Universitat
de les Illes Balears

DOCTORAL THESIS

2024

OCUPAT: Occupational Project in Patients with Hereditary Transthyretin Amyloidosis

Aina Isabel Gayà Barroso

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Doctoral Programme in Psychology

OCUPAT: Occupational Project in Patients with Hereditary Transthyretin Amyloidosis

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I DECLARE:

That the thesis titles **OCUPAT: Occupational Project in Patients with Hereditary Transthyretin Amyloidosis**, presented by Aina Isabel Gayà Barroso to obtain a doctoral degree, has been completed under my supervision and meets the requirements to opt for an International Doctorate.

For all intents and purposes, I hereby sign this document.

Signature:

Palma de Mallorca, 30.10.2023

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I DECLARE:

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Caminante, son tus huellas
el camino y nada más;
Caminante, no hay camino,
se hace camino al andar.
Al andar se hace el camino,
y al volver la vista atrás
se ve la senda que nunca
se ha de volver a pisar.
Caminante no hay camino
sino estelas en la mar.

Antonio Machado

Acknowledgements

What we do is part of who we are

I would like to express my sincere gratitude to my supervisors, Inés Losada and Eugenia Cisneros, for their unwavering support and guidance throughout the whole of my professional project and its objectives. It was their competence and their encouragement that made it possible for me to complete this adventure. I can't thank you enough for all you've done, and in particular for believing in my ideas.

Once again, my thanks go to you, Mari Euge. Thank you for advising me and, above all, for being my sister. Without you, I would not have been able to do this PhD. and so many other things.

I am deeply grateful to Milena Paneque for her constructive feedback, for suggesting and for allowing me to come to CGPP to do my research. I am also grateful for all she has done to help and support me. However, Milena, I wanted to thank you for having allowed me to feel at home in another country, thank you for that gift: Porto.

I would also like to thank my colleagues at the Hospital Universitario Son Llàtzer for all their efforts and collaboration during my Ph.D. In particular, I would like to thank Juan González and Adrián Rodríguez for the help they have given me in the writing of my thesis and for the support they have given me during this trip.

Thank you so much, Sara Pérez. Your helpful insights and guidance have been fundamental since I graduated. I am here today because of many of your advices.

To my mother and father, I am eternally grateful. I realise now: You are and always will be my reason. Nobody will ever trust me as much as you do. I love you so much.

Thank you for allowing me to see the world in a different way and teaching me to be.

I'd also like to thank my friends for all they have done to support me through these three exciting years. I would not have been able to complete this amazing life experience without their encouragement, patience and motivation.

Cova, Clara, Paula, Aina, Sonia, La ra, Ali, Francisco, Tiago, Ana, Raquel, Daniela, Inés, Catarina, Carmen, Jorge and “Al ser de luz” too, whatever their countries or tongues: GRACIAS. After three years, I realise that good friends are the ones who are there in good times and especially in all the times.

Obrigada Sempre!

In particular, I would like to thank my "new family" in Portugal for all the love and understanding that they have shared with me. Ever since my arrival in Porto, I have had the feeling of being at home. To be honest, I think that this is without a doubt one of the greatest gifts that this project has given me.

Leyre, more than a friend, a kind of sister since the biology joined us, only one thing: "Lo hacemos y ya veremos, pero hazlo".

Finally, I would like to thank everyone who participated in my study for taking the time to share their everyday lives with me. Without you this work would not have been possible. ABEA: Aina, Tina, Katy, Cristina, Joel, Frederich, Xisca, Magdalena, Mercedes, Bel, Antonia, Gloria, Paula, Maria del Mar, Miquel, Segismundo.

And for you, Catilena, aqui tens una companya. You have been the driving force behind the idea and behind me.

Finally, I would like to dedicate the last paragraph to myself.

Remember what matters. You have laughed, you have cried, you have changed, you have been wrong and you have tried again, you have come to a new place alone and you have started, you have wanted to explain what occupational therapy means to you and you have done it and you are still doing it.

All I can add is: Go on and fight, just as your mother taught you to do every day when she left you at the gate of the school.

It is not a matter of proving anything to anybody, it is a matter of being on your way.

To walk. To live.

Thank you. Thank you for this wonderful opportunity.

Aina Gayà Barroso.

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Presentation

Summary

Variant transthyretin amyloidosisis a rare genetic disease with a variable prognosis and complex to understand. It was first described in 1952 by the Portuguese neurologist Corino Andrade as a peculiar form of peripheral neuropathy; familiar atypical generalized amyloidosis with special involvement of the peripheral nerves, reason why it is also known as Andrade's disease (Andrade, 1952).

In Europe, the incidence of variant transthyretin amyloidosis varies widely. In Portugal, Sweden, Majorca and Cyprus, variant transthyretin amyloidosis with Polineuropathy is endemic and one particular mutation predominates. Cases are more sporadic in the rest of Europe. More than 140 different mutations in the TTR gene have been described. However, the Val30Met variant transthyretin amyloidosis is the most common (Connors et al., 2003).

The first symptoms of the disease usually appear in the third decade of life. Patients usually experience severe physical limitations due to this genetic mutation, but psychological, social and occupational effects have also been described (Luigetti et al., 2020). Existing research suggests that being diagnosed affects daily life (Buades-Reinés et al., 2016).

However, in addition to the clinical approach, no type of intervention has been described that focuses on the maintenance, improvement and adaptation of the daily life of patients and their carers after the diagnosis, a competence that is partly within the discipline of occupational therapy.

This study, in collaboration with the Centre for Predictive and Preventive Genetics (Porto), is a research project carried out by the Multidisciplinary Unit for Hereditary Transthyretin Amyloidosis at the Son Llàtzer University Hospital. It is based on the analysis of the presence, knowledge, access and interventions related to occupational therapy, with the diagnosis of variant transthyretin amyloidosis.

Specifically, this project aims to analyse the effectiveness of an occupational intervention in patients with variant transthyretin amyloidosis. A sample of individuals was assessed at baseline and at the end of the intervention to compare scores on scales and interviews. It was expected that

the daily living approach would have multiple benefits, based on the literature on occupational interventions in patients with different diagnoses. The aim was to have a concrete knowledge of what the benefits would be, in which parameters of the daily life of these patients these benefits would be real and what their magnitude would be.

On the other hand, given the importance of psychosocial support in this typically Portuguese, genetic, degenerative and incurable disease, we also considered it necessary to carry out an exploratory study to better understand the situation of occupational therapy in Portugal, with an Iberian perspective. To answer this question, we designed the study in which interviews were conducted with Portuguese and Spanish occupational therapists. We believe that this study has contributed to a more accurate description of the vision of this discipline, which is still developing.

Our aim is to illustrate the impact of this disease on the occupational dimension and on daily life, to open up avenues for future research, to stimulate reflection on possible lines of intervention and to highlight the importance of a differentiated multidisciplinary team, complemented by professionals in the psychosocial field, such as occupational therapists.

This research is presented in a compendium of five scientific articles. The psychosocial burden of patients and carers is analysed in the first part. This is followed by a study of patients' access to occupational therapy, identification of occupational needs and implementation of an occupational programme in patients with a diagnosis of variant transthyretin amyloidosis. To complete and complement the thesis, an Iberian study on occupational therapy was carried out.

Finally, we present the discussion of the results already obtained in the articles, the limitations that we could see inherent in this project, general conclusions and a proposal for the future. We also present the documents used throughout the study, which are referred to in the annexes.

Resum

L'amiloïdisi variant de la transtirretina és una malaltia genètica i rara amb un pronòstic variable i complex d'entendre. Va ser descrita per primera vegada el 1952 pel neuròleg portuguès Corino Andrade, i és precisament la causa per la qual també és coneguda com a Malaltia d'Andrade (Andrade, 1952).

La incidència d'aquesta malaltia varia àmpliament a Europa. A Portugal, Suècia, Mallorca i Xipre, l'amiloïdisi variant de la transntiretina amb polineuropatia és considerada una malaltia endèmica, amb una prevalença més gran d'una mutació particular. Per contra, a la resta d'Europa, els casos són més esporàdics. S'han descrit més de 140 mutacions diferents en el gen TTR, però, la variant Val30Met de l'amiloïdisi variant de la transtiretina és la més reconeguda (Connors et al., 2003).

Els primers símptomes de la malaltia solen aparèixer en la tercera dècada de vida, en la qual els pacients solen experimentar limitacions físiques greus imposades per aquesta mutació genética. Per una altra banda, s'han descrit també efectes psicològics, socials i ocupacionals (Luigetti et al., 2020). Les investigacions fins ara suggereixen que el diagnòstic té un impacte directe a la vida diària (Buades-Reinés et al., 2016).

No obstant això, a més de l'abordatge clínic, no s'ha descrit cap tipus d'intervenció que se centri en el manteniment, millora i adaptació de la vida diària dels pacients i els seus cuidadors després del diagnòstic, una competència que està en part, dins de la disciplina de la teràpia ocupacional.

Aquest estudi és un projecte de recerca dut a terme per la Unitat Multidisciplinària d'Amiloïdisi Hereditària per Transtiretina de l'Hospital Universitari Son Llàtzer, en col·laboració amb el Centre de Genètica Predictiva i Preventiva (Oporto) i es basa en l'anàlisi de la presència, coneixement, accés i intervencions relacionades amb la teràpia ocupacional i el diagnòstic d'aquesta malaltia.

En l'àmbit específic, aquest projecte té com a objectiu analitzar l' efectivitat d' una intervenció ocupacional en aquests pacients, avaluant una mostra d' individus a l' inici i al final de la intervenció per comparar les puntuacions en escales i entrevistes. D' acord amb la literatura sobre intervencions ocupacionals en pacients amb diferents diagnòstics, queda constància que l' enfocament de la vida diària té múltiples beneficisL'objectiu d'aquest estudi és tenir un coneixement concret de quins serien els beneficis, en quins paràmetres de la vida diària aquests pacients es beneficien i quina seria la seva magnitud.

D'altra banda, també considerem necessari realitzar un estudi exploratori per comprendre millor la situació de la teràpia ocupacional a Portugal, amb una perspectiva ibèrica, atesa la importància

del suport psicosocial en aquesta malaltia típicament portuguesa, genètica, degenerativa i incurable. Per respondre a aquesta pregunta, dissenyem l'estudi en el qual es van dur a terme entrevistes amb terapeutes ocupacionals portuguesos i espanyols. Es considera que aquest estudi ha contribuït a una descripció més precisa d' aquesta disciplina, encara en procés de desenvolupament.

La nostra perspectiva pretén il·lustrar l'impacte d'aquesta malaltia en la dimensió ocupacional i en la vida quotidiana, obrir vies per a futures investigacions, fomentar la reflexió sobre possibles línies d'intervenció i destacar la importància de comptar amb un equip multidisciplinari diferenciat, complementat per professionals de l'àmbit psicosocial, com els terapeutes ocupacionals.

Un compendi de cinc articles científics presenta aquesta investigació. La primera part analitza la càrrega psicosocial dels pacients i cuidadors. A això li segueix un estudi sobre l'accessibilitat i el coneixement dels pacients sobre la teràpia ocupacional, la identificació de les necessitats ocupacionals i la implementació d'un programa ocupacional en pacients diagnosticats amb amiloïdosi variant de la transtiretina. Per completar la tesi i com a complement, es va realitzar un estudi ibèric de la situació de la teràpia ocupacional.

Finalment, presentem la discussió dels resultats ja obtinguts en els articles, les limitacions que puguem veure inherents a aquest projecte, conclusions generals i una proposta per al futur, així com els documents utilitzats al llarg de l' estudi, esmentats en els annexos.

Resumen

La amiloidosis variante de la transtirretina es una enfermedad genética y rara con un pronóstico variable y complejo de entender. Fue descrita por primera vez en 1952 por el neurólogo portugués Corino Andrade, y es precisamente la causa por la que también es conocida como Enfermedad de Andrade (Andrade, 1952)

La incidencia de esta enfermedad varía ampliamente en Europa. En Portugal, Suecia, Mallorca y Chipre, la amiloidosis variante de la trasntirretina con polineuropatía es considerada una enfermedad endémica, con una prevalencia mayor de una mutación particular, Por el contrario, en el resto de Europa, los casos son más esporádicos. Se han descrito más de 140 mutaciones diferentes en el gen TTR, sin embargo, la variante Val30Met de la amiloidosis variante de la transtirretina es la más común (Connors et al., 2003).

Los primeros síntomas de la enfermedad suelen aparecer en la tercera década de vida, en la que los pacientes suelen experimentar limitaciones físicas severas impuestas por esta mutación genética, pero también se han descrito efectos psicológicos, sociales y ocupacionales (Luigetti et al., 2020). Las investigaciones hasta la fecha sugieren que el diagnóstico tiene un impacto en la vida diaria (Buades-Reinés et al., 2016).

Sin embargo, además del abordaje clínico, no se ha descrito ningún tipo de intervención que se centre en el mantenimiento, mejora y adaptación de la vida diaria de los pacientes y sus cuidadores tras el diagnóstico, una competencia que está en parte, dentro de la disciplina de la terapia ocupacional.

Este estudio es un proyecto de investigación llevado a cabo por la Unidad Multidisciplinar de Amiloidosis Hereditaria por Transtiretina del Hospital Universitario Son Llàtzer, en colaboración con el Centro de Genética Predictiva y Preventiva (Oporto) y se basa en el análisis de la presencia, conocimiento, acceso e intervenciones relacionadas con la terapia ocupacional y el diagnóstico de dicha enfermedad.

A nivel específico, este proyecto tiene como objetivo analizar la efectividad de una intervención ocupacional en estos pacientes, evaluando una muestra de individuos al inicio y al final de la intervención para comparar las puntuaciones en escalas y entrevistas. De acuerdo a la literatura sobre intervenciones ocupacionales en pacientes con diferentes diagnósticos, queda constancia de que el enfoque de la vida diaria tiene múltiples beneficios. El objetivo de este estudio es tener un conocimiento concreto de cuáles serían los beneficios, en qué parámetros de la vida diaria estos pacientes se benefician y cuál sería su magnitud.

Por otro lado, también consideramos necesario realizar un estudio exploratorio para comprender mejor la situación de la terapia ocupacional en Portugal, con una perspectiva ibérica, dada la importancia del apoyo psicosocial en esta enfermedad típicamente portuguesa, genética, degenerativa e incurable. Para responder a esta pregunta, diseñamos el estudio en el cual se realizaron entrevistas con terapeutas ocupacionales portugueses y españoles. Se considera que este estudio ha contribuido a una descripción más precisa de esta disciplina, todavía en proceso de desarrollo.

Nuestra perspectiva pretende ilustrar el impacto de esta enfermedad en la dimensión ocupacional y en la vida cotidiana, abrir vías para futuras investigaciones, fomentar la reflexión sobre posibles líneas de intervención y destacar la importancia de contar con un equipo multidisciplinar diferenciado, complementado por profesionales del ámbito psicosocial, como los terapeutas ocupacionales.

Un compendio de cinco artículos científicos presenta esta investigación. La primera parte analiza la carga psicosocial de los pacientes y cuidadores. A esto le sigue un estudio sobre la accesibilidad y el conocimiento de los pacientes sobre la terapia ocupacional, la identificación de las necesidades ocupacionales y la implementación de un programa ocupacional en pacientes diagnosticados con amiloidosis variante de la transtirretina. Para completar la tesis y como complemento, se realizó un estudio ibérico de la situación de la terapia ocupacional.

Finalmente, presentamos la discusión de los resultados ya obtenidos en los artículos, las limitaciones que pudimos ver inherentes a este proyecto, conclusiones generales y una propuesta para el futuro, así como los documentos utilizados a lo largo del estudio, mencionados en los anexos.

Index of Abbreviations

AA: Amyloid serum A protein

AADL: Advanced activities of daily living

A-AL: immunoglobulin light chain amyloidosis

ABEA: Asociación Balear de la Enfermedad de Andrade

ADL: Activities of Daily Living

AMILO: Asociación Española de Amiloidosis

AO: Age of Onset

AOTA: American Occupational Therapy Association

ATTRv: Variant Transthyretin Amyloidosis

ATTRv -PN: variant transthyretin amyloidosis with polyneuropathy

BADL: Basic Activities of daily living

BI: Barthel índex

CMT: Charcot-Marie-Tooth disease

DNA: Deoxyribonucleic acidFAP: Familial Amyloid Polyneuropathy

ENTO: National School of Occupational Therapy

FEDER: Federación Española de Enfermedades Raras

IADL: Instrumental activities of daily living

LT: Liver transplantation

MDT: Multidisciplinary team

MOHO: Modelo de ocupación humana

Norfolk QoL-DN: Norfolk Quality of Life-Diabetic Neuropathy

OT: Occupational Therapy

PGD: preimplantation genetic diagnosis

PND: polyneuropathy disability

QoL: Quality of Life

RAs: Reconstruction Aides

RDs: Rare diseases

SF-36: The Short Form (36) Health Survey

TTR gene: transthyretin gene

WEMWBS: The Warwick-Edinburgh Mental Wellbeing Scales

WFOT: World Federation of Occupational Therapists

WHO: World Health Organization

Table Index

Nacho Varela

Chapter I
Daily life

1 Rare diseases

1.1 General aspects

The disease may be rare, but the people who suffer from it are not
Anonymous

To set the scene for this work, a brief general review of the basic concepts related to the diagnosis of rare diseases is considered necessary.

What is a rare disease?

Rare diseases (RDs) are numerous and heterogeneous in their nature and in their geographical distribution. Few are preventable or curable. Most are chronic and many cause early death. In spite of their heterogeneity, rare diseases share common characteristics that are related to their rarity. This calls for a comprehensive response from the public health sector (Nguengang Wakap, et al., 2020).

At present, between 3.5 % and 5.9 % of the world's population is affected by rare diseases.

When is a disease considered to be rare?

Although there is no universal definition of RD, it has been suggested that, in the current political and legal context, RD is closely linked to defining RD based on point prevalence (Richter, et al., 2015). A prevalence threshold is explicitly or implicitly used in existing definitions.

In the European Union (Eurodis, n.d.), a disease is considered rare if it affects fewer than 1 in 2,000 citizens.

How many rare diseases have been the subject of research?

To date, 7,000 rare diseases have been identified in Spain. New diseases are regularly described in the medical literature. The number of rare diseases also depends on the degree of specificity in the categorization of the different pathologies (FEDER, n.d.).

The consequences

Medical expertise is scarce because of the low prevalence of rare diseases. This means that little is known, care is inadequate, and research is limited. Patients with rare diseases are the orphans of healthcare systems. Despite their large numbers, they are often denied diagnosis, treatment and the benefits of research (Nguengang Wakap, et al., 2020).

Rare diseases can be hidden behind relatively common symptoms. The result is misdiagnosis and delays in treatment. Rare diseases are typically disabling and debilitating. A rare disease patient's quality of life (QoL) is affected by lack or loss of autonomy due to the chronic, progressive, degenerative and often life-threatening aspects of the disease (O'Connor, et al, 2023).

Diagnosis

On the basis of Spanish data, it is estimated that a person with a rare disease waits an average of 4 years for a final diagnosis, with 20% of cases taking 10 years or more to receive an accurate diagnosis.

The time to diagnosis for the patient, according to this information and the data provided by FEDER, is

- Not having been seen by a multidisciplinary team or not having received medical treatment (29.37%).
- The wrong type of treatment is offered (17.9% of individuals).
- A clinical and psychosocial deterioration has been observed (31.26% of the individuals).

Treatments

Due to the rarity of rare diseases, there may be no specific treatment for the disease, treatment options may not be widely available or there may be a need for special attention.

Multidisciplinary approaches often benefit patients. This involves health professionals with experience in their specific condition, which is another complex reality as, due to this characteristic, there are not many specialised professionals with the necessary skills to carry out the right approaches (Meyer et al., 2022).

The main treatment options currently available to people with a rare disease diagnosis are (Meyer et al., 2022):

- **Supportive care**: Rare diseases are characterised by the lack of specific curative treatments for rare diseases. Supportive care is essential in this situation.
- Supportive care is defined as a service that provides information, empowerment and strategies, management of symptoms and their consequences, provision of physical, psychological and OT care, and management of any complications that may arise.
- **Gene therapy:** For certain rare genetic diseases, advances in genetic research have led to the development of gene therapy. In this approach, the underlying cause of the disease is corrected by modifying or replacing damaged genes.
- **Stem cell transplants:** Stem cell transplantation is used to treat certain rare diseases, particularly those affecting the blood or immune system, including bone marrow and haematopoietic stem cell transplantation. This option is used in order to replace cells that are not working properly with healthy cells.
- **Pharmacological treatments:** For some rare diseases, pharmacological treatments and medications may be used for symptom management or disease control.
- **Surgical treatments:** For some rare diseases, surgery may be necessary to correct structural deformities or for the removal of tumours or damaged tissue.
- Surgery varies depending on the condition and the symptoms experienced.
- **Orphan Drugs:** Orphan drugs are medications specifically designed to treat rare diseases.
- **Lifestyle:** Daily lifestyle management to manage symptoms and improve overall health may be recommended for all rare diseases. Multidisciplinary interventions may include psychology, social work, occupational therapy, nutrition, physiotherapy or podiatry.

Social Exclusion

Regarding rare diagnosis and social context, 43% of Spanish patients diagnosed with these characteristics felt discriminated against in at least some situation because of their disease or related symptoms. The most important contexts of discrimination reported by patients are: social relations (56.91%), health resources (43.21%), education (32.46%) and daily life (62.56%) (FEDER, n.d.).

There is therefore a clear need for intervention in order to help people with a diagnosis to manage their daily lives.

Daily life dimension

As reported by Feder, patients:

- 33.74% reported having less free time.
- 10% report that they have had a loss of job opportunities as a result of the disease.
- 12.03% have had a reduction in their working hours due to incompatibility or lack of adaptation as a result of symptoms.
- 10.45% have had a loss of educational opportunities as a result of the disease.

These data are proving to be very important in developing this project.

Patient advocacy groups

For individuals, carers and families affected by a rare disease diagnosis, patient advocacy groups are an important source of support. They focus specifically on raising awareness, empowerment, social justice, providing information, services and resources, advocating for better healthcare systems and promoting research within this community.

We would like to highlight some of the key patient advocacy organisations around the world. These include:

EURORDIS - Rare Diseases Europe:

EURORDIS: is the leading alliance of rare disease patient organisations in Europe.

It focuses on defending the rights and responsibilities of people with rare diseases. It also promotes research initiatives in the field.

Global genes:

Global Genes: is a global rare disease advocacy organisation. It is based in the United States. They work to connect and empower rare disease communities, provide education and awareness, and promote new initiatives, following the lead of other organisations.

National Organization for Rare Diseases (NORD):

This organisation is the leading independent advocacy and research organisation representing all patients and families affected by rare diseases in the United States.

Rare Diseases International (RDI):

It is defined as a global alliance of patient organisations in the rare disease community. They promote policies and initiatives that benefit patients worldwide, in collaboration with other interested international organisations.

Rare diseases South Africa (RDSA):

To improve the QoL for people with rare diseases in the region, this leading organisation in South Africa provides support, resources, advocacy, education, start-up and awareness campaigns.

Canadian Organization for Rare Diseases (CORD):

It is the Canadian organisation whose mission is to improve the QoL of Canadian rare disease patients. Their work is in line with the work of the other organisations in this list.

UK Rare Disease Forum:

The Forum brings together several rare disease organisations in the UK to co-ordinate efforts to promote better medical and psychosocial care for rare disease patients.

National Registry of Patients with Rare Diseases and Biobank (Spain):

This Spanish organisation focuses on rare diseases. It has a patient registry and a biobank to promote research and the best QoL for patients.

These are just a few examples. There are many other rare disease associations, groups and federations around the world, each dedicated to education, awareness raising, social justice, support for individuals and families living with a rare disease, and the advancement of research and treatment options, not only at the medical level, but also at the psychosocial level.

What are the prospects for progress in psychosocial interventions for rare diseases?

In all rare diseases, to improve the well-being of people affected by these diseases, researchers from different disciplines are increasingly working in networks to share their research and make progress more efficiently.

Not only medicine, but also psychology, genetic counselling, occupational therapy, physiotherapy and social work are finding new hope in the prospects offered by new research. This is certainly the case with this ground-breaking project.

2 Variant Transthyretin amyloidosis

2.1 Types of Amyloidosis

Systemic amyloidoses are a group of complex diseases in which misfolded proteins accumulate in the tissues and cause progressive damage to the organs. Immunoglobulin light chain amyloidosis (AL) is the most common type. It is caused by clonal plasma cells that produce misfolded light chains (Merlini et al. (Merlini et al, 2011).

More than 140 different mutations in the TTR gene have been identified and about 30 types of amyloidosis have been described. More are expected to be identified as research continues. The most common types are AL amyloidosis, AA amyloidosis, ATTRv amyloidosis, TTR wild-type and localised amyloidosis (Mayo Clinic, n.d.).

One of the proteins that can cause amyloidosis is the protein transthyretin (TTR), whose function is to transport thyroxine and retinol. The disease is caused by the presence of a mutant protein identified on chromosome 18, which leads to the deposition of extracellular insoluble amyloid fibrils. Transthyretin (TTR) or prealbumin is a 127 amino acid protein. It is synthesised in the liver. (Munar-Qués, et al., 2005).

Transthyretin amyloidosis (ATTR) is the name given to all amyloidoses caused by transthyretin accumulation. Within ATTR, a clear distinction is made between two types (Mayo Clinic, n.d.):

ATTRv: The deposition of the TTR protein is due to a mutation in the deoxyribonucleic acid (DNA) so that the protein is not made properly.

ATTR wild-type: this is the same protein (not altered in its production) that is pathologically deposited in tissues and organs. That is why it is called "wild type".

Historic view of Variant Transthyretin amyloidosis

ATTRv was first described in northern Portugal, in the region of Póvoa do Varzim, by Corino de Andrade in 1952. The disease was defined as an autosomal dominant neurodegenerative disease characterised by a progressive progression of neurologic symptoms, including dementia. (Coutinho, 1989).

Familial amyloid polyneuropathy (FAP), paramyeloid disease, hereditary transthyretin amyloidosis (hATTR) or "Doença dos Pezinhos" or even Andrade's disease are equivalent terms for this neurological form of systemic disease affecting the peripheral nervous system (Santos, et al., 2021).

The mutation in the transthyretin gene leads to a mutated protein. This is deposited in the connective tissue as amyloid plaques. In 1939, the neurologist Corino de Andrade observed the first patient with ATTRv and claimed that there were many relatives with similar symptoms in his home town of Póvoa de Varzim (Portugal) and in his family (Coutinho et al., 1980). This person, who was part of a family of fishermen, began to feel a tingling and burning sensation in his lower limbs; he had been burnt, cut by fishing nets without realising it, had diarrhoea and significant weight loss. (Lopes et al., 2017).

Corino de Andrade first reported Portuguese familial amyloidosis in the Brain Journal twelve years later, in 1952. In 1962, as a result of his research, he founded the Centre for the Study of Neuropathology. This is now the Centre for Paramyeloid Studies (CEP). The disease quickly spread to other areas through migratory flows and fishing trips along the Portuguese coast: Póvoa de Varzim, Vila do Conde, Barcelos, Braga, Porto, Vila Nova de Gaia, Unhais da Serra, Figueira da Foz and Lisbon, among others. (Saraiva, 2015).

Recently, ATTRv has been described as a multisystem disease. This means that more than one system or organ is affected. In this sense, the disease of ATTRv is not only a polyneuropathy, as it has been described so far. Other symptoms would have been ignored by Dr Corino Andrade. That is why his application was not completed. For this reason, other names that have been used in the past, such as "familial amyloid polyneuropathy", have been abandoned. They are imprecise. As research has progressed, it has become clear that all of the amyloid diseases are caused by proteins that build up in the tissues and organs. (ABEA, n.d.).

To date, ATTRv has been described as a systemic disease that results from the extracellular deposition of insoluble transthyretin (TTR) amyloid fibrils in a variety of organs and tissues (Ando et al., 2013).

Diagnostic process:

ATTRv can be difficult to diagnose correctly. Some people are initially misdiagnosed. It is essential for healthcare professionals to be familiar with the symptoms, diagnostic process and treatments, as different treatments are available that can alter the natural history of ATTRv (Ando et al, 2022).

To diagnose ATTRv, the following process is used:

Genetic test:

This is a blood test. Its main objective is to identify the genetic mutation that causes ATTRv. In non-endemic regions, full sequencing of the transthyretin gene is recommended.

Symptoms:

The presence of disease symptoms, such as those described in the Symptoms section, is valuable information. Usually, the disease first appears with mild symptoms. These include dizziness, tingling in the feet, diarrhoea and involuntary weight loss.

Specific tests:

These are a variety of diagnostic tests that look for the presence of amyloid in tissues and organs and/or detect changes in nerve function.

The most common include

- Electromyogram
- Biopsies
- Echocardiogram
- Urine 24 hours
- Electrocardiogram
- Holter ECG

The family background:

ATTRv is an inherited disease. This means that there is a 50% chance of the disease being passed from parent to child during each pregnancy. As an inherited disease, ATTRv is inherited in an autosomal dominant pattern and has variable penetrance. This means that not all people who carry the mutation will develop the disease.

Therefore, being a carrier does not mean having symptoms. It means having the potential to develop the disease and pass it on to those born.

Genetic testing should be carried out on first-degree and ascending relatives of a confirmed ATTRv case in a family. Depending on the results, offspring will also be tested. The family tree is

very useful for the identification of all relatives in the same family who may be at risk of being carriers. It must be as detailed as possible, showing the index case and blood relatives: ascendants, descendants and collaterals with all nuclear families forming the extended family. (Saraiva, 2015).

It is also important to try to include as many generations as possible.

Signs and symptoms:

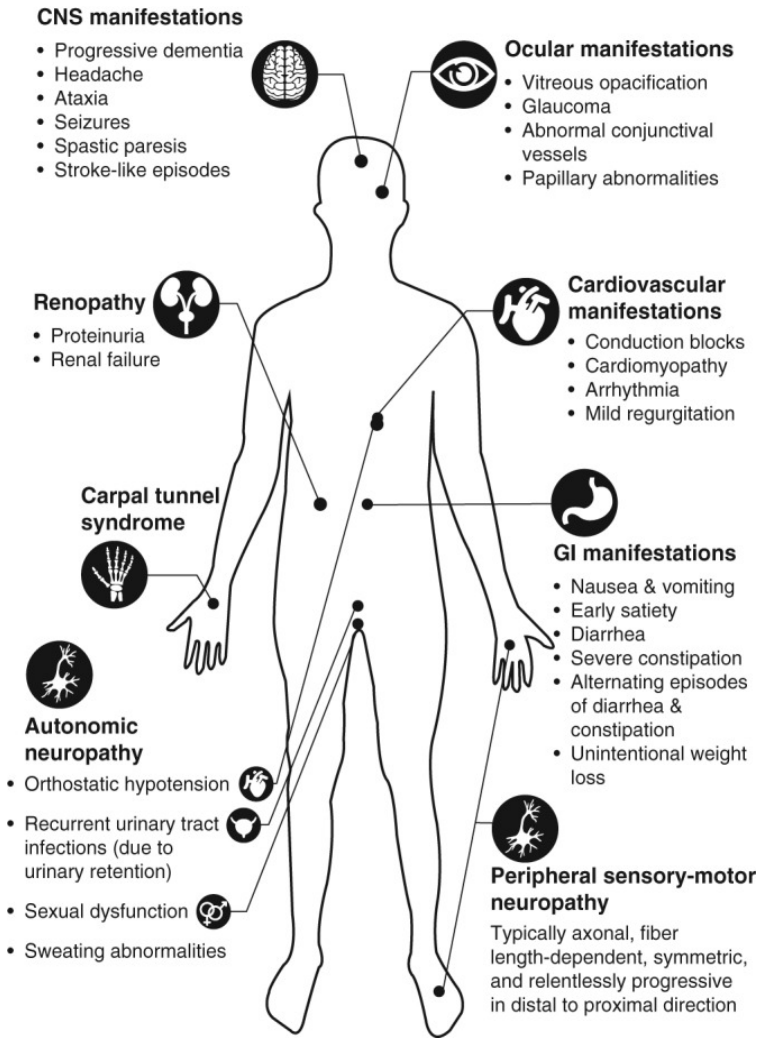

Figure 1. Clinical features in association with TTR-FAP. CNS, central nervous system; GI, gastrointestinal tract. Excerpted from Conceição, et al, 2016.

There are a number of warning signs that can be used to aid in the diagnostic process. The presence of a symmetrical and progressive sensorimotor neuropathy or the presence of any of the following symptoms may indicate the possible presence of this disease.

Some of the symptoms are:

- Early autonomic dysfunction
- Cardiac dysfunction
- Dysfunction of the gastrointestinal tract
- Weight loss
- Carpal tunnel syndrome
- Renal disorders
- Vitreous opacities

The symptoms of ATTRv depend on the type of variant and the organ/tissue where the amyloid deposition occurs.

In the case of the Val30Met mutation, the most common symptoms are:

Table 1. Main symptoms described in ATTRv

Peripheral Nervous System	**Tingling** **Neuropathic Pain** **Weakness and loss of sensation in the feet that can go up to the hands**
Central Nervous System	Epileptic seizures Temporary loss of mobility in an arm or leg Headache Cognitive impairment Ataxia
Autonomous Nervous System	Loss of consciousness Diarrhoea alternated with periods of constipation Sexual impotence Sweating alterations
Cardiovascular System	Heart rhythm disturbances Shortness of breath when walking Fluid retention Chest pain
Vision	Vision loss Glaucoma Pupil changes
Kidneys	Recurrent urinary tract Infections Kidney failure Proteinuria

Note: Luigetti et al., 2020.

Different stages of the disease have been described, in which the disease manifests itself with specific characteristics. The following is a description of the main stages of the disease, along with a description of the symptoms.

If the disease is left untreated, ATTRv will usually lead to death from disease-related causes within 10 to 15 years. To date, the prognosis for ATTRv patients has improved significantly. This is due to the availability of several treatments that can slow the progression of the disease. However, a cure for the disease has not yet been achieved.

The symptoms of the disease are classified by two models, Coutinho and Yamamoto, as follows:

Table 2. Coutinho and Yamamoto disease stages

Coutinho		Duration of stage	Yamamoto	
Stage 1	The disease is limited to the lower limbs Walking without any help. Slight weakness of the extensors of the big toes.	5.6 ± 2.8 years	PND I	Sensory disturbances in extremities Preserved walking capacity
Stage 2	Motor signs progress in lower limbs with steppage and distal amyotrophies, the muscles of the hands begin to be wasted and weak. The patient is by then obviously handicapped but can still move around, although needing help.	4.8 ± 3.6 years	PND II PND IIIa PND IIIb	Difficulties walking but without the need for a walking stick One stick or one crutch required for walking. Two sticks or two crutches required for walking
Stage 3	The patient is bedridden or confined to a wheelchair, generalized weakness and areflexia.	2.3 ± 3.1 years	PND IV	Patient confined to a wheelchair or a bed

Note: Adapted from Adams, 2013.

Available treatments

Several disease-modifying treatments have been shown to be effective and safe in reducing ATTRv over the past decade. Liver transplantation (LT) was the first treatment to be offered. It works by eliminating amyloidogenic mutated TTR produced by the liver. However, limitations of this option have been described, despite its efficacy: As a surgical procedure, it is of course associated with an implicit risk and with a limited number of transplants that are adaptable. In general, it leads to further disease progression due to the deposition of amyloid fibrils in the eye and choroid plexus. For these reasons, there is a need for research and development of new treatments that take into account the pathogenic TTR pathway, stabilising the mutant TTR protein, preventing its dissociation and the formation and deposition of amyloid fibrils (Conceição, et al., 2016).

The current treatments can be divided into three groups:

Table 3. Major clinical treatments and psychosocial interventions in use to date

Symptomatic treatment	Enteropathogenic treatment	Clinical trials
Its purpose is to calm/relieve/diminish the symptoms of the disease.	Its purpose is to stop/slow down the progression ATTRv and the damage caused by amyloid deposits.	Removal of amyloid deposits CRISPR gene editing
Pharmacological or non-pharmacological symptomatic treatments. These can be physical therapy, occupational therapy, diet and nutrition, podiatry, psychology, etc.	Available treatments are: TTR synthesis inhibitors TTR stabilizers liver transplant	

Note: Ando and others, 2022.

1 LT

Liver transplantation was the first therapeutic strategy for this disease. It has been performed on hundreds of ATTRv patients worldwide and until 2011 was the only treatment that could stabilise disease progression.

To prevent the formation of new amyloid deposits, the liver of the person producing the mutated transthyretin TTR is replaced with a liver from a donor that produces TTR without the mutation. Transplantation early in the course of the disease is most successful. This means that it is done before the heart and nerves have become too damaged.

Unfortunately, a cure has not yet been found. Even after transplantation, there is still progression of the disease, particularly in the eyes, heart and central nervous system. It appears that amyloid deposits formed prior to liver transplantation act as a template for TTR to deposit as amyloid. Therefore, the normal TTR protein that is produced by the new liver will accumulate on top of the pre-existing amyloid deposits that contain the abnormal TTR.

Today, LT is only available to those patients who are aware of the other options available and wish to undergo it. Similarly, the decision to start pharmacological treatment does not affect access to transplantation.

2 Transthyretin (TTR) synthesis inhibitors:

Inhibitors are used to try and stop the TTR protein from getting made. Two options are available:

Patisiran

This is a drug that tries to stop the production of the mutant transthyretin protein. It prevents the build-up of deposits. It is given intravenously. It is given every 21 days, usually in hospital. Additional drugs, such as corticosteroids, may be needed to prevent possible side effects.

It has been shown to be effective in slowing the progression of ATTRv at the nerve and heart levels, with few side effects.

It is indicated for polyneuropathy and in stages I and II of the disease.

Inotersen

This treatment is used to try to prevent the production of the mutated transthyretin protein. It is given by subcutaneous injection every 7 days. It is taken at home.

It has also been shown to be effective in the slowing of disease progression at the level of the nerves. One of the main side effects reported is low blood platelets. This is rare. Fortnightly blood tests are therefore necessary for monitoring of platelet levels. It is indicated for polyneuropathy and in stages I and II of the disease.

3 TTR stabilisers

Transthyretin TTR stabilisers aim to prevent the TTR protein from forming amyloid deposits.

Tafamidis

This is a pill taken once a day. It tries to prevent the mutant protein, transthyretin, from being deposited in different tissues and organs in the body. Trials have shown that 60% of people treated with Tafamidis respond. The rest have only a partial response.

With a long history of use, it is a safe drug. There have been few reports of side effects. It is indicated to treat polyneuropathy, but only in the early stages of the disease and when hospitalised.

2.4 Removing amyloid plaques:

There are currently no drugs approved to remove amyloid plaques. There are currently no drugs on the market for amyloid removal, but one is currently in clinical trials (PRX004).

2.5 CRISPR gene editing:

There are currently no approved gene editing drugs for the treatment of ATTRv. They are currently being tested in clinical trials (CRISPR-Cas9).

3 Population general description

To describe general characteristics, the population with a diagnosis of ATTRv was studied.

In relation to the age of onset (AO), as reported in Buades-Reinés et al. (2016). The results from Mallorca show that the AO is 45.7 years of age. This is similar to the results obtained in some other Mediterranean areas, such as Cyprus, where the AO is 46 years.

Earlier AO was found in the main endemic areas, Portugal 33.5, Japan 35.6 and Brazil 32. A later age of onset of 56.7 years has been reported from the north of Sweden.

Considering that the causal mutation is probably the same in Portugal and Majorca, and that patients who have no relatives with the disease in other areas (sporadic cases) have a later AO, the difference in age of onset between Portuguese and Majorcan cases may be due to environmental or epigenetic factors.

The results confirm that the AO is similar in men and women. This was shown by Buades-Reinés et al. (2016). The results show a 51/49% ratio in Mallorca. Similar results have been reported in several other regions, such as Brazil (45% to 55%) and Cyprus (44% to 56%). A later AO in women has been described in Portugal and Brazil. No gender differences were found in Sweden, Cyprus or the previous series from Mallorca.

Depending on the diagnosis and the relationship with family history, Buades-Reinés et al (2016) explain that a high proportion of patients had a positive family history, mostly on the father's side. Family history was more common in asymptomatic carriers, as expected. An earlier diagnosis was also observed in these individuals, and many symptomatic patients received a liver transplant. The mean time to transplantation was just over 2.5 years from diagnosis.

Regarding symptoms, Buades-Reinés et al (2016) describe that in previous observational studies of endemic foci associated with early-onset AO (<50 years), such as in Japan and Portugal, ATTRv is clinically characterised by sensorimotor dysfunction and severe dysautonomic symptoms. In contrast, progressive sensorimotor symptoms (especially in the upper limbs) with only moderate dysautonomia are clinically characteristic of patients with late-onset AO (≥50 years of age) without family history and from non-endemic foci. However, it is not yet known why there are differences between patients with early-onset AO and those with late-onset AO.

Finally, the paucity of data on patients' employment activities in previous studies was noted by Buades-Reinés et al. (2016). This is an important reality that needs to be considered. When considering the real impact of the disease on the lives of those affected, this is particularly important. It is essential for a project such as this one, where the aim is to have occupational interventions for people with the diagnosis. The assessment of occupational aspects during this project revealed that most patients were not working or were retired due to their ATTRv symptoms.

4 Psychosocial impact of Variant Transthyretin amyloidosis diagnosis

A person acts and feels not in accordance with what things really are, but in accordance with his mental image of reality
Eric Berne

QoL is defined by the World Health Organization (WHO) as:

> A person's perception of his position in life within the cultural context and value system in which he lives and in relation to his goals, expectations, standards and concerns. It is a very broad concept that can be influenced in a complex way by the physical health, the psychological state, the level of autonomy and the social relationships of an individual (WFOT, n.d.).

Thus, QoL would be related to how individuals perceive their position in life, the context and values in which they live, and their goals, expectations, aspirations and concerns. An assessment of the satisfaction or dissatisfaction that individuals feel in relation to their ability to participate in daily activities is taken into account in this description. For example, in relation to work, the ability to study, drive, prepare a meal, hold a job or use public transport.

A period of inability to carry out the activities of daily living has been recognised as a public health problem (Fuchs, et al., 2022). It represents a progressive and highly disabling deterioration. It has physical, psychological, social and occupational consequences.

Patients diagnosed with ATTRv may perceive their QoL to be affected by the anxiety associated with lack of autonomy, as well as the symptoms themselves. This is an area that has not yet been the focus of research. The importance of psychosocial support in influencing patients' daily lives after diagnosis has been shown in several studies. It can even have an impact on adherence to treatment, recovery, subjective perception of the disease, rehabilitation and adaptation to the disease. This means: Psychosocial support for people who have been diagnosed is part of a network that helps them to remain independent. (Damy, et al., 2022).

Psychosocial resources are currently scarce, in line with the research developed in this project and the psychosocial needs identified in ATTRv patients. This means that patients do not receive the full range of helpful non-medical interventions when diagnosed.

5 Occupational therapy

5.1 Occupational therapy definition

Occupational Therapy (OT) is a person-centred health discipline concerned with promoting health and well-being through occupation and activities of daily living. Enabling people to engage in meaningful participation and activities is the primary goal of OT. (Sakellariou and Pollard, 2017).

Occupational therapists achieve this outcome by working with individuals and communities to improve people's ability to engage in activities they want, need or expect, or by adapting activities or environments to better support activity engagement (Santos del Riego, 2005).

Occupational Therapy can be used to intervene in several areas, including:

- Education (class participation, reading, writing, use of school equipment).
- Recreation (playing).
- Social participation (socialising, self-regulating).
- Daily activities (grooming, eating, getting dressed, personal hygiene).
- Instrumental ADLs (meal preparation, shopping, money and medication management, grocery shopping).

- Sleeping and resting (sleeping hygiene, relaxing, managing stress).
- Managing health (self-care).
- Work life

5.2 Historical vision of Occupational Therapy

Since the dawn of civilization, activity, movement and occupation have been the subject of therapeutic use. A journey through history from its origins to the present day will be undertaken to better understand and contextualize OT (Morrison et al., 2011). This will be in the context of the fundamental role of occupation from the point of view of different communities at different times.

The Chinese believed that disease was caused by organic inactivity in 2600 BC. They used physical training through exercise as a means of health promotion and even, in their belief, as a means of ensuring immortality (Tipton, 2014).

In Egypt, temples were built around 2000 BC. Melancholiacs flocked to these temples to alleviate their illness. Leisure, games and recreation were used as therapeutic tools in these temples. A few years later, in the Greek and Roman cultures, important figures such as Aesculapius, Hippocrates and Galen began to appear. They were already advocating the therapeutic use of moving and working. Exercising has always been seen as something positive, not only for the physical action it involves, but also for how it affects the mind, as is said: "The best exercises are those which do not only train the body, but also give pleasure to the mind. (...) Activity is nature's best doctor. It is indispensable for the happiness of man". (Santos del Riego, 2005).

This was followed in classical Greece by Escupalius in 600 BC, as APETO notes. He used song, music and literature to alleviate delirium and founded a hospital in the city of Pergamum. Between 600 BC and 200 AD, Pythagoras, Thales of Miletus and Orpheus used music to treat various illnesses. Hippocrates recorded wrestling, reading and work for well-being. To improve disturbed minds, Cornelius Celsus (14-37 AD) recommended music, conversation, reading and exercise to exhaustion. It is important to note that his recommendations were for occupations appropriate to each person's temperament. This is what we would now call a person-centred approach to occupational therapy. Later, Seneca (55BC-39AD) recommended work for every kind of mental disorder, and Galen (129-199AD) promoted occupational treatment, stating that work was the best natural medicine and the basis of human happiness (APETO, n. d.).

Scientific interest turned to the analysis of movement between 1250 AD and 1700 AD. Rhythm, posture and energy were studied by writers such as Leonardo, Descartes and Bacon. Ramazzini emphasized prevention over treatment and the importance of observing the patient while working (WFOT, n.d.).

However, it was not until the 18th and 19th centuries, with the rapid development of psychology, anatomy and physiology, that these new principles had an impact on physiotherapy and occupational therapy (APETO, n.d.).

In 1780, Tissot recommended violin-playing, sewing, hammering or bell-ringing as therapeutic interventions, and classified occupational exercises as active, passive or mixed. Nine years later, Pinel, who believed that human work, rigorously performed, was the best guarantee of good morals and discipline, introduced work therapy at the Bicetre asylum, prescribing physical exercises and manual occupations for mental illness (APETO, n.d.).

Physiotherapy and the beginnings of OT were introduced during the First World War. However, it was not until the Second World War that OT had official recognition. Towards the end of the war, the United States War Department employed Reconstruction Aides (RAs) to help wounded and troubled soldiers recover from the panic, depression, stress and anxiety of return home. Civilian women were these RAs. They worked in military hospitals, both at home and abroad, to help the soldiers to rebuild their lives (APETO, n.d.). OT RAs taught vocational skills and set personal goals to distract the injured while increasing their productivity and motivation. Military hospitals recognized the need to help soldiers with psychiatric and orthopedic problems between 1939 and 1945, during the Second World War. The profession expanded again with the war effort. In 1944, the OT was recognized by the United States Army. It created special training courses for OT Assistants (OTAs) (WFOT, n.d.).

History suggests that as part of the discovery of the value of occupation as a treatment, the OT profession emerged in the late nineteenth century. However, since the beginning of civilization (Newton, 2007), occupation has been used as a tool to treat a variety of conditions.

Pinell's moral treatment

Moral treatment developed in response to the approach to mental illness. At first, people with mental illness were seen as animals and delinquents. According to this point of view, these people were dangerous. They were excluded from society because they could not be responsible for their own actions. Beating and chaining were also common practices in lunatic asylums. However, not everyone agreed with the view that mental illness was incurable (Pelletier and Davidson, 2015).

This view began to change as the causes of mental illness began to be seen as changing from somatic to psychological. Thus, from the humanist movement, there was a development of moral treatment.

Within moral treatment, two important figures have been identified: Samuel Tuke and Philippe Pinel. Pinel, who triggered patients, established common rules in the treatment of psychoses (Trent, n.d.).

The patients have to have an organization of all the activities that they do.

For the treatment of idleness, work and organised activity become a treatment.

Occupational Therapy in the twentieth century

OT has evolved over the years. Three countries, the United States, the United Kingdom and Spain, have played an important role in the development of the profession since the beginning of the 20th century. As has been presented (Reed, 2018):

United States

Adolf Meyer, a psychiatrist working in the late 19th and early 20th centuries, provided the philosophical basis for the development of OT. Meyer accepted that the rhythms of life (BADL, IADL, AADL) needed to be kept balanced. This was achieved by carrying out habitual activities, with a healthy lifestyle providing the basis for general wellbeing.

In 1892 he made the observation that: "The proper use of time in some useful and rewarding activity seems to be a fundamental problem in the treatment of neuropsychiatric patients".

A founding father of the field is Dr William Rush Dunton Jr. A psychiatrist, he began using OT to treat the mentally ill as early as 1895.

Herbert J. Hall, an early twentieth century physician, prescribed OT to individuals as a form of medicine to help direct interests and regulate life. He established a workshop in 1906. He used hand weaving, wood carving and ceramics to treat neurasthenia.

Hall argued that: "An important factor in maintaining physical, mental and moral health is the proper occupation of the hand and mind.

In 1919, the basic principles for occupational therapists were established by Dr William Rush Dunton Jr:

- Occupation is as necessary to life as eating and drinking.
- Every human being needs to have a physical and a mental occupation.
- Every man must have occupations in which he can have practice or diversion.
- These are the most necessary when the vocation has gone out or has become unpleasant.
- Everyone must have at least two occupations: inner and outer.
- A large number of them will be a broadening of interests and intelligence.
- Occupation can heal the sick mind, body and soul.

The following points briefly summarise the valuable legacy received from these ancestors:

- Considering manual and creative work as restorative.
- Using the patient's needs, values and interests to stimulate constructive activity.
- Addressing pathology by using existing skills and developing new ones.
- The importance of establishing and maintaining a therapeutic process through interpersonal relationships and interactions.
- Reducing limitations by developing techniques and equipment.
- Acceptance of the basic advice that meaningful and purposeful occupation can be a positive force in the influence of an individual's health status.

Great Britain

The introduction of OT in Scotland was during the First World War. OT was introduced into a modern psychiatric hospital by Dr Henderson in 1919. In 1925 the first occupational therapist, Margot Fulton, was in post in the UK. It was not until 1930 that the first OT school was in place, at Dorset House. This was followed within a few years by schools in London, Northampton and Exeter. Six years later, the first department and training centre was established at the Astley Ainslie Hospital in Edinburgh by Dr Cuningham, with the help of an occupational therapist from Canada (Reed, 2018).

Spain

The National School of Occupational Therapy (ENTO) was established in 1967 and is governed by a Board of Governors.

On 27 June 1967, BOE No. 152 published a decree stating that OT would be governed by ENTO, which would be attached to the National School of Health as a subsidiary body and would be managed by a Board of Directors made up of a President, a Vice-President, five members and a Secretary (APETO, n.d.).

From the beginning, ENTO's criterion was to adapt its programmes to those of the World Federation of Occupational Therapists (WFOT). It maintained a link with this federation until its definitive integrity (13 August 1970). It was in this way that it acquired its titles, its recognition and its international validity. The studies of the TO seem to be finally recognised as a university degree of intermediate level with the law on the reform of the universities (BOE 20/10/1990). The first of these schools was set up at the University of Zaragoza. These were joined by the Complutense University of Madrid, the Red Cross School of Tarrasa and the CEU of Talavera de la Reina (APETO, n.d.).

The decree that regulates the validation of degrees obtained in the former ENTO was published in the BOE in 1996. OT is in many ways a young discipline. However, there is a remarkable number of references from ancient times in the scientific literature, especially in the beginnings of neurology and psychiatry (E. Fossey 2001). These bring us closer to the origins and the main foundations that guide the profession today.

5.3 Occupation as an intervention tool.

A regular activity or hobby.
A situation in which an army or group of people move into and takes control of a place.
The fact of living in or using a building.
A job.
A regular activity.

The occupational therapist intervenes through occupation and through the person's daily life. To understand occupation as a tool, it is important to consider the dimensions of occupation and the domains from which OT can address these dimensions (WFOT, n.d.).

The main dimensions of occupation are:

- Activities of Daily Life (ADL), basic, instrumental and advanced, such as: Personal care: eating, bathing, dressing and ambulating; Transportation.
- Leisure time: For mental and physical relaxation, stress reduction and reduction of medication use.
- Work: Domestic tasks (watering plants, vacuuming, making beds), finding and re-finding work.

According to these dimensions, the occupational therapist profile will be present in different areas as follows (APETO, n.d.):

- People with physical and mental disabilities.
- Geriatrics: OT is used in the treatment of both healthy and sick elderly people. For the healthy elderly, with their limitations and the progressive and cumulative losses typical of old age, OT should be preventive. It should maintain optimal autonomy. For the disabled elderly, in order to promote their reintegration into their social and family environment, comprehensive rehabilitative OT is offered.
- Pediatrics: The aim is the promotion of the child's personal autonomy in all areas of his or her life and the provision of skills appropriate to each stage of development.
- Social exclusion: the occupational therapist intervenes and works to promote or re-establish routines in situations of isolation or lack of activity for social reasons.
- Home OT: The occupational therapist facilitates or maximizes the patient's level of functioning in a domestic context through personal care, home care and recreational therapy activities.
- Education: The OT trains children to be more active participants in the education process.

5.4 Occupation and quality of life

The quality of life is more important than life itself
Alexis Carrel

Occupation is of fundamental importance to people and has a direct impact on their quality of life and general wellbeing, as it has been explained in previous sections (APETO, n.d.).

The person's abilities are assessed and goals and objectives are set by the occupational therapist. These will be developed during the course of the intervention and are subject to ongoing evaluation. This is followed by assessing the person's autonomy and independence. The result is an improvement in the person's aptitudes, attitudes and skills for the correct performance of general activities. (De las Heras, 2009). All these processes are carried out using Activity ans Activity is seen as the essence or means of treating. It is not an end in itself, but a means to the achievement of general objectives set by the therapeutic team (Stoewen, 2017).

Activity is important in everyday life because it is part of it and because it involves the process of doing. It is necessary for human beings to exist and survive and it is under the control and guidance of culture and environment (WFOT, n.d.). Most of all, it is something that can be learned.

Some of the characteristics of OT activities for the achievement of the above goals may be:

Each activity needs to have a meaningful purpose and a specific goal or objective to be achieved.

The activities need to be meaningful or relevant to the individual in question. There is a need for commitment to participation and persistence in order to achieve a positive outcome. Discipline and commitment are required.

The activity should not only be an increase or maintenance of the individual's level of functioning. It should also prevent possible or future limitations from occurring.

Generic models cannot be used in the OT discipline. All activities must be adapted to the needs of the individual and their environment.

All activities are assessed by the occupational therapist to understand and sequence the whole process.

To make them functional in the occupational process, adaptations must be made.

6 Model of Human Occupation (MOHO)

¿We are what we do or we do what we are?

Several theoretical occupational models exist. In this project, we are going to focus on MOHO and what it offers. We feel it is important to break down certain concepts in relation to the model used in this project.

What is an occupational model?

An occupational model is a theory that explains some aspect of human behaviour, approaches practice from an OT perspective and provides tools for applying this theory. An essential part of an occupational model is research. The theory and tools are tested and improved through research. (Kilhefner, 2004).

Human Occupational Model

The Model of Human Occupation (MOHO) explains how occupations are motivated, modelled and performed in everyday settings, according to Kielhofner (2004). This model has been described as the most cited and most widely used model of practice with a focus on occupations in the world.

History and origins

To understand how this conceptual model has developed from theory to practice, it is necessary to look at the history of OT. As a psychosocial discipline, OT is concerned with the validation of its own knowledge outside of the medical discipline. It does this by setting real and tangible goals for intervention with the individual. Around these central concepts (Kilhefner, 2004) an OT paradigm has been developed.

In challenging the discipline to move beyond the mechanistic paradigm and towards what would become the contemporary paradigm of OT, Mary Reilly was a key figure in the profession. Her hypothesis about the audacity of OT was emblematic of the time when he said: "Through the use of his hands, powered by his mind and will, man can influence the state of his own health". (De las Heras, 2009).

Mary Reilly was concerned with the recovery of the professional identity. She sought its reaffirmation in the training programmes of the OT and proposed a return to the principles of moral treatment. She also developed her own postulates, used as a frame of reference, which she called professional behaviour, and had as principles, the relevance of motivation to the profession, the importance of the sense of time in the performance of professions and the influence of the environment and its support or obstacle to adaptation. On the other hand, she highlighted the concept of exploration as the basis of all learning. She linked it to play, developing a game theory with other professionals (De las Heras, 2009).

These principles have been developed by a number of occupational therapists who have contributed to the return to the occupation-centred paradigm. These include Gary Kielhofner and Janice Burcke, who began to develop the Human Occupation Model in 1975 from their doctoral research. Its contributions to the development of knowledge in OT are described as an emerging paradigm, up to the conceptualisation of the current paradigm, which has facilitated the reaffirmation of the professional identity. Other disciplinary perspectives have been added to this development, such as Occupation Science and other occupation-centred models of practice, such as the Canadian Model of Occupational Performance, among others (De las Heras, 2009).

The development of the Model of Human Occupation since its inception created a conceptual basis at the beginning of the discipline, in the pre-paradigm of occupation, and together with other representatives of the discipline, contributed to the positioning of the contemporary paradigm, in what Polatajko called 'an occupation-centric discipline' (De las Heras, 2015).

The model of human occupation was first published in 1980. It grew out of Gary Kielhofner's master's thesis and practice in the mid-1970s. It has always been a collaborative effort, although Kielhofner was the main person working on MOHO.

What is the MOHO approach?

MOHO seeks to explain how occupation is motivated, how it is structured and how it is performed. MOHO provides a broad and integrative view of human occupation (Park, et al., 2019) by offering explanations for these diverse phenomena.

Within MOHO, the human being is conceptualised as being made up of three interrelated components:

- Volition: the motivation to engage in occupation.

- Habituation: the process by which occupation is organised into patterned or routinised performance.
- Capacity or performance: the physical and mental abilities that are the basis of skilled occupational performance.

Park, et al.'s (2019) definition of MOHO emphasises that an understanding of the physical and social environment is critical to an understanding of human occupation. These concepts provide a clearer and more accurate explanation of how these factors interact to influence people's daily activities, and why problems arise when chronic disease, deficits or environmental factors limit occupation, and have remained consistent throughout nearly three decades of model development.

The evolution of the components of the MOHO model

Since 1985, the development and evolution of this discipline has been made possible by the collaboration of theorists and researchers with occupational therapists. There has also been evidence-based development of the model, both conceptually and through the use of different assessment and intervention tools (Kilhefner, 2004).

The context

The context is a fundamental principle that needs to be taken into account in a professional intervention. Respect for autonomy has been included as a key factor in ethics applied to medicine (Kilhefner, 2004) as a result of the recognition of vulnerability in health care contexts.

The different contexts and their influence on the profession were defined by De las Heras, 2009:

Environments and contexts

When a patient participates in an activity, this activity takes place in a social and physical environment. This environment is in turn in a context. In the OT Practice Framework, environments and contexts are used to describe the importance of considering all aspects that influence people. Such conditions can be internal and external.

The environment is the physical and social setting in which a person's day-to-day life takes place. Natural or artificial surroundings and objects are the physical environment. The social environment is the set of relationships and expectations of people, groups and organisations that the individual is a part of.

Contexts are various conditions in or around the patient. The cultural context includes traditions, beliefs, patterns of activity, norms of behaviour and expectations accepted by the society of which the patient is a member.

A person who finds it difficult to function in one context may succeed by moving to another. For a better understanding of this idea, here is a description of the different environments and contexts:

Physical environment

The main characteristics of the place where an activity takes place are described as the physical environment. Performance capabilities, performance patterns, demands of activities performed and patient limitations should be considered as part of the assessment process when considering occupational therapy intervention.

Social environment

Individuals are embedded in a social environment based on people and social relationships. This means that people integrate into their living system some expectations based on other people and organisations in which they participate.

In OT, the social environment is important throughout the intervention process. Not only is the patient being intervened upon, but there will also be an impact on carers and family members.

Cultural context

Cultural differences can have an impact on people's participation or well-being.

There are established behaviours that need to be followed depending on where the person lives or the organisations to which they belong. In addition, people are entitled to different resources and opportunities depending on the laws that govern a place. These attitudes need to be analysed for an appropriate OT programme.

Personal context

The personal context is defined as all those characteristics that make up the person and are not related to their condition or health. In addition to age, gender, socio-economic level and level of education achieved, the level occupied in a company or in a type of work can be included.

Temporal context

The moment or stage at which the occupational situation changes is important to examine. The causes, the illness or the situation that leads people to request or to receive OT services are occurring.

Virtual context

Advances in technology mean that we are constantly in touch and in touch with each other. It is important to know if patients know about the different means of communicating, if they know how to use them or if they are essential to perform some of their activities.

7 Knowledge about Occupational Therapy

7.1 Professional identity

In the mid-twentieth century, World Health Organisation (WHO) established an enduring definition of health as "a state of complete physical, mental and social well-being" rather than "the absence of disease". This definition now recognises a wide range of personal, social, economic, occupational and other factors as determinants of both individual and community health. (WHO, n.d.).

The WHO describes health as a factor of everyday life. It is a function of social and personal resources as well as physical capabilities. It proposes two basic strategies for the development of health in communities around the world (WHO, n.d.). These strategies are

Advocacy for the political, environmental, economic, social, cultural, biological and behavioural conditions essential for health.

Empowerment of people to promote health potential and prevention rather than cure.

WHO calls for collective action in different sectors of society to set and achieve the goals, with an emphasis on social justice and equity. This is because the determinants of health and well-being, such as serious illness and death, lie outside health services. It is in this area that a key role in health promotion would be played by social health professionals such as psychologists, occupational therapists, social workers, genetic counsellors, etc. A fundamental idea for the development and promotion of OT as a social health discipline is that: "People's health is significantly affected by changes in the way they live, work and spend their leisure time. (WFOT, n.d.).

Universal Declaration of Human Rights

The importance of all people being able to participate in occupations is recognised in the United Nations Universal Declaration of Human Rights. However, the opportunity to do so is not available to everyone. Mental health professionals and occupational therapists should be particularly concerned about this. Lack of health and occupation can be the cause of physical, mental and long-term disability (Whalley, 2008).

However, as Whalley (2008) affirms, occupational therapists need to be aware of their privileged position and obligations in promoting occupational rights and freedoms at all levels. These rights and freedoms include 'free development of personality' through practice, free choice of work with 'just and favourable remuneration', time and opportunity for rest and leisure, adequate educational opportunities, opportunity for participation in the governance of a country, participation in community and culture.

Yet in many parts of the world, these rights are not in place. Some of them are overlooked even in the most highly developed countries (Buettner-Schmidt and Lobo, 2012). Reality differs from theory. Some of the difficulties currently faced by occupational therapists include a shortage of trained professionals, lack of collective motivation, low impact on the scientific community and difficulty in defining their own profession.

7.2 Occupational therapist professional reality

Hammond, (2004) defines the professional identity of OTs as a fundamental concept that encompasses not only the role of these professionals in the workplace, but also the unique values, beliefs, skills, abilities and responsibilities. This set of concepts defines who occupational therapists are as healthcare professionals and guides their interventions with patients as well as their contribution to the wider healthcare system and working with other professionals.

There are some key aspects to consider when defining professional identity in occupational therapy. These are (Hammond, 2004)

General values:

OT is based on a set of core values which include person-centred care, multidisciplinary collaboration, empathy, advocacy and a commitment to ethical practice.

Occupational approach:

As a key tool in their approach, OT professionals advocate the importance of 'occupation' in people's daily lives. Occupation refers to the range of activities and tasks in which people regularly participate, such as caring for the body, working, recreating and playing.

OTs should intervene to improve people's overall wellbeing and quality of life when changes or limitations occur that prevent or hinder participation in these important activities.

Education and training:

Occupational therapists need specific training that enables them to work with the necessary skills in order to establish and define their professional identity.

This means that they need to be educated and trained. University education is essential for the development and definition of the professional and the profession.

Holistic vision:

OT professionals take a holistic and comprehensive approach to psychosocial care. They consider the physical, psychological, social, occupational and environmental factors that affect a person's ability to engage in meaningful activities. They look at the person as a whole.

It is this holistic perspective that distinguishes OT from other health care disciplines that focus on a specific aspect of the person.

Person-centred care:

Person-centred care is the occupational therapist's professional identity. Occupational therapists consider the goals and preferences of each patient as an individual, rather than as a generic factor. They work collaboratively to develop individualised intervention plans. They aim to achieve meaningful goals.

Multidisciplinary team:

OTs work as part of a multidisciplinary team and collaborate with other professionals. They work with other professionals such as doctors, physiotherapists, speech therapists and social workers. This ensures a holistic approach.

The professional identity of occupational therapists is emphasised by this collaborative approach. On the contrary, a reality described so far is the lack of knowledge about the occupational therapist profile, which is an obstacle to the access to this discipline as well as to the promotion of the profession between patients and health care professionals (Moore, 2006).

Professional ethics:

Occupational therapists work within a code of ethics. This code guides professional behaviour, sets parameters for action and regulates interventions. Ethical principles are a fundamental part of occupational therapists' identity, such as confidentiality, informed consent and professional competence.

On the other hand, these professionals are part of professional groups or associations. This allows them to work in a homogeneous way, according to the same standards of action and with the support of the group itself.

Legislation:

Occupational therapists, both individually and collectively, advocate for rights and well-being. One of the roles is to remove barriers of any kind that limit participation in meaningful activities. This includes promoting the creation of accessible environments as well as policy changes.

Continuous learning:

Continuous learning and updating of the profession are linked to the professional identity of OTs.

Professionals need to keep abreast of the latest research, techniques and interventions in order to provide effective services. They also need to undertake activities that enable the profession to be disseminated and developed.

Society impact:

OT professionals enable people of all ages and abilities to live meaningful lives and have a significant impact on society.

In summary, the professional identity of occupational therapists includes a commitment to person-centred interventions, values, a holistic vision, a multidisciplinary approach, ethical behaviour and education. All of these aspects fully define the role and responsibilities of occupational therapists

as well as their work to promote health and well-being using occupation as a means to an end (Walder, et al., 2022).

8 Occupational approach in chronic diseases

OT plays a key role in managing and rehabilitating people with chronic conditions (Hammond, 2004). Chronic conditions are defined as: "long-term conditions that require ongoing medical care and psychosocial support. These conditions and symptoms often have an impact on a person's ability, and sometimes their context, to engage in meaningful activities of daily living and maintain their quality of life (Kristensen, 2012).

Occupational therapists are specifically trained to address functional limitations (physical, psychological, social, environmental and occupational) and challenges associated with chronic conditions (Walder, et al., 2012).

Managing daily living activities

The physical, psychological, social and occupational abilities of patients and carers change as a result of living with chronic illness. OT professionals are tasked with analysing the overall demands of activities that are meaningful to each person and assessing the fit between the abilities and challenges posed by those activities and the environment (Lambdin-Pattavina and Pyatak, 2022).

Occupational therapists can make recommendations to conserve energy, reduce or prevent pain, simplify activities, improve safety and ease of work in a given environment.

Lambdin-Pattavina and Pyatak (2022), provide an approximation of the areas where the occupational therapist has an essential role in the approach to chronic disease.

Health and routines

Managing chronic conditions involves learning specific tasks for managing health. These tasks may include regularly monitoring blood pressure or weight; preparing meals according to specific needs or dietary restrictions; administering oral, injected or inhaled medications; or increasing physical activity. One of the occupational therapist's roles is to ensure that these activities are carried out consistently and regularly in order to be correct and effective, and that they are integrated into existing routines. OT professionals work with patients to integrate health managing tasks into daily routines and, where necessary, to incorporate accommodations to simplify the demands of these

tasks. These professionals are particularly skilled in the management of lifestyles that fit into existing routines and patterns, so that changes in routines are timed and are more likely to be properly integrated (Walder, et al, 2022).

Emotional well-being

OTs understand the emotional changes associated with living with a diagnosis of chronic illness. Anger and depression, feelings of uncertainty about the future, and changes in relationships with the environment, among others, are fundamental aspects to consider in developing an appropriate occupational intervention. It is important to recognise that wellness is more than managing symptoms or performing daily tasks. The therapist focuses on what is meaningful and develops personalised interventions (Walder, et al., 2022).

Patient assessment

To understand the limitations and needs of people with chronic conditions, therapists conduct comprehensive assessments using interviews and standardised scales. The aim of the assessment is the identification of physical, cognitive, emotional, occupational and social abilities in order to develop a specific individual intervention (Walder, et al., 2022).

Goal setting

Occupational therapists will work with the patient to set realistic goals for the short, medium and long term.

Self-management

OTs provide people with information about their chronic condition and teach self-management strategies. These include techniques for symptom management, medication management and stress reduction. On the other hand, they enable developing coping strategies, behaviours, habits, routines and lifestyle adjustments (Lambdin-Pattavina and Pyatak, 2022).

Conserving energy

For people with chronic fatigue, such as those with multiple sclerosis or fibromyalgia, occupational therapists offer energy conservation techniques to help maximise performance at work (Walder, et al., 20-22).

Assistive technology and assistive technology products

Occupational therapists can recommend and provide training in using assistive devices and coping strategies to promote autonomy (Walder, et al., 2022).

Pain management

Occupational therapists can implement and teach pain management strategies such as ergonomic modifications, relaxing techniques, and phased activities (Walder, et al., 2022).

Psychosocial support

Emotional and psychological challenges are associated with many chronic conditions. Occupational therapists offer emotional support and strategies to manage anxiety, depression and other mental health issues with an occupational perspective (Lambdin-Pattavina and Pyatak, 2022b).

Work

Following on from this work, Fields and Smallfield (2022) describe therapists' interventions with people with chronic conditions in order to return to work or maintain employment. Interventions may focus on recommending adaptations and modifications to the workplace.

Treatment adherence

Occupational therapists work with patients to improve their adherence to treatment. This includes medication management, exercise routines and dietary changes (Lambdin-Pattavina and Pyatak, 2022).

Carer support

Occupational therapists also provide education, awareness and support to carers, helping them to understand the needs of patients with chronic conditions and teaching them strategies to provide better care and support (Lambdin-Pattavina and Pyatak, 2022).

Advocacy and promotion of social justice

OT can intervene with people with chronic conditions in a variety of settings, including schools, businesses and healthcare systems, to ensure they receive necessary accommodations and services equitably. In general, the focus of OT in chronic illness is on improving the functional independence and quality of life of the individual, while at the same time helping them to cope effectively with the new daily life after diagnosis. The main aim is to enable people to live as independently and as fully as possible in spite of the challenges posed by a chronic health condition (Fields and Smallfield, 2022).

9 Occupational approach in rare diseases

Lou (2002) states that given the involvement of the occupational therapist in chronic diseases, the involvement and role of the occupational therapist in rare diseases could be considered:

- General assessment: occupational therapists carry out assessments and follow up to identify the challenges faced by people living with rare diseases. The OT assessment will cover the physical, cognitive, emotional, social and vocational aspects.
- Goal setting: occupational therapists work together and in collaboration with the patient to set realistic goals. The focus of these goals is on the improvement of occupational performance and day-to-day living.
- Accommodation Occupational therapists work with adaptive strategies and techniques to manage the functional limitations caused by the symptoms of the rare disease, so that the person can continue to develop meaningful activities.
- Modifying the environment: A rare disease may be associated with symptoms that require modifying the environment to improve access, safety or use.
- Conserving energy: Fatigue is a common symptom of many rare diseases, as it is with chronic diseases. Occupational therapists work with energy conservation techniques to help people manage their energy levels effectively. This can reduce the impact on daily life.
- Pain management: therapists can provide pain management strategies, such as ergonomic guidelines, relaxation techniques and aids to daily living, for rare diseases associated with chronic pain.

- Cognitive rehabilitation: Rare diseases can have an impact on cognitive function. Occupational therapists work with rare disease patients to improve memory, attention, problem solving and executive function.
- Emotional support: The emotional impact of living with a rare disease should be addressed as a factor of daily life. Occupational Therapists provide emotional support, coping strategies and occupational mental health interventions to individuals and their families.
- Social participation: Occupational therapists help people to maintain or regain their social roles and to participate in meaningful activities. This may include programmes to help reintegrate into the community and training in social skills.
- Multidisciplinary team: Occupational therapists work closely and collaboratively as part of a multidisciplinary team. This may include genetic counsellors, doctors, physiotherapists, speech therapists, dieticians, social workers and other professionals.
- Education and awareness raising: Occupational therapists educate patients and their families about the rare disease, the progression of the disease and possible treatment options from a vocational point of view. They also promote awareness and understanding of how the disease affects people's lives, among healthcare professionals and the community at large.

OT in the rare disease context is highly specialised and aims to improve the person's overall wellbeing and autonomy, quality of life and subjective perception of the disease (Meyer et al., 2022).

The scarce bibliography published in the field of OT and specifically in the field of rare diseases makes this pioneering project a new avenue of investigation in which specific occupational interventions for people with a rare disease diagnosis are being studied and developed.

10 Occupational Approach in Variant Transthyret in amyloidosis

10.1 Occupational impact of ATTRv diagnosis

This pathology is very limiting in daily life (Buades-Reinés et al., 2016). Due to the lack of research, it is still not clear what the real impact is on daily life after an ATTRv diagnosis, an area that is being studied.

11 The Patients Advocacy groups

People with people

Historically, patient organisations have played an essential role as social actors in defending collective rights and providing services, support and resources to patients, carers and their families living with a rare disease. They typically provide services including health information, empowerment and education, social awareness, rehabilitation, social support, psychological and legal support (Patterson, et al., 2023).

The main functions of patient advocacy groups according to Patterson, et al. (2023) are

To advocate and provide legal representation:

Patient advocacy groups are advocates for the rights and interests of patients with ATTRv. Their main goals are health policy, investigation, research funding and access to treatment, general care and raising general awareness.

Public awareness

Patient advocacy groups provide essential information and educational resources on the diagnosis of ATTRv and medical or psychosocial treatment. They support patients and their families in their understanding of their situation, treatment options and available resources. On the other hand, health care professionals have information and awareness about people with a rare diagnosis. This information is valuable. It empowers patients to make informed choices and decisions about their own health situation.

Benefits available

To provide opportunities to build relationships, share experiences and offer support to people with similar conditions. For emotional, social and occupational well-being, these support networks can be essential.

Funding

Funding or promotion of research initiatives in relation to your diagnosis. You can work with researchers and advocate for increased research funding to advance treatments and cures.

Awareness and education programmes

Patient advocacy groups often organise awareness campaigns for the general public and educational events for healthcare professionals and policymakers. The main idea of these campaigns is to reduce stigma and improve general understanding.

Treatment and care

These organisations work to ensure that people have free access to appropriate treatment. They can campaign for insurance coverage, government funding and better access to mental health services.

Policy and health system engagement:

Patient advocacy groups work with policy makers and health care providers to shape health care policy and practice. They also provide general information about patient conditions, specific guidelines, standards of care and legislation.

Quality of life

Patient advocacy groups focus on improving the overall quality of life of people with rare diseases. This includes considering the psychosocial professions, facilitating access to support resources, and promoting research into symptoms, management and rehabilitation.

Idea of community

Creating a sense of community for people and their families faced with a rare disease diagnosis. Associations often organise events, congresses, conferences and workshops to facilitate interaction, increase the visibility of the disease, and provide a platform to share experiences and knowledge not only between patients and families, but also with professionals, allowing the creation of better services.

Empowerment

Patient advocacy groups can provide training to empower people to advocate for their own interests and those of the community. These programmes teach self-advocacy skills, disease awareness, communication and recognition of the health care system.

In conclusion, patient advocacy groups play a fundamental role in improving the daily lives of people living with a diagnosed rare disease. They enable positive changes within the healthcare system that benefit all patients by acting as powerful advocates, sources of information and support (Patterson, et al., 2023).

The current associations in Spain with a focus on support for people with ATTRv are:

Asociación Balear de la enfermedad de Andrade **ABEA**	
Asociación Valverdeña de la enfermedad de Andrade. **ASVEA**	
Asociación Canaria de la enfermedad de Andrade. **ACEA**	
Amiloidosis Visible	

Nacho Varela

Chapter II

To do or not to do (that is the question)

1 Thesis objectives. OCUPAT: OCUpacional Project in Patients with hereditary Transthyretin amyloidosis

1.1 General objectives

- To develop a specific project related to occupational therapy and Variant Transthyretin Amyloidosis.
- Contribute to the bibliography on the subject to help provide new avenues for scientific research to follow.

1.2 Specific objectives

- To assess the basic occupational situation of patients diagnosed with variant transthyretin amyloidosis.
- To promote the motivation and interest of people who have lost their ability to work.
- To acquire, improve and develop the skills needed to achieve a professional performance that is as satisfactory and as functional as possible.
- To take a holistic and humanistic approach to raising awareness of variant transthyretin amyloidosis.
- To include the occupational therapist in the work team dealing with patients with rare diseases through outreach work with patients and other professionals.
- To analyse the situation of occupational therapy from the point of view of the Iberian Peninsula, between Spain and Portugal.

1.3 Hypothesis

- Primary hypothesis: The application of a specific occupational therapy programme for ATTRv will improve patients' occupational autonomy and quality of life.
- Second hypothesis: Patients with ATTRv who are approached from an occupational perspective will score lower on depression and occupational scales compared to those who do not receive any intervention.

2 First manuscript. Val50Met hereditary transthyretin amyloidosis: not just a medical problem, but a psychosocial burden

González-Moreno, J., Gayà-Barroso, A., Losada-López, I., Rodríguez, A., Bosch-Rovira, T., Ripoll-Vera, T., Usón, M., Figuerola, A., Descals, C., Montalà, C., Ferrer-Nadal, M. A., & Cisneros-Barroso, E. (2021). Val50Met hereditary transthyretin amyloidosis: not just a medical problem, but a psychosocial burden. Orphanet journal of rare diseases, 16(1), 266. https://doi.org/10.1186/s13023-021-01910-5.

3 Second manuscript. Accessibility to Occupational Therapy Services for Hereditary Transthyretin Amyloidosis

Gayà-Barroso, A., González-Moreno, J., Rodríguez, A., Ripoll-Vera, T., Losada-López, I., Gili, M., & Cisneros-Barroso, E. (2022). Accessibility to Occupational Therapy Services for Hereditary Transthyretin Amyloidosis. International journal of environmental research and public health, 19(8), 4464. https://doi.org/10.3390/ijerph19084464.

4 Third manuscript. Establishing Occupational Therapy Needs: A Semi-Structured Interview with Hereditary Transthyretin Amyloidosis Patients

Gayà-Barroso, A., González-Moreno, J., Rodríguez, A., Ripoll-Vera, T., Losada-López, I., Gili, M., Paneque, M., & Cisneros-Barroso, E. (2022). Establishing Occupational Therapy Needs: A Semi-Structured Interview with Hereditary Transthyretin Amyloidosis Patients. International journal of environmental research and public health, 19(18), 11721. https://doi.org/10.3390/ijerph191811721.

5 Fourth manuscript. Occupational practice in patients with hereditary transthyretin amyloidosis, a qualitative study.

Gayà-Barroso, A., González-Moreno, J., Rodríguez, A., Ripoll-Vera, T., Losada-López, I., Gili, M., Paneque, M., Pérez-Martínez, S., & Cisneros-Barroso, E. (2023). Occupational practice in patients with hereditary transthyretin amyloidosis, a qualitative study. Orphanet journal of rare diseases, 18(1), 352. https://doi.org/10.1186/s13023-023-02964-3.

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Chapter III

Tell me what you do and I'll tell you who you are

1 Discussion

Whether personal, professional or social, the impact of being diagnosed with ATTRv appears to affect daily life, roles, routines and habits, and future goals (González, et al., 2021). Access to OT offers an opportunity to re-establish a new normality for people with the diagnosis as well as their families (Gayà, et al., 2022b).

In what follows, we discuss our experience in applying an OT programme to patients diagnosed with ATTRv. The results obtained after 6 months of follow-up encourage us to open a new line of research, since having an occupational therapist as part of an intervention team represents an improvement for this approach as a global concept. After analysing the results, the main hypothesis of the study was confirmed. That is, the application of specific OT programmes reduces dependency and favours the autonomy and quality of life of patients. However, more research is needed regarding the second hypothesis, that an occupational intervention of a short duration, 6 months, would not be sufficient to obtain better scores on the measurement scales.

Despite this limitation, 80% of the patients stated that the presence of the occupational therapist in the process of intervening and approaching after receiving the diagnosis of ATTRv was essential (Gayà, et al., 2022a).

1.1 Not just a medical problem, but a psychosocial burden

The impact of patients and carers on the psychosocial dimension was analysed in the first article. Our results showed that there is a burden on patients and their families. Patients, carers and their families face challenges that go beyond medical issues. The main impact of this disease is on quality of life and mental health, not only for patients but also for asymptomatic carriers and their relatives. According to this information, for 88.5% of participants, the diagnosis of the disease or the identification of the disease-related mutation was considered a life-changing experience, leading to changes in their study, work, daily life, family and future plans.

A survey was carried out to find out what the participants thought of the medical care they received during the diagnosis and follow-up of their disease. In both scenarios, patients rated their care as "remarkable" or "excellent". A multidisciplinary team follows the patients in the study. This multidisciplinary approach aims to meet all the medical needs of these patients in a timely manner. This avoids delays in diagnosis and any necessary treatment. The multidisciplinary approach also aims to reduce the number of hospital visits required by these patients. Regular health education

programmes are organised to provide up-to-date information about the disease. Patients appreciate the existence of a dedicated medical team for this rare disease, according to the survey results. This reflects the fact that needs are met at the medical level, but not at the level of professions such as psychology, social work or OT.

Although clinical needs are met, the results of this research are of great interest as they reflect a specific need shared by families diagnosed with ATTRv, the need for a non-clinical approach (González, et al., 2021).

1.2 Access to occupational therapy

Although the first article recorded the level of satisfaction with the clinical care received by patients, a strong need was expressed for a comprehensive approach involving psychosocial disciplines not yet included in the team, such as occupational therapists. For this reason, the possibility of including the therapist in the team was raised and it was considered necessary to know the patients' level of knowledge of this profession.

Although our results show that ATTRv affects quality of life and the holistic approach is not yet a reality in this group, most patients had never heard of OT and were not familiar with the functions of occupational therapists (Gayà, et al., 2022a). Many patients associated OT with the idea of pastime or entertainment (Darawsheh 2018), a concept that distanced them from the idea of being able to count on the occupational therapist as part of the multidisciplinary team. As previously reported, lack of knowledge about OT can lead to delays in accessing OT services. In addition, the time between the onset of symptoms, the progression of ATTRv patients and the provision of OT services was extremely delayed, despite its crucial role in the implementation of compensatory strategies. The limited existing literature on this discipline also hinders access to OT. Education of health professionals in the multidisciplinary teams treating ATTRv patients, patients and occupational therapists may be essential to ensure that all patients have access to OT and receive appropriate interventions in a timely manner to meet their specific needs.

Of great importance to this project are the occupational therapists' reflections. The therapists' own difficulties in delimiting their work, the lack of protocols and strategies for approaching patients correctly, combined with the lack of a clear definition of their work, make it difficult for the therapists themselves to work in OT.

The same applies to the education and training of therapists, which is increasingly adequate, but at the same time in short supply, as the title of this thesis indicates. Due to the lack of appropriate

doctoral programmes and opportunities for therapists, a doctorate in psychology was undertaken by an occupational therapist.

1.3 Identification de occupational needs

The reality of OT is also reflected in the lack of standardised scales, specific to the discipline, for the measurement of parameters that are relevant for the subsequent application of interventions. The lack of standardised scales is one of the limitations of this project. Identifying needs through a protocol would allow better vocational interventions to be offered in a shorter period of time (Gayà-Barroso et al., 2022b).

The third point of this occupational project was to identify occupational needs by means of standardised scales and a semi-structured interview. The aim was to initiate a specific occupational intervention for each of these needs. The scales that were chosen covered the dimensions of daily life, mental health and symptoms. The interview focused on occupational questions, both before and after diagnosis. The aim was to assess the subjective impact of the diagnosis.

The results of the research showed that although high scores were obtained for the BADL and IADL, this probably reflects the fact that most of the patients included in the study were in stage I of the disease. This situation of independence is expected to change over time as the disease is heterogeneous and progressive. Indeed, in a related study, more than 80% of patients reported that the diagnosis or identification of the disease was a life-changing event that changed their plans and day-to-day activities.

Secondly, it was difficult to collect comprehensive data on baseline occupational conditions due to the lack of knowledge and specific scales, and the paucity of literature on occupational performance in patients with ATTRv. However, the diagnosis of ATTRv has a significant impact on certain aspects of general well-being, such as emotional well-being, social functioning or general well-being, as shown by the results of the SF-36 questionnaire and the WEMWBS scale. Accordingly, for occupational therapists whose profession is focused on promoting health and well-being through occupational interventions, the diagnosis of ATTRv is likely to present a challenging situation.

Patients who have been involved in OT intervention programmes have been shown to experience significant benefits in their daily lives in similar studies of OT and rare diseases. The importance of OT interventions to address levels of dependency in conditions such as CMT is highlighted. Therefore, this information could be extrapolated to ATTRv disease (Matyjasik-Liggett, 2013).

However, most studies using OT in rare diseases have many significant limitations and poor results. To date, this project is the only one using OT in ATTRv patients. A fact that, once again, reflects the lack of OT services aimed at working with people diagnosed with rare diseases and the need to offer this discipline after the diagnosis of these characteristics.

The potential of occupational therapy

Two articles concluded the project. Firstly, the analysis of the effectiveness of an occupational programme and, finally, a broader vision of OT, through the development of a qualitative study with an Iberian vision of Spain and Portugal in terms of knowledge and real situation of this discipline.

OT is defined as the health profession responsible for the promotion and maintenance of health through the use of occupation ((Kilhefner, 2004) The results obtained after applying an occupational intervention programme show that only two scores of ATTRv patients had worsened after a 6-month intervention period according to the Norfolk scale, the main hypothesis was that in addition to ATTRv, patients were diagnosed with other diseases with symptomatology that affected the final scores. To know whether OT would be effective and could contribute to improving final scores, more research and longer OT programmes are needed. It is unclear whether this decline was due to disease progression, or to lack of meaningful activities and to having to give up one's own life. Due to the small sample size and short intervention period, this finding is inconclusive.

According to the Barthel Index (BI) and the Lawton and Brody Instrumental Activities of Daily Living (IADL), the WEMWBS scale and the SF-36 questionnaire, patients achieved similar scores or slight improvements in some aspects, improvements that could be related to the OT programme, but further research is needed to validate this information.

Emphasising the importance of a multidisciplinary team approach, patients recognised that their current situation was likely to deteriorate as their disease progressed and emphasised the need for interventions to maintain general wellbeing. In terms of occupation, 29% (4 patients) of the 14 participants took up new hobbies. These included Pilates, ceramics and martial arts. 21% (3 patients) enrolled in academic courses. 14% (2 patients) became involved in cultural projects. One patient (7%) found a new job. All patients recognised the importance of continuing therapy sessions and the essential role played by the occupational therapists during the intervention period. In their view, the project was a valuable resource for people with rare diseases beyond ATTRv. This was due to the flexibility of the project and its subjective impact. The need for the project to

continue as a valuable resource for rare disease patients was agreed by both the patients and the patient advocacy group.

In addition, it should be noted that patients expressed difficulty in numerically measuring mood states or limitations in daily life, as the concept of daily life is not stable and can be difficult to characterise. Nevertheless, an ideal combination for improving patients' quality of life and occupational balance is the proper implementation of OT projects in parallel with treatment. The psychosocial dimension of ATTRv, specifically OT, should also be further explored. OT remains underutilised in healthcare and in the treatment of ATTRv, despite its unique benefits.

Significant improvements in patients' daily autonomy could be achieved by developing OT for rare diseases, providing adequate training for occupational therapists and integrating their professional profile as part of the multidisciplinary team. This is of benefit not only to the patient, but also to the health care system as a whole, through a reduction in the patient's dependency needs in daily life.

The OT with an Iberian perspective

Although it is challenging, this thesis aims to conclude with a vision of OT beyond Spain. Although characterising a profession is a complex task, the aim of this study was to provide a more comprehensive vision of how OTs see their profession at an Iberian level.

Six Portuguese and four Spanish occupational therapists were interviewed and surveyed. The results of this research revealed themes and sub-themes that were common to both countries. The occupational therapists mentioned the presence of the professional profile in the multidisciplinary team in specific areas. Many other areas were not mentioned. In short, both Portuguese and Spanish occupational therapists reported a significant lack of theoretical foundations of the profession. This leads to a devaluation of the role of the therapist. This reality is in addition to the fact that some of the therapists in this study report that OT was not their first choice of study. They entered this discipline as a second or third option, a fact that may influence the motivation and involvement of professionals in their work. Lack of identity and motivation and low impact on the health system may be related to this finding. Professionals who do not have a definition of their role or who do not believe in the potential of their work are a reflection of the current Iberian reality.

On the other hand, the areas in which they work are very limited. Therapists in both countries commented that working in geriatrics, paediatrics and physical rehabilitation is where OT is most valued. In these contexts, the occupational therapist works with a large number of patients in short

intervention times. This makes it more difficult to provide care and achieve goals, a situation that leads to professional fatigue and burnout. Another reality related to this discipline is that, in some cases, occupational therapists are aware that resources are not sufficient for the patient, who has to pay for private sessions in order to achieve the objectives. These are a number of reasons that guide the development of the profession.

On the basis of all these data, it is considered that the presence of occupational therapists in the approaches offered to patients and as a complement to pharmacological treatment could be a factor that improves the quality of life of patients and maintains the sustainability of the health care system. However, the resources available to occupational therapists are limited and inaccessible to a large part of the population, and there is a lack of professional foundation and professional identity, which hinders the development and implementation of person-centred OT programmes in a multidisciplinary approach.

Findings are consistent with previous articles. The path and development of OT is characterised by a lack of resources and knowledge about OT.

All that can be offered by OT

To conclude, the impact on daily life, roles, routines and habits following the diagnosis of a rare disease such as ATTRv is more than obvious. It is clear that this dimension has not yet been addressed by professionals with the skills to provide effective interventions.

Therefore, as mentioned in the previous sections, in order to demonstrate their work and its effectiveness to those involved, occupational therapists in rare diseases need to differentiate themselves from other areas of work.

The pioneering nature of this work should only serve to further motivate other professionals to continue to contribute to it and to the profession generally.

This is a profession with great potential and contributions to patients, multidisciplinary teams and the health system, which is still far from being widely recognised in the Iberian Peninsula.

Any occupational intervention for people diagnosed with ATTRv must carefully consider each of the issues raised in this paper. To continue with a basic working principle: an occupational therapist is an intervener, not an entertainer.

Limitations

It is important to highlight the main limitations identified in this study with the aim of contributing to future OT projects that focus on working with people diagnosed with a rare disease.

With regard to the sample

The recruitment of the sample, which was initiated through ATTRv patient associations and specific centres, was an important limitation of this thesis. Consequently, the results can only be extrapolated to patients diagnosed with ATTRv who belong to these associations. Conducting this research study with general profiles, and not only with people who are members of associations, would be important. In addition, the fact that the participants were recruited through the associations and that the data collection was focused on psychosocial and occupational aspects in order to avoid a focus on medical aspects has certainly meant that additional information that might have been of interest - such as information on the patient's diagnosis and medication - was not collected.

The small size of the cohort was another important limitation. Unfortunately, there is no centralised registry for ATTRv disease in Spain. This makes it difficult to reach all patients. In addition, the survey was distributed online, and many elderly people or patients in advanced stages of the disease have difficulty accessing a computer. For future studies, the use of paper questionnaires should be considered, or the possibility of answering the questionnaire by telephone for the population in advanced stages of the disease. The survey was only available online for a short time. This may have contributed to the low response rate.

The length of the intervention

Finally, the duration of the intervention is considered as limiting, as a vocational intervention lasting 6 months is a very limited time to obtain potentially valuable results. The possibility of continuing this project is suggested. Regular sessions should be held so that patients can start and finish their occupational plan and obtain real results.

The research's innovative nature

This study is a pioneering global evaluation of the availability and use of OT services for ATTRv patients in Spain. The results show the need for further research into specific factors that influence daily life and possible strategies to address them. Despite this, there has been a lack of significant bibliography throughout the work carried out, as reflected in this thesis.

On the other hand, in order to observe its potential applicability elsewhere, the implementation of the project in the ATTRv group has begun to be applied to other groups diagnosed with rare diseases. There is a need for more research and studies related to occupational therapy in rare diseases and the power of occupations as a working tool.

2 Future proposal

At the end of such a project, after three years of total commitment, both professional and in many cases personal, I have to say that some views and approaches differ from those at the beginning.

The focus of this section will be a brief explanation of areas for improvement as well as future steps to be taken at the conclusion of this journey.

About the Intervention

This dissertation gathers information on new psychosocial approaches in people diagnosed with rare diseases, specifically ATTRv. It is based on the occupational person-centred model. A six-month intervention was carried out. After the first and last evaluation, the methodological limitations resulted in a significant lack of positive results. This means that an intervention of at least one year would be necessary to start new research. The length of time varies from case to case and person to person.

On the other hand, online interventions could be a good way to reach these patients for a more controlled and thorough follow-up. This seems to be particularly important today due to the time, place and cost limitations of face-to-face care. It would therefore be very interesting to conduct further research on the effectiveness of online interventions.

About the sample of patients

It should be noted that a limitation of the results obtained is the small number of patients who took part in the project, fourteen. Consideration should be given to the dissemination of the study in order to have a larger number of participants and results with greater impact within the scientific community.

On the other hand, the importance of working with other groups, including those less frequently diagnosed, must be stressed. This has been done for several months. Replication of the project is a further step towards improving its effectiveness.

Taking the project to the Community level

The importance of OT care for individuals and families following the diagnosis of a rare disease such as ATTRv will be highlighted at the end of this project. This research aims to reinforce the need for OT care in working groups dealing with rare diagnoses as part of a multidisciplinary team.

To date, this work has been replicated with satisfactory results in other populations. These include familial spastic paraparesis, fetal alcohol syndrome, tuberous sclerosis and butterfly skin.

In addition, there is a need for further research into the effectiveness of different OT approaches in different rare disease diagnoses so that they can be incorporated into routine clinical practice.

To eat, to drink, to go to work or to spend time with the family. These are all important activities in our lives. They have little value until they are lost.

Daily life is one of the most valuable things a human being can have. Daily life is one of the most valuable things a person can have, and it must be taken care of and respected.

3 Conclusion

General conclusions

This project was conducted to contribute to understanding the impact of occupational therapy interventions on people diagnosed with a rare disease such as ATTRv. General and specific conclusions have been drawn.

The psychosocial interventions offered to patients diagnosed with ATTRv require the activity and creativity of the occupational therapist. The integration of occupational therapists in health centres and multidisciplinary teams would be beneficial, feasible and desirable. This would provide tools to complete people's lives through meaningful occupations. This is a pioneering study mapping the OT services currently available to patients diagnosed with ATTRv.

Specific conclusions

Daily life

The diagnosis of ATTRv has a direct impact on daily life, not only for patients but also for carers and relatives.

This project has provided important considerations that can contribute to the best possible approach for patients with ATTRv. The use of an occupational needs assessment protocol, as well as the implementation of an occupational therapy programme, reinforces a paradigm shift in this discipline's approach to this disease.

Patients

This project not only provides important information for other disciplines. It is also a way of understanding the needs of patients after diagnosis. This is a simple reality that in many cases is underestimated.

OT Scales

Tools for assessing occupational therapy clients are severely lacking. By establishing a protocol of standardised scales and an interview, as described in this project, the necessary information to initiate an effective occupational intervention can be obtained.

Access

For patients, families and even professionals, OT is not yet an accessible service. Several causes directly affect the development of the profession and its recognition. The lack of a bibliography to support the work of occupational therapists was identified as one of the main issues.

Research on professional reasoning in OT is empirical in nature. Most of the studies are qualitative. Compared to other health professions, there is a paucity of studies that focus on information processing, reasoning modalities and distinguishing features of professional reasoning in OT.

Perception

The perception and role of OT in multidisciplinary teams is directly related to the different perceptions of professional reasoning. Occupational therapists do not have a sense of appreciation from patients or other professionals. This conclusion is reached for several reasons, which are explained throughout this thesis. However, it seems that when approaching people diagnosed with a rare disease such as ATTRv, few actions have been taken to promote occupational therapy as a necessary profession.

Improving theoretical knowledge in OT, as it requires specific learning of skills related to professional reasoning, is directly related to increasing the effectiveness of interventions.

Contributions

In the light of these findings, the inclusion of occupational therapists in multidisciplinary teams is advisable in order to

Encourage patients to engage in meaningful activities following diagnosis.

To recognise the patient's need for person-centred care.

To Understand how occupation works and how being diagnosed affects their day-to-day lives.

Promotion of the occupational therapy approach as a way of giving meaningful content to people's lives after diagnosis.

To carry out an intervention focused on the person and their daily life. To maintain autonomy and quality of life.

Promotion of knowledge of the profession.

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Annexes

1 Fifth Manuscript. Occupational therapy in the Iberian Peninsula. A qualitative study

Abstract Summary

Occupational therapy is an integral part of integrated care and support in the health care system. However, its application in the context of rare diseases has been the subject of little research. The aim of this final manuscript was the investigation of the OT situation and services for patients with a diagnosis of ATTRv in Portugal and Spain. A structured interview was used to collect opinions on the reality of OT from six Portuguese and four Spanish occupational therapists. Four interrelated dimensions were considered. The first was a general view of psychosocial resources in health, addressing questions related to interventions offered to patients; the second and third were professional views, asking questions about the role of OT in the Portuguese and Spanish psychosocial reality; and the fourth dimension was a social view, addressing questions about the role of OT in the Portuguese and Spanish psychosocial reality. It was found that there are currently no occupational therapists providing therapeutic interventions for patients with ATTRv in either Portugal or Spain.

Article

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Keywords

Transthyretin amyloidosis, occupation, occupational therapy, polyneuropathy, snowball sampling, and intervention.

Abstract

Occupational therapy is part of integrated care/support in health services. However, there has been little research on how it is applied to rare diseases. The aim of this study was to investigate the occupational therapy situation and services offered to patients diagnosed with hereditary transthyretin amyloidosis in Portugal and Spain. A structured interview was carried out by an occupational therapist with six Portuguese occupational therapists and four Spanish occupational therapists in order to collect opinions about the reality of occupational therapy. Four interrelated dimensions were addressed. The first was a general perspective on psychosocial resources in health care, addressing questions related to the interventions offered to patients; the second and third was an occupational perspective, asking questions about the role of occupational therapy in the Portuguese and Spanish psychosocial realities; and the fourth dimension was a social perspective that addressed questions about the role of occupational therapy in the Portuguese and Spanish psychosocial realities. All questions asked in the study ranged from a general approach to a more specific one and were focused on patients diagnosed with hereditary transthyretin amyloidosis. It was found that there were no occupational therapists currently providing therapeutic interventions to patients with hereditary transthyretin amyloidosis in either Portugal or Spain. The results of the non-existence of occupational therapy services show that this group of patients does not have

access to such a valid form of occupational intervention. This highlights the importance of developing and introducing the discipline of occupational therapy in a multidisciplinary approach, by creating potential strategies to promote its use.

Introduction

Hereditary transthyretin amyloidosis (ATTRv) has been defined as a heterogeneous, genetic and rare disease (Ando et al., 2013). This disease is caused by a mutation in the transthyretin gene. To date, more than 140 mutations in the TTR gene have been identified, with Val50Met reported to be the most common variant (Connors et al., 2003). ATTRv presents in many different forms, with significant variability in signs and symptoms between patients, and is therefore defined as a multisystemic disease (Reinés et al., 2014).

Amyloid fibrils, derived from the accumulation of unstable conformations of transthyretin, are the origin of amyloid deposition. These fibrils can be deposited in various organs, most commonly the heart, gastrointestinal tract, and kidneys, and are the cause of a variety of functional disorders (Luigetti et al., 2020). The Val50Met variant, the most common, is particularly prevalent in the two largest foci in Portugal and Spain. These are located in northern Portugal, Majorca, and Valverde del Camino (Spain), where ATTRv is defined as endemic (Munar-Qués et al., 2005).

Neuropathic symptoms directly related to peripheral sensory and motor damage are the main features of this disease (Conceição & De Carvalho, 2007). Symptoms are progressive and disabling throughout the four main disease stages (I, II, III, and IV), with severe damage in the final stage (Tornero Estébanez et al., 2007). Autonomic dysfunction and sensory impairment progress from the lower limbs to the upper limbs. Thus, autonomy and dependence in activities of daily living are directly affected as the dysfunction progresses (González-Moreno et al., 2021). In the final stage, individuals with ATTRv become extremely dependent and die approximately 10 to 13 years after the onset of symptoms without proper treatment (Gayà-Barroso et al., 2021a). Daily activity impairment that ATTRv patients and carers experience at all stages of the disease has been described as difficulties that affect personal autonomy, occupational balance, and daily living skills (Damy et al.,2022). According to this information, occupational therapy (OT), through the development of specific interventions for these patients, can be fundamental in reducing activity limitations, improving personal independence, promoting autonomy, and empowering patients to overcome disease-related barriers (Welsby et al., 2019). OT has been described as a health profession concerned with promoting health and well-being through daily activities and occupations (Lee et al., 2012). Therefore, the occupational therapist's role is to develop an intervention strategy in all situations related to daily living and occupational risks, whether physical,

mental, social, occupational, or environmental, to enable patients diagnosed with ATTRv to develop, regain, maintain and/or compensate daily abilities (Gayà-Barroso et al., 2021b).

In the field of health care, the term multidisciplinary team is used to describe the approach to treatment planning that involves different health care professionals, experts in different specialities. The idea was explored from the perspective of including occupational therapists in ATTRv teams and recognising this discipline among patients and professionals (Legg et al., 2017). For this reason, different evidence, useful for future studies, will be shown in this pioneering study of the reality of OT between two countries such as Portugal and Spain.

The aim of this study is to analyse the reality of OT with an Iberian vision between Spain and Portugal, in order to characterise the reality of this discipline in the two countries, using the tree analysis system in order to define the main themes and subthemes.

Methods

This is an exploratory and qualitative study. It sought to understand the experiences of occupational therapists from an Iberian perspective. The qualitative approach provides a heterogeneous and comprehensive perspective of occupational therapists' experiences and how they view the current circumstances of the profession.

Snowball sampling was used as a qualitative research method. It was used to contact occupational therapists in Portugal and Spain. This system is described as a sampling method in which an interviewee informs the researcher of at least one other interviewee, in this case occupational therapist. The interviewee then provides the name of at least one other interviewee, and so on, until the sample grows like a rolling snowball (Kirchherr & Charles, 2018). This study has aimed to explore the situation of OT with an Iberian vision, from general to specific areas, focusing on the OT services offered to patients with ATTRv. This whole process allowed us to focus on the themes that emerged. However, it is recognised that the researcher's interpretations are fundamental, as they are what give meaning to the narratives.

Participants

Occupational therapists in Portugal and Spain were interviewed in a structured way. There were six occupational therapists in Portugal and four occupational therapists in Spain. Of the Portuguese occupational therapists, one was working as a teacher. Two were professionals who had completed their studies and the rest were working as occupational therapists. All Spanish occupational therapists worked at hospitals or clinics in geriatrics, paediatrics, or physical rehabilitation.

Procedure

Participants were recruited through an ATTRv support institution: Centro de Genética Preditiva e Preventiva (Porto), located in an endemic region in northern Portugal. This procedure included inclusion criteria, namely, that participants had to be occupational therapists, aged 18 years or older, and resident in Portugal and Spain.

In addition, an information sheet was prepared containing the objectives and important data about the study. This sheet was given and/or read to the participants when they made contact with the occupational therapist in charge of the study. Participants were then contacted by telephone or video call by the occupational therapist. An appointment was made for the interview.

Between October and April 2022-2023, all interviews were conducted by two authors (AG, MP). The interviews were audio-recorded and transcribed with the participants' consent. All interviews, which lasted on average about 40-50 minutes, were conducted by the occupational therapist. Questions were asked about occupational services in Portugal and Spain, specifically about (a) general questions, (b) psychosocial services in the Portuguese and Spanish health systems, (d) occupational approach, (e) role and benefits of occupational therapy in psychosocial interventions, (f) access and referrals to occupational therapy.

Data Analysis

In each interview, information and emerging subthemes were noted. Subsequently, in order to make a preliminary mapping of the emerging subthemes, each interview was reread. More specifically, each interview was reread and analysed inductively. This process involved identifying, comparing, and coding subthemes. Sub-themes were identified after carrying out this procedure. The AG and MP coded a preliminary mapping of the subthemes. Finally, they were grouped into different categories. This resulted in themes. Finally, with the aim of understanding the experience in a holistic way, a narrative prototype was created in which the themes and subthemes were related by means of the selective coding technique.

Ethical Considerations

Ethical approval was granted by the Ethics Committee of the Balearic Islands and the Research Commission of Hospital Universitario Son Llàtzer (Decision number: IB 4587/21 PI). Participants in the interview were informed that the study was confidential, anonymous, and voluntary. Informed consent was collected verbally due to the virtual nature of the interviews.

Results

The results indicated four themes and respective subtopics that are interrelated in Figure 1.

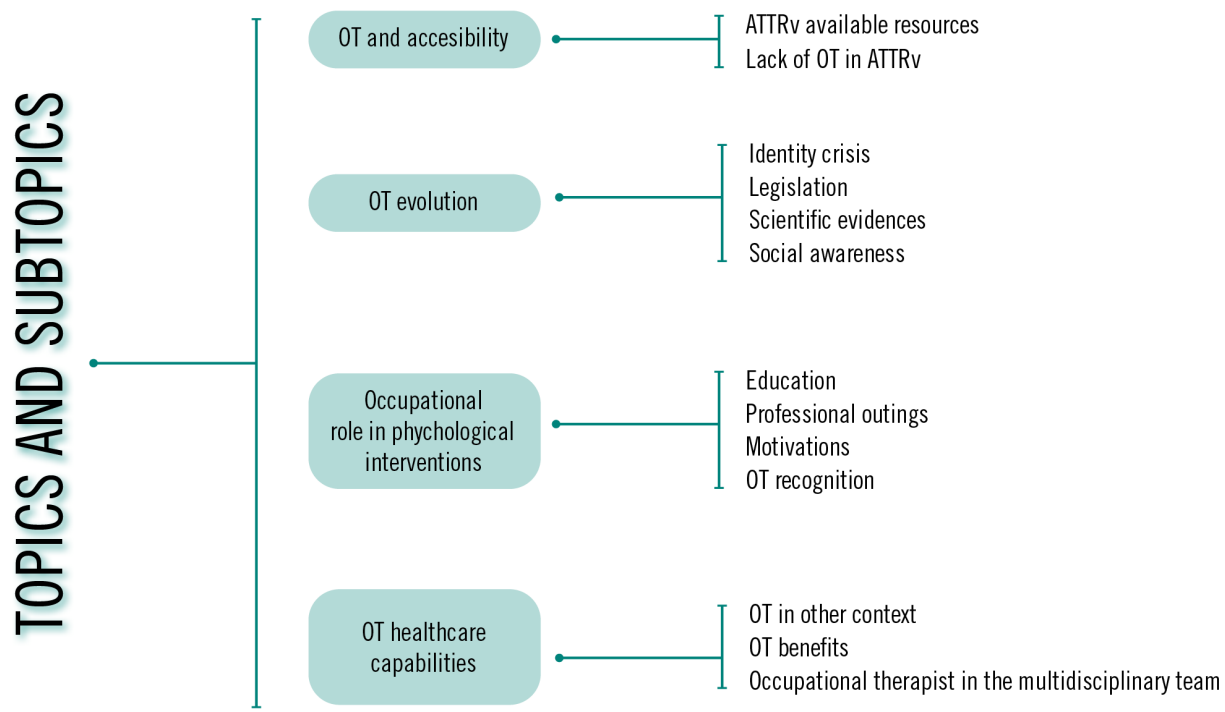

Figure 1. General themes and subthemes of the study

Topic 1: OT and accessibility

According to the main theme of the study related to the psychosocial and occupational services offered to ATTRv patients, all participants reported that they were no longer aware of any psychosocial or occupational therapy service for ATTRv or had no data on the disease.

> I work in the clinical field, in rehabilitation. But it is true that I have 10 years of experience working with patients with amyloidosis. I have been working with this profile for many years, but at the moment there are not many cases. Now the patients would be cases with a longer follow-up because of their dependency situation. But not because of the diagnosis. (Portuguese Occupational Therapist).
>
> If we consider rheumatoid arthritis as a rare disease, I have experience. But I have no experience or knowledge of ATTRv". (Spanish Occupational Therapist).

> I don't know, I have never worked with rare diseases. (Portuguese and Spanish occupational therapist).

Given this reality, occupational therapists also refer to multidisciplinary teams, as well as the availability of resources and interventions from a general perspective. Most participants reported that their experiences with psychosocial services were with four different professions: psychologists, social workers, speech and language therapists, and other occupational therapists. This experience shows the presence of different professionals in the general health services working together with the occupational therapist. However, this reality does not exist in all institutions, according to some participants. in some cases, multidisciplinary teams that include occupational therapists do not exist.

> As a psychosocial: OT, social worker, speech therapy and psychology, depending on the institution ... in the clinical services, in the health sector, there is usually also physiotherapy, nursing, neuropsychology ... often. It is important to work as a team to achieve the objectives". (Spanish Occupational Therapist).

> Social work and OT sometimes, but not always. In the field of geriatrics, they are resources that are widely used because of the characteristics of the patients and their families" (Spanish occupational therapist).

> Basically, I work in the Continuous Care Unit of Santa Casa and it is specifically a physical rehabilitation unit for all those patients who have no other support at home and who need to be hospitalised to be able to carry out an intervention in different disciplines such as: physiotherapy, occupational therapy, psychology, social work, etc.. (Portuguese Occupational Therapist)

On the other hand, it is important to emphasise the idea expressed by some occupational therapists: Individuals need to use private resources in order to get the right approach. Otherwise, only a few sessions are received and they are far apart in time.

> In my opinion, occupational therapists in Portugal are very poorly paid and poorly recognised. This is my vision. In order to achieve the goals we want to achieve, in many cases the services are not sufficient and people have to pay for private sessions. (Portuguese Occupational Therapist).

> Generally, in psychology and OT sporadic sessions are offered, so if a person wants the service, they usually do it themselves (privately), which is the only way to have sessions with effective continuity. (Spanish occupational therapist).

Topic 2: OT evolution

According to the interviewees, ignorance about the profession is linked to various causes, which were discussed during the interview. The answers focused on the lack of a clear concept of the role of the occupational therapist within the psychosocial field.

> I think that the first thing to emphasise and the first thing to do is to identify what we are doing in an institution. If we have a theoretical basis for what we are doing and show our knowledge and our own information as therapists, we will achieve the final goal. (Spanish occupational therapist)

> There is a great lack of knowledge about the role of occupational therapists, both on the part of patients and other health professionals, and therefore referral is not always made if they don't know us. .... However, the paradigm is changing. At the moment there is more demand for these services, especially in paediatrics". (Portuguese occupational therapist)

> No, few people in Spain have knowledge of this discipline. In the Spanish health system, the services are still unknown. There is a long way to go. I think that therapy is the eternal unknown in the health sector. (Spanish occupational therapist).

The occupational therapists interviewed mentioned a lack of resources in addition to a lack of knowledge and resources to meet occupational needs. This lack of resources is linked to poorer interventions and outcomes. The perception of the profession is also affected by this situation.

> It's a shame, but in general they are not fully addressed, there is a lack of both human and material resources, so in the end both patients and family solve their needs in the best way. (Portuguese occupational therapist.)

> All interventions depend on this: the rehabilitation strategies and possible adaptations that the therapist will implement. Unfortunately, there is a lack of resources. This affects the general approach. (Spanish occupational therapist.)

Following this idea, a Portuguese occupational therapist mentioned a law that makes the presence of an occupational therapist mandatory in services provided in the context of general health care. As a result, in both countries the OT was perceived as a health care profession that was in touch with everyday life. The occupational therapist would be responsible for working on this aspect if the activities of daily living could not be carried out independently and functionally. However, some occupational therapists reported that this was not the case in all institutions. In some cases, there was no OT in multidisciplinary teams.

> No, it's still hard to get to know Occupational Therapy in Portugal. One important thing is that from 2022 there is a law obliging all national health services to have an occupational therapist. This is a essential step forward for us, because in theory we have to be there. But it

> is true that as a profession we are moving very slowly. I believe that in the near future we will start to be known. Professionals will start to refer and in general they will know who the occupational therapist is. (Portuguese occupational therapist)

> The therapist works with people's daily life, from the simplest activities like getting dressed to the most complex ones like looking for a job. Whenever there is a change in daily life, the OT should be there, although at the moment this is not the case. (Portuguese occupational therapist)

> They are beginning to realise the importance of this figure in the field of early intervention. Although I think that interventions in general do not have an impact. The vast majority do not know us. So, from my point of view, although it is essential, the role is now relative. (Spanish occupational therapist)

In addition, through their previous experiences, the participants also mentioned the significant lack of approach to the daily life of people with any kind of illness. For example:

> There is a very important lack of intervention in daily life, we do not really know how these people work at home, what is their routine, how they live. We cannot know in 10 minutes of intervention what is being done now. This is not compatible with an effective intervention... (Portuguese occupational therapist)

> Unfortunately, in the hospital health service you intervene mainly in BADL. The IADL and the leisure and free time activities depend on the ability of the occupational therapist, who tries to create an environment that is as real as possible... Advanced activities require resources that you often do not have. Therefore, the OT focuses on talking to the family and advising them, but they cannot assess in situ, and then they are not reassessed by an OT, so these are areas that tend to be forgotten... because nobody asks them. (Spanish occupational therapist)

Regarding the impact of OT, those interviewed believe that the lack of references and studies that demonstrate the impact of the profession leads to low social awareness of the discipline.

> In my opinion, there is a lack of awareness in terms of lectures, seminars, and health fairs, so that both professionals and patients are aware of our role. (Portuguese occupational therapist)

> In my opinion, occupational therapy is in need of a lot of theoretical foundations that would open a door to our profession. And above all, to be present in team meetings that work with patients... we are still not there. (Spanish occupational therapist)

When participants were asked about working with activities of daily living, therapists were aware that all patients referred to their services needed OT. However, due to a lack of knowledge, both professional and civil, this service was often not accessed.

> Absolutely. Although I don't know if all patients with difficulties in their daily life reach the OT. (Portuguese occupational therapist)

> Yes, much. When patients come to the service, the OT is a necessity". (Spanish occupational therapist)

In relation to access to OT and referral, the study participants linked this subtheme to knowledge of the profession. Lack of knowledge determines access. However, in general, the occupational therapists commented: "Doctors are the main person who usually refers patients. In some cases, it can also be done by the school, in the case of children and by the social worker. They do not refer patients if they are not aware of the role of the occupational therapist.

> In my case, yes, and what I've experienced is that we've got the same role and the same importance within the team. When you work in an institution with therapists, the other professionals know us. But I think that in general, if they have not worked with us, they do not know of our existence. (Portuguese occupational therapist)

> In the case of children, I know that it's different, because in the early stimulation centres, the figure of the occupational therapist is increasingly recognised and appreciated for its impact on the children. They are usually referred by doctors, although whenever a professional sees that there is a change in the activities of daily living, he could refer, but it doesn't happen yet. (Spanish occupational therapist)

Topic 3: Lack of training

Occupational therapists commented that OT was not the first option to study. This is relevant information for an approach to the current situation of OT in both countries. To better understand the situation of this profession, the lack of knowledge and motivation about the profession, the lack of references, and professional excursions should have been taken into account.

> First of all, I have to explain that occupational therapy was not my first choice. I did not consider becoming an occupational therapist. My first choice was physiotherapy, you could say that occupational therapy came into my life by chance". (Portuguese occupational therapist).

> Patients are not aware of your role at first. I think this is due to a mix of elements, lack of research, lack of training in universities, lack of protocols, lack of social awareness... for example, occupational therapy was not my first choice of study, in fact I didn't know what it was when I started. (Spanish occupational therapist)

> It would be nice to have more references from professionals who could explain professional opportunities, possibilities for the future. It's important to be motivated and to achieve professional goals. However, this is not the reality for us. (Portuguese occupational therapist)

Topic 4: OT healthcare capabilities

Despite the reality described throughout the interview, all occupational therapists agree that OT plays a fundamental role in the therapeutic approach. After all, we all have a 'daily life' and living is a significant occupation.

> There are many, I think that our occupations are the axis of our day, if this is changed.... It is the occupational therapist who has the tools. Even if it is not yet seen or defined that way". (Portuguese occupational therapist). "I would include it, either as continuous treatment or as advice and training of personnel in basic activities, instrumental and advanced activities... adaptations, energy saving techniques... in general as a health educator". "Remember" that people go walking, celebrate birthdays with relatives, go to the cinema, have sex.... (Spanish occupational therapist)

> The presence of the OT would be very beneficial if each professional did not see the participation of the other as an "intrusion" and if there was a clear division of tasks, because this is often lost sight of... (Spanish occupational therapist)

In contrast, occupational therapists reported that OT involvement in multidisciplinary teams was important and should be seen as a necessity in both countries.

> As I said: "Many professionals forget that people with a diagnosis also have a life". (Spanish occupational therapist)

> Sure, it would help with all the aspects that were commented during the interview. I do not know the disease, but therapy plays a fundamental role when there is a change in daily life. (Spanish occupational therapist)

> As far as occupational therapy is concerned, we work effectively with people's daily lives. We do a lot of things and we give a lot of meaning to the things of daily life. Therapists are already bored of talking about just doing meaningless things, jumping, making things. We work with people every day. We should not be ashamed to work with a patient on grooming or dressing, or teaching them to brush their teeth. We are just one more point of view. (Portuguese occupational therapist)

Finally, it is worth noting that OT has been developed in a number of fields, such as paediatrics. An idea that could be extrapolated to rare diseases.

> However, it is a changing paradigm and there is currently a greater demand for these services, especially in paediatrics. Maybe in the future it will happen in other areas too. (Portuguese occupational therapist)

> I really believe that in Spain, geriatrics and paediatrics are the two areas where occupational therapy is most appreciated. I hope that it will gradually be applied in more areas of daily life. (Spanish occupational therapist)

Discussion

To the best of our knowledge, this is a pioneering study in the mapping of OT services currently on offer in the Iberian Peninsula. Specifically, in Portugal and Spain, this study explored the reality of the OT services available to patients diagnosed with ATTRv. In order to understand the current situation of OT in Portugal and Spain, the study started by conducting an interview. The interview had four thematic focuses. However, no occupational therapists were found who were currently working with ATTRv or rare diseases. In addition, after diagnosis, patients do not have access to interventions to keep their daily life and autonomy as functional as possible. In spite of this reality, some articles on OT and ATTRv have been found. They describe the access to discipline and the identification of professional needs in order to initiate approach strategies (Gayà-Barroso et al., 2022b). For the health care system, this situation is a challenge. The occupational therapist could be a key professional in multidisciplinary teams in circumstances related to occupational imbalance (Quinn & Hynes, 2021).

However, the exposed reality of ATTRv contrasts with the benefits already identified in interventions in other rare diseases such as Charcot-Marie-Tooth (CMT), as well as recent articles published on the impact of OT in ATTRv patients (Matyjasik-Liggett et al., 2013). Although the limitations of the study, as well as the small samples, need to be taken into account, the data obtained are very informative and positive as soon as the occupational therapist intervenes in the occupational risks of people diagnosed with a rare disease.

All therapists agreed that OT plays an important role in the holistic approach to patients. This is due to the fact that the concept of everyday life has not been fully addressed from the psychosocial dimension (Santos et al., 2021). Themes and subthemes common to both countries were identified in the results of this research. Occupational therapists mentioned the presence of the professional profile in the multidisciplinary team in specific areas such as geriatrics or early intervention. Many other areas were not mentioned. In short, both Portuguese and Spanish occupational therapists reported a significant lack of theoretical foundations of the profession, which leads to a devaluation of the role of the therapist. This reality is in addition to the fact that some of the therapists in this study report that OT was not their first choice of study. They entered this discipline as a second or third option, a fact that may influence the motivation and involvement of professionals in their work. Lack of identity and motivation and low impact on the health system may be related to this finding. A reflection of the current Iberian reality are professionals who do not know how to define their functions or do not believe in the potential of their work.

On the other hand, the areas of work are very limited. Therapists in both countries commented that working in geriatrics, paediatrics, and physical rehabilitation is where OT is most valued. In

these contexts, the occupational therapist works with a large number of patients in short intervention times. This makes it more difficult to provide care and achieve goals, a situation that leads to professional fatigue and burnout. Another reality related to this discipline is that, in some cases, occupational therapists are aware that the resources are not sufficient for the patient, who has to pay for private sessions in order to achieve the objectives. These are a number of reasons that guide the development of the profession.

Occupational therapists' views on referrals and general access were the last important finding. Patients are referred to the service in institutions where the OT profile exists. However, occupational therapists feel that OT is not part of the multidisciplinary approach. In many cases, patients who should receive occupational interventions are not referred to them. This may be due to the education and training of healthcare professionals.

A complex approach, even when recognising the importance of roles, routines, and habits, following the diagnosis of a rare disease such as ATTRv. It should be emphasised that in studies investigating the reasons why patients do not have access to OT, most respondents reported that professionals had never recommended OT (Bolt et al., 2019) Our results suggest that ATTRv patients do not have access to OT services, which, as shown, could benefit them. Although OT is provided in primary care in several European countries, this is not the reality in Spain or Portugal, where OT is still underused for patients requiring occupational interventions (Cavaglion et al., 2023). Several factors may explain this, such as the relatively low number of OT professionals in Spain and Portugal, the unfamiliarity of patients, the lack of referrers, the lack of knowledge, as well as the lack of theoretical basis of OT to support the interventions (Welsby et al., 2019). In addition, other studies show that in rare diseases such as CMT, participants usually report that their occupational therapist is unfamiliar with the symptoms and difficulties of daily life, and the professional is not able to provide an effective intervention (Matyjasik-Liggett et al., 2013). However, due to the small sample size and global nature of the results, our findings are inconclusive. Further research is therefore clearly needed, including research on the outcomes of specific OT interventions used in the general health care system and in the ATTRv population. OT promotes occupation as a protective factor for health, participation and well-being, focusing on habits and routines that tend to achieve maximum occupational balance (Gayà-Barroso et al., 2022).

Limitations

The fact that the sample was recruited through professional contacts is an important limitation of this study. Consequently, the findings can only be extrapolated to the specific contexts of the occupational therapists included in the study. The inclusion of other occupational therapists working in hospitals and other types of settings would be important in future studies.

The small size of the cohort was another important limitation. Unfortunately, there was a low level of participation in this study. Allies such as professional associations, hospitals, and other institutions involved in rare disease diagnosis would be needed.

Conclusion

This study has shown that OT is rarely used among ATTRv patients in Portugal and Spain, mainly due to lack of awareness. Our results seem to indicate that there are no OT specialists for ATTRv. Not even occupational therapists specialise in rare diseases. Occupational therapists in both countries perceive a significant lack of theoretical framework and recognition among patients and other professionals. This, together with the lack of research, means that occupational therapy is still an unknown and undervalued profession from an Iberian point of view.

Data

Institutional Review Board Statement

Ethical approval was granted by the Ethics Committee of the Balearic Islands and the Research Commission of Hospital Universitario Son Llàtzer. Decision number: IB 4587/21 PI.

Conflict of Interest

The authors have no competing interests to declare.

Authors' contributions

AGB MP led the study conceptualization. AGB and MP recruited occupational therapists. AGB and MP performed the interviews and designed the intervention. AGB and MP analysed the data and wrote the first draft of the paper. AGB, MP, ECB, ILL, JGM, ARR, TRV, and MG contributed to the discussion on the protocol development and provided critical feedback on drafts of the manuscript.

Acknowledgements

The authors wish to thank all professionals who participated in this study.

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2 Etical approval

DICTAMEN DEL COMITÉ DE ÉTICA DE LA INVESTIGACIÓN DE LAS ISLAS BALEARES

Esther Cardo Jalón, Secretaria Técnica del Comité de Ética de la Investigación de las Islas Baleares,

CERTIFICO:

Que este Comité, en la sesión celebrada el día 30 de junio (nº 08/2021), evaluó los aspectos metodológicos, éticos y legales del proyecto de investigación:

Código CEI: IB 4587/21 PI

Código de protocolo: TTR-TO

Título: ESTUDIO SOBRE LA ACCESIBILIDAD A LA TERAPIA OCUPACIONAL EN PACIENTES DIAGNOSTICADOS DE AMILOIDOSIS HEREDITARIA POR TRANSTIRETINA

Promotor: Inés Losada López / Eugenia Cisneros Barroso

Versión y fecha de protocolo: 1.0 de 2-6-2021
Versión y fecha de HIP-CI: 1.0 de 2-6-2021

Que se acuerda emitir DICTAMEN FAVORABLE para la realización de este proyecto de investigación en los siguientes centros:

Investigador principal	Centro	Servicio
Inés Losada López y Eugenia Cisneros Barroso	Hospital Universitario Son Llàtzer	Medicina Interna

Que el CEI-IB, tanto en su composición como en los PNT, cumple con las normas de Buena Práctica Clínica (CHMP/ICH/135/95) y con la legislación vigente que regula su funcionamiento, teniendo en cuenta que en el caso de que algún miembro participe en el estudio o declare algún conflicto de interés no habrá participado en la evaluación ni en el dictamen.

Que, a la fecha de aprobación de este estudio, la composición del CEI-IB es la siguiente:

Presidente:	Francisco Campoamor Landín	Farmacología clínica
Vice-presidenta:	Cristina Villena Portella	Biología
Secretaria:	Esther Cardo Jalón	Neurología pediátrica
Vocales:	Alberto Alonso Fernández	Neumología

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Enrique Colás Ruiz	Cirugía general y del aparato digestivo
Cristina Corbillo Colom	Representante de los pacientes
Magdalena Esteva Cantó	Medicina preventiva y salud pública
Francisco Javier Fanjul Losa	Medicina interna
Miquel Fiol Sala	Medicina intensiva
José Fuster Salvá	Oncología médica
Rocío Gómez Juanes	Psiquiatría
Gemma Jiménez Guerra	Microbiología
Raquel Marsé Fabregat	Oncología médica
Antònia Mas Cantallops	Derecho
Cristina Moreno Mulet	Enfermería
Antònia Obrador Hevia	Biología
Jaume Orfila Timoner	Medicina interna
Carmen Pata Iglesias	Farmacia atención primaria
José Ignacio Ramírez Manent	Medicina familiar y comunitaria
Pere Riutord Sbert	Estomatología
Montserrat Rodríguez Delgado	Química
Juan Rodríguez García	Medicina preventiva y salud pública
Mónica Sanz Muñoz	Farmacia hospitalaria
Llorenç Socías Crespí	Medicina intensiva
Javier Ureña Morales	Derecho

Palma, 7 de julio de 2021

comite d'ètica de la investigació
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3 Informed agreement

TÍTULO DEL ESTUDIO:

CÓDIGO DEL PROMOTOR:

PROMOTOR:

INVESTIGADOR PRINCIPAL *(médico del estudio, si excepcionalmente se trata de otro profesional se debe indicar. Incluir nombre, servicio y teléfono)*:

CENTRO:

Yo, *(nombre y apellidos)*,

- ◯ He leído la hoja de información que se me ha entregado.
- ◯ He podido hacer preguntas sobre el estudio.
- ◯ He recibido suficiente información sobre el estudio.
- ◯ He hablado con: *(nombre del investigador).*
- ◯ Comprendo que mi participación es voluntaria.
- ◯ Comprendo que puedo retirarme del estudio (*si se recogen muestras, añadir:*) y solicitar la destrucción de mi muestra, siempre y cuando no haya sido anonimizada:

Cuando quiera.

Sin tener que dar explicaciones.

Sin que esto repercuta en mis cuidados médicos.

- ◯ Comprendo que, si decido retirarme del estudio, los resultados obtenidos hasta ese momento podrán seguir siendo utilizados (*si se recogen muestras, añadir*:) pero que no se realizarán nuevos análisis de mi muestra, siempre y cuando no haya sido anonimizada.

En el caso de que los resultados de la investigación proporcionen datos que me puedan interesar a mí o a mis familiares: (*indicar una de las casillas*)

- ◯ Quiero ser informado.

- ◯ No quiero ser informado, pero acepto que mi médico contacte con mis familiares si dichos resultados les pueden afectar.
- ◯ Comprendo que tengo los derechos de acceso, rectificación, supresión, oposición, limitación del tratamiento de datos, incluso a trasladar mis datos a un tercero autorizado (portabilidad), de acuerdo con lo dispuesto en la Ley Orgánica 3/2018, de 5 de diciembre, de protección de datos de carácter personal y garantía de los derechos digitales.
- ◯ Presto libremente mi conformidad para participar en el estudio y doy mi consentimiento para el acceso y utilización de mis datos en las condiciones detalladas en la hoja de información al paciente.

(*En caso de solicitar muestras para el proyecto, y tener previsto almacenarlas para otros estudios, deberá incluir el siguiente párrafo, ofreciendo al participante una o varias de las siguientes opciones, de acuerdo con la información facilitada en la hoja de información*)

Al término de la investigación mi muestra podrá ser:

- ◯ Destruida
- ◯ Anonimizada
- ◯ Incorporada en una colección cuyo responsable es el investigador *(indicar nombre completo del investigador responsable de la colección),* que se encuentra en *(indicar lugar)*, para continuar siendo utilizada en el estudio de *(indicar la línea de investigación)*.
- ◯ Almacenada en el biobanco *(indicar cuál)* para poder ser utilizada en otras investigaciones, posiblemente no relacionadas con el estudio inicial para el cual consintió.

Firma del paciente: Firma del investigador:

Nombre: Nombre:

Fecha: Fecha:

4 Patient information

INSTRUCCIONES

La Hoja de Información al Paciente y el documento de Consentimiento Informado no deben entenderse únicamente como requisitos procedimentales para llevar a cabo la investigación pretendida, sino como parte imprescindible del proceso, por lo que resulta primordial configurar un documento útil y entendible para los participantes, que abarque los principales aspectos de la investigación y muy especialmente aquellos que puedan afectarle.

Aunque se debe entregar al sujeto la información por escrito, en todos los casos es obligado explicársela de forma oral.

En este modelo aparecen en letra normal los aspectos que quedan fijos para todos los estudios, *en cursiva los aspectos variables dependiendo de las características del estudio, pero que deben cumplimentarse obligatoriamente*, y *en rojo aparecen instrucciones y aspectos que son variables dependiendo de las características del estudio pero que podrían no tener que aparecer en determinados casos (intentar no superar MÁXIMO 10 PÁGINAS y tamaño de la fuente 12)*

HOJA DE INFORMACIÓN AL PACIENTE PARA LA REALIZACIÓN DE ENSAYOS CLÍNICOS, PROYECTOS DE INVESTIGACIÓN Y ESTUDIOS GENÉTICOS ***(eliminar el que no proceda)***

(Versión y fecha de la HIP)

TÍTULO DEL ESTUDIO:

CÓDIGO DEL PROTOCOLO:

PROMOTOR:

INVESTIGADOR PRINCIPAL: (médico del estudio, o en su caso otro profesional)

CARGO, UNIDAD, CENTRO:

TELÉFONO:

CORREO ELECTRÓNICO:

INTRODUCCIÓN

Nos dirigimos a usted para informarle sobre un estudio en el que se le invita a participar. El estudio ha sido aprobado por el Comité de Ética de la Investigación de las Islas Baleares, de acuerdo a la legislación vigente, y se lleva a cabo con respeto a los principios enunciados en la declaración del Helsinki y a las normas de buena práctica clínica.

Nuestra intención es tan solo que usted reciba la información correcta y suficiente para que pueda evaluar y juzgar si quiere o no participar en este estudio. Para ello lea esta hoja informativa con atención y nosotros le aclararemos las dudas que le puedan surgir después de la explicación. Además, puede consultar con las personas que considere oportuno. Si tiene alguna duda diríjase a *(investigador principal.)*.

DESCRIPCIÓN GENERAL

(Este apartado no debería exceder de una página. La información contenida debe ser relevante, expresada en términos claros y comprensibles para los sujetos, no se deben usar tecnicismos)

Se debe explicar en qué consiste y qué objetivo persigue el estudio al cuál se le invita a participar, así como la metodología (ej. cuando el estudio sea doble ciego, se debe decir que "ni el médico ni el paciente sabrán cuál es el tratamiento que va a recibir"), cuánto tiempo dura, los inconvenientes y riesgos derivados del estudio (número de visitas y pruebas complementarias a las que se someterá, reflejando claramente cuales se van a hacer de forma extraordinaria a su asistencia habitual por su participación en el estudio).

Debe explicarse el procedimiento de "asignación al azar" (cuando proceda), y los criterios generales por el que el sujeto es seleccionado.

Se debe indicar el número total de sujetos que se van a incluir.

TRATAMIENTOS ALTERNATIVOS (si procede)

Se debe explicar brevemente, si existen, las otras alternativas terapéuticas eficaces en la actualidad para el tratamiento de su enfermedad, que podría recibir en caso de no participar en el estudio (incluso se debe explicar que podría recibir los mismos fármacos que se le ofrecen en el estudio como sería el caso por ejemplo de EC fase IV).

Añadir que el médico del estudio le dará más información si lo desea.

OTRA INFORMACIÓN RELEVANTE *(si procede)*

Se debe indicar si el fármaco está o no está autorizado/comercializado y sus indicaciones.

Se debe explicar brevemente la experiencia con el fármaco.

Se deben indicar las responsabilidades del participante con relación a las visitas, actividades del estudio, y que tiene que notificar cualquier acontecimiento adverso al médico/investigador del estudio

Cualquier nueva información referente a los fármacos utilizados en el estudio y que pueda afectar a su disposición para participar en el estudio, que se descubra durante su participación, le será comunicada por su médico lo antes posible.

Si usted decide retirar el consentimiento para participar en este estudio, ningún dato nuevo será añadido a la base de datos y puede exigir la destrucción de todas las muestras identificables previamente retenidas para evitar la realización de nuevos análisis, si bien los responsables del estudio podrán seguir utilizando la información recogida sobre usted hasta ese momento, a no ser que usted se oponga expresamente.

También debe saber que usted puede ser retirado del estudio en caso de que los responsables del estudio lo consideren oportuno, ya sea por motivos de seguridad, por cualquier acontecimiento adverso que se produzca por la medicación en estudio o porque consideren que no está cumpliendo con los procedimientos establecidos. En cualquiera de los casos, usted recibirá una explicación adecuada del motivo que ha ocasionado su retirada del estudio.

Si a usted se le retira del estudio por alguno de los motivos expresados, su médico le rescribirá un tratamiento adecuado a su enfermedad.

Al firmar la hoja de consentimiento adjunta, se compromete a cumplir con los procedimientos del estudio que se le han expuesto.

ESTUDIOS EN MENORES DE EDAD y/o PERSONAS CON LA CAPACIDAD MODIFICADA PARA DAR SU CONSENTIMIENTO *(si procede)*

Hasta los 16 años debe existir también el consentimiento de los titulares de la patria potestad o de la tutela.

Le informamos que, al tratarse de la participación de su hijo, que tiene 12 años cumplidos, se le va a entregar a él mismo una hoja de información y consentimiento informado adaptados a su

capacidad de entendimiento y deberá firmarlos (si el menor es mayor de 12 años). Asimismo, se garantiza al menor el acceso a la información relativa a la utilización de su muestra cuando éste alcance la mayoría de edad y tendrá también el derecho a la revocación del consentimiento y, en el caso de no ejercerlo, este documento se considerará vigente (si hay recogida de muestras). Además, de acuerdo a la legislación vigente, los responsables del estudio han puesto en conocimiento del Ministerio Fiscal que se van a incluir menores de edad en el mismo si es necesario.

El documento de consentimiento informado de los padres será válido siempre que está firmado por uno de ellos con el consentimiento expreso o tácito del otro, este hecho debe quedar suficientemente documentado.

Asimismo, el menor siempre debe ser escuchado incluso si tiene menos de 12 años, si tiene madurez suficiente.

BENEFICIOS Y RIESGOS DERIVADOS DE SU PARTICIPACIÓN EN EL ESTUDIO

Se deben comentar los beneficios esperados para el sujeto y la sociedad y añadir, obligatoriamente, que es posible que no obtenga ningún beneficio para su salud por participar en este estudio. No se puede inducir a la participación.

Se debe explicar brevemente la experiencia previa referente al estudio, los posibles acontecimientos adversos (en términos que el sujeto pueda comprender y de forma concisa, si se conocen porcentajes se deben incluir) (si procede).

Incluir cualquier tipo de riesgo debido a las pruebas que se realizan como consecuencia del estudio.

La realización gratuita de las pruebas y el seguimiento estricto no deben ser incluidas como beneficio de la participación al paciente.

Explicar los posibles inconvenientes vinculados con la donación y obtención de la muestra, incluida la posibilidad de ser contactado con posterioridad con el fin de recabar nuevos datos y/o obtener otras muestras (si procede).

En el caso de participación de mujeres en edad fértil debería existir un apartado específico sobre el embarazo o lactancia (si procede).

Se deben incluir los riesgos conocidos del fármaco sobre el feto o advertir que se desconocen.

Se debe indicar que, en el caso de producirse un embarazo, se solicitará la recogida de datos y los datos de salud del recién nacido. Se debe informar del periodo en el que se recoge la información y que se garantizará el cumplimiento del Reglamento General de Protección de Datos.

Si se considera necesario evitar el embarazo, debe precisarse qué método(s) anticonceptivos se recomiendan. Se aconseja que, si se tiene que utilizar un método anticonceptivo doble, el coste asociado al segundo método anticonceptivo será reembolsado por el promotor.

Efectos sobre la pareja del sujeto participante, si está en edad fértil, se debe especificar la obligación de informar a su pareja sobre cualquier tipo de contraindicación.

En el caso de embarazo de la pareja de un participante masculino, esta información se debe solicitar a través de un consentimiento específico.

SEGURO

Cuando proceda: la realización de una investigación que comporte un procedimiento invasivo en seres humanos exigirá el concierto de una póliza de seguros que cubra los posibles daños y perjuicios que puedan derivarse para la persona participante en el estudio.

En el caso de ensayos clínicos de baja intervención (art.2j del RD 1090/2015, por el que se regulan los ensayos clínicos con medicamentos) y siempre que la póliza del centro de investigación cubra la investigación clínica no será necesario otro tipo de seguro.

Se recomienda que si el paciente tiene otros seguros, debe ponerse en contacto con la compañía para determinar si su participación en un estudio clínico puede afectar a su póliza.

El promotor del estudio ha concertado una póliza de seguros (número de póliza) con la compañía (nombre compañía) que se ajusta a la legislación vigente y que cubre todos los perjuicios que pudieran producirse en relación con su participación en el estudio.

CONFIDENCIALIDAD

Responsable del tratamiento: (investigador y datos de contacto)

Finalidad de la recogida de datos: (investigación biomedica)

Destinatarios de la información: (indicar si habrá o no cesión a terceros)

Plazo máximo de conservación de los datos: (indicar el plazo máximo de conservación de los datos)

El tratamiento, la comunicación y la cesión de los datos de carácter personal de todos los sujetos participantes se ajustará a lo dispuesto en la Ley Orgánica 3/2018, de 5 de diciembre, de protección de datos de carácter personal y garantía de los derechos digitales.

De acuerdo a lo que establece la legislación mencionada, usted puede ejercer los derechos de acceso, rectificación, supresión, oposición, limitación del tratamiento de datos, incluso a trasladar sus datos a un tercero autorizado (portabilidad), para lo cual deberá dirigirse al investigador principal responsable de tratamiento en las siguientes direcciones (Se debe incluir dirección física y electrónica)

Sus datos serán tratados informáticamente y se incorporarán a un sistema automatizado de datos de carácter personal que cumple con todas las medidas de seguridad de acceso restringido al objetivo descrito en este documento.

Para garantizar la confidencialidad de la información obtenida, (a continuación se dan varios supuestos posibles, elija el que corresponda a su estudio)

1) anonimización: Sus datos y la muestra recogida serán sometidos a un proceso de anonimización, con lo que será irreversiblemente disociada de sus datos personales, de tal manera que será imposible identificar a quién pertenecen posteriormente.

2) codificación o pseudoanonimización: Sus datos y la muestra estarán identificados mediante un código y solo el médico del estudio y colaboradores podrán relacionar dichos datos con usted y con su historia clínica. Por lo tanto, su identidad no será revelada a persona alguna salvo en caso de urgencia médica, requerimiento de la Administración sanitaria o requerimiento legal.

Sólo se transmitirán a terceros y a otros países los datos imprescindibles necesarios para poder realizar el estudio, y que en ningún caso contendrán información que le pueda identificar directamente, como nombre y apellidos, iniciales, dirección, nº de la Seguridad Social, etc. En el caso de que se produzca esta cesión, será para los mismos fines del estudio descrito y garantizando la confidencialidad como mínimo con el nivel de protección de la legislación vigente en nuestro país.

El acceso a su información personal quedará restringido al médico del estudio/colaboradores, autoridades sanitarias, al Comité de Ética de la Investigación de las Islas Baleares y personal

autorizado, cuando lo precisen para comprobar los datos y procedimientos del estudio, pero siempre manteniendo la confidencialidad de los mismos de acuerdo a la legislación vigente.

(Si se trata de una investigación del Servicio de Salud,) Igualmente se le informa que usted podrá realizar cualquier consulta sobre este tratamiento ante la Delegación de Protección de Datos del Servicio de Salud de las Islas Baleares la cual tiene su sede en la calle de la Reina Esclarmunda, 9, de Palma (Islas Baleares) y su correo electrónico de contacto es dpd@ibsalut.es.

(Si no se trata de una investigación del Servicio de Salud) Igualmente se le informa que usted podrá realizar cualquier consulta sobre este tratamiento ante la Delegación de Protección de Datos de organismo promotor de la investigación, la cual tiene su sede en la calle dirección postal, de localidad y su correo electrónico de contacto es *correoelectronico@correo.com*.

En cualquier caso, usted puede dirigirse a la Agencia Española de Protección de Datos para cualquier reclamación derivada del tratamiento de sus datos personales.

COMPENSACIÓN ECONÓMICA

Su participación en el estudio no le supondrá ningún gasto y le serán reintegrados los gastos extraordinarios (por ejemplo: comidas y traslados) (*en los casos en los que se haya previsto por las características del estudio compensar a los pacientes por el tiempo dedicado al estudio o por las molestias que se le ocasionen).*

Su médico (indicar si recibirá o no una compensación económica por su participación en este estudio) y ha declarado (si existe o no) conflicto de intereses.

PARTICIPACIÓN VOLUNTARIA

Este apartado se redactará según el modo de protección de los datos de los individuos y el modo de tratamiento de las muestras.

En el supuesto de datos (y muestras si procede) anónimos o anonimizados se redactará como sigue:

Debe saber que su participación en este estudio es voluntaria y que puede decidir no participar en él, sin que ello tenga ninguna influencia en la relación con su médico o el tratamiento que debe usted recibir y sin dar ningún tipo de explicación.

En el supuesto de datos (y muestras si procede) codificados o pseudoanonimizados se redactará como sigue:

Debe saber que su participación en este estudio es voluntaria y que puede decidir no participar o cambiar su decisión y retirar el consentimiento en cualquier momento, sin dar ningún tipo de explicación, así como solicitar la destrucción de la muestra *(si hay recogida de muestras),* sin que por ello se altere la relación con su médico o el tratamiento que debe usted recibir.

Si usted decide revocar su consentimiento, no se recogerán nuevos datos, ni se realizarán nuevos análisis de la muestra, pero esta revocación no afectará a las investigaciones realizadas hasta el momento.

RECOGIDA DE MUESTRAS BIOLÓGICAS *(si procede)*

Explicar para qué finalidad concreta se requiere su muestra, dónde, cuándo y cómo se obtendrán, el procedimiento de recogida de muestra, lugar de realización del análisis y destino de la muestra al término de la investigación.

Los posibles riesgos derivados del procedimiento realizado para la obtención de estas muestras quedan cubiertos por el seguro del estudio.

La muestra será codificada y tratada confidencialmente durante la duración de este estudio, mediante un código que sólo el investigador y personal de su equipo podrá vincular con usted para preservar su identidad.

En caso de que sea necesario algún dato o muestra adicional, su médico contactará con usted para solicitar de nuevo su colaboración. Se le informará de los motivos y se le solicitará nuevamente el consentimiento, si es necesario.

No percibirá ningún beneficio económico por la donación de las muestras y la cesión de los datos aportados, ni tendrá derecho sobre posibles beneficios comerciales de los descubrimientos que se puedan conseguir como resultado de la investigación efectuada.

Las muestras se analizarán en el laboratorio *(nombre laboratorio)* y serán almacenadas durante *(número de años)* años, en previsión de que fuera necesario algún análisis adicional relacionado con los objetivos del estudio. Durante este proceso el responsable de las muestras será el promotor del estudio.

En caso de conservación de las muestras para usos futuros, se almacenarán en *(nombre repositorio)* durante *(número de años)* años.

(Sólo si está previsto conservar las muestras más allá del estudio/proyecto se deberá incluir el siguiente apartado; si no está previsto, se debe informar que al término del estudio/proyecto se destruirá la muestra y sus derivados):

Al término de la investigación, su muestra podrá ser *(puede ofrecerle al paciente una o varias de las siguientes opciones. El paciente debe dar su consentimiento expreso mediante casillas de verificación en el consentimiento informado):*

- Destruida.
- Anonimizada (es decir, se destruirá completamente el vínculo que relaciona dicha muestra con usted, con lo que ni el investigador ni ninguna otra persona del equipo será capaz de identificar de nuevo a quién pertenece su muestra).
- Incorporada en una colección cuyo responsable es el investigador *(indicar nombre completo del investigador responsable de la colección)* que se encuentra en (*indicar dónde*), para que continúe siendo utilizada en el estudio de *(indicar la línea de investigación).*
- Almacenada en el biobanco (*indicar cuál*) para poder ser utilizada en otras investigaciones, posiblemente no relacionadas con el estudio inicial para el cuál consintió. Desde el biobanco se podrán ceder las muestras para proyectos autorizados, posiblemente también en el extranjero, con un dictamen favorable previo del comité científico y del comité de ética del biobanco. Usted podrá dirigirse al biobanco para recabar información sobre los proyectos en los que se hayan utilizado sus muestras.

ANÁLISIS GENÉTICOS *(si procede)*

Las muestras se utilizarán para realizar análisis genéticos para *(detallar finalidad, ej. para poder identificar genes o marcadores genéticos de los que sea portador y que puedan predisponer al desarrollo de una enfermedad específica o que puedan condicionar su respuesta a un tratamiento concreto…).* Tendrán acceso a los resultados de los análisis (detallar quién).

En caso de obtenerse descubrimientos inesperados transcendentes sobre su salud o la de sus familiares, podrá serle comunicado si así lo ha manifestado en el apartado correspondiente al final de este documento. En el caso de no querer ser informado y la información sea necesaria para evitar un grave perjuicio para la salud de sus familiares biológicos, se podrá informar a los afectados o a su representante legalmente autorizado, previa consulta al Comité de Ética Asistencial del centro.

Si usted desea conocer los resultados, su médico se los suministrará o le dirigirá hacia donde puedan darle consejo genético tanto a usted como a sus familiares.

AGRADECIMIENTO

Sea cual sea su decisión, tanto el promotor como el equipo investigador quieren agradecer su tiempo y atención. Usted está contribuyendo al mejor conocimiento y cuidado de su enfermedad, lo que en el futuro puede beneficiar a multitud de personas.

5 ATTRv patients' questionnaire

Datos sociodemográficos

- Ud. es:
  - Paciente con AhTTR
  - Portador de una mutación del gen AhTTR
  - Familiar de un paciente con AhTTR

- ¿Presenta Vd. una mutación AhTTR con polineuropatía?
  - Sí
  - No

- ¿Cuál es el estadío FAP de su enfermedad?
  - Portador asintomático
  - Estadio I: no precisa ayuda para la deambulación
  - Estadio II: precisa ayuda para deambulación
  - Estadío III: silla de ruedas/ encamado

- ¿Cuánto tiempo ha pasado desde que le diagnosticaron amiloidosis AhTTR?
  - No aplicable
  - Menos de 2 años
  - 2-5 años
  - 5-10 años
  - Más de 10 años

- Indique su sexo
  - Mujer
  - Hombre

- Indique su edad
  - 18-30 años
  - 30-40 años

- 40-50 años
- Más de 50 años

- Indique el estado civil
  - Soltero
  - Casado o con pareja de hecho
  - Divorciado
  - Viudo

- ¿Cuántos hijos tiene?
  - 0
  - 1
  - 2
  - 3
  - Más de 3

- ¿Cuál es su nivel educativo?
  - Sin estudios
  - Educación primaria o Secundaria
  - Formación profesional
  - Formación universitaria o superior

- ¿Cuál es su situación laboral?
  - Empleado
  - Desempleado
  - En baja laboral
  - Jubilado
  - Estudiante

Antecedentes familiares de AhTTR

- Enfermó alguno de sus pades de AhTTR
  - No
  - Madre
  - Padre
  - Ambos

- ¿Falleció alguno de sus padres a causa de AhTTR?
  - No
  - Madre
  - Padre
  - Ambos

- (Si aplica) ¿Qué edad tenía su madre/padre cuando enfermó de AhTTR?
  - 18-30 años
  - 30-40 años
  - Más de 40 años

- (Si aplica) ¿Qué edad tenía su madre/padre cuando falleció por AhTTR?
  - 18-30 años
  - 30-40 años
  - 40-50 años
  - Más de 50 años

- Aparte de sus padres, tiene algún familiar diagnosticado de AhTTR?
  - 0
  - 1
  - 2
  - Más de 2

- (Si aplica) ¿Qué edad tenía Vd. cuando su madre/padre enfermó de AhTTR?
  - Menos de 18 años

- 18-25 años
- Más de 25 años

- (Si aplica) ¿Qué edad tenía Vd. cuando su madre/padre falleció por AhTTR?
  - Menos de 18 años
  - 20-25 años
  - Más de 25 años

- ¿Ha cuidado Vd. de algún familiar enfermo de AhTTR?
  - No
  - Madre
  - Padre
  - Otro familiar

Influencia del conocimiento/contacto con la AhTTR en su propio diagnóstico:

- ¿Hubo algún factor que le ayudara a entender su propio diagnóstico de AhTTR?
  - Ninguno
  - La enfermedad de mis padres
  - La enfermedad de otros familiares
  - Otros (detallar)

- ¿Cuál es su principal fuente de información sobre la enfermedad?
  - Asociación de pacientes
  - Mi médico
  - Familiares con la enfermedad
  - Internet
  - Otros

Diagnóstico, consejo genético y tiempo hasta la obtención de los resultados:

- ¿Tuvo Ud. acceso al consejo genético sobre AhTTR?
  - Sí
  - No

- (Si aplica) ¿Qué motivó su solicitud de consejo genético sobre AhTTR?
  - La enfermedad de mis padres
  - La enfermedad de mis parientes
  - La solicitud de mi médico
  - Mi propia solicitud

- ¿Tuvo dificultad para acceder a las pruebas genéticas?
  - Sí
  - No

- (Si aplica) ¿Qué barreras se encontró para acceder a las pruebas genéticas?

- ¿Tuvo miedo por conocer el resultado de las pruebas genéticas?
  - Sí
  - No

- ¿Cuánto tiempo tardó en conocer los resultados de las pruebas genéticas desde que se las hizo?
  - Menos de 6 meses
  - 6 meses- 1 año
  - Más de 1 año

Impacto de la AhTTR en la vida del paciente

- ¿Ha influido su diagnóstico de AhTTR en sus planes de estudios/laborales?
  - Sí
  - No

- En caso afirmativo, concrete de qué manera

- ¿Ha influido su diagnóstico de AhTTR en sus planes de formar una familia?
  - Sí
  - No

- En caso afirmativo, concrete de qué manera:

- ¿Se ha visto obligado a cambiar de vivienda debido a su AhTTR?
  - Sí
  - No

- ¿Ha necesitado contratar a un cuidador debido a su enfermedad?
  - Sí
  - No

- ¿Ha influido la enfermedad en sus ingresos?
  - Sí
  - No

- ¿Ha condicionado la enfermedad su círculo de amistades?
  - Sí
  - No

- ¿Cuenta con algún tipo de ayuda estatal debido a su enfermedad?
  - Sí
  - No

- En caso afirmativo, ¿cuál?

- ¿Pertenece Ud. a alguna asociación relacionada con su enfermedad?
  - Sí
  - No

- En caso afirmativo, ¿cuál?

Impacto psicológico o emocional tras el diagnóstico de la AhTTR

- ¿Ha tenido algún problema psicológico/psiquiátrico asociado a su diagnóstico de AhTTR?
  - Sí
  - No

- En caso afirmativo, ¿Cuáles?
  - Depresión
  - Ansiedad
  - Otros (concrete)

- (Si aplica) ¿Cuándo se han presentado estos problemas?
  - En el momento del diagnóstico
  - El año siguiente al diagnóstico
  - Varios años después del diagnóstico
  - Son problemas permanentes desde la aparición de los primeros síntomas

- (Si aplica) ¿Ha requerido ayuda de algún profesional para tartar estos problemas?
  - Sí
  - No

- En caso afirmativo, ¿qué tipo de profesional le ha ayudado?
  - Psicólogo
  - Psiquiatra
  - Otros (concrete)

- (Si aplica) ¿Ha requerido alguna medicación para tratar estos problemas?
  - Sí
  - No

- En caso afirmativo, ¿qué tipo de medicación?

Área de mejora

Por favor, califique del 1 al 10 su grado de satisfacción con los siguientes aspectos (1= nada satisfecho, 10= muy satisfecho).

- Atención recibida en lo relativo a su diagnóstico

  1 2 3 4 5 6 7 8 9 10

- Atención recibida en lo relativo a su enfermedad

  1 2 3 4 5 6 7 8 9 10

- ¿Qué piensa que podría mejorarse en la atención recibida en cuanto a su diagnóstico?
  - Atención médica
  - Atención psicológica
  - Aspectos sociales
  - Otros (concrete)

- En caso afirmativo, ¿qué tipo de mejoras relativas al diagnóstico de la AhTTR le parecen necesarias?

- ¿Qué piensa que podría mejorarse en la atención recibida en cuanto a su enfermedad?
  - Atención médica
  - Atención psicológica
  - Aspectos sociales
  - Otros

- En caso afirmativo, ¿qué tipo de mejoras relativas a la AhTTR le parecen necesarias?

Cuidadores

Datos sociodemográficos

- ¿Cuál es su relación con el paciente diagnosticado de amiloidosisAhTTR al que Ud. cuida?
  - Padre o madre
  - Hijo
  - Cónyuge
  - Pariente (especificar)

- ¿Cuánto tiempo ha pasado desde que diagnosticaron amyloidosis AhTTR al paciente?
  - Menos de 2 años
  - 2-5 años
  - 5-10 años
  - Más de 10 años

- ¿Cuál es el estadío FAP de la AhTTR del paciente?
  - Portador asintomático
  - Estadio I: no precisa ayuda para la deambulación
  - Estadio II: precisa ayuda para la deambulación
  - Estadio III: silla de ruedas/encamado

- ¿Cuántas horas diarias dedica al cuidado de este paciente?
  - Menos de 2
  - 2 - 5 horas
  - 5 - 10 horas
  - Más de 10 horas

- Indique su sexo
  - Mujer
  - Hombre

- Indique su edad
  - 18 - 30 años
  - 30 - 40 años
  - 40 - 50 años
  - Más de 50 años

- Indique su estado civil
  - Soltero
  - Casado o con pareja de hecho
  - Separado
  - Divorciado
  - Viudo

- ¿Cuántos hijos tiene?
  - 0
  - 1
  - 2
  - 3
  - Más de 3

- ¿Cuál es su nivel educativo?
  - Sin estudios
  - Educación primaria o secundaria
  - Formación profesional
  - Formación universitaria o superior

- ¿Cuál es su situación laboral?
  - Empleado
  - Desempleado
  - En baja laboral
  - Jubilado
  - Estudiante

- ¿Padece Ud. alguna enfermedad relevante?
  - Sí
  - No

- En caso afirmativo, ¿qué enfermedad?

Síntomas del paciente

Señale únicamente aquellos síntomas de AhTTR que ha experimentado el paciente al que Vd. cuida y califique del 1 al 10 la carga que este síntoma ha supuesto para Vd. como cuidador en el último año (1= nada problemático, 10= muy problemático).

- Dificultad para caminar, debilidad muscular

1 2 3 4 5 6 7 8 9 10

- Entumecimiento, calambres, hinchazón o dolor de piernas o pies

1 2 3 4 5 6 7 8 9 10

- Cansancio, mareos, pérdida de equilibrio

1 2 3 4 5 6 7 8 9 10

- Estreñimiento, diarrea, náuseas/vómitos, pérdida de peso/apetito

1 2 3 4 5 6 7 8 9 10

- Incontinencia fecal o urinaria

1 2 3 4 5 6 7 8 9 10

- Alteraciones de la vista

1 2 3 4 5 6 7 8 9 10

- Disfunción eréctil

1 2 3 4 5 6 7 8 9 10

- Ansiedad/depresión

1 2 3 4 5 6 7 8 9 10

- Insomnio

1 2 3 4 5 6 7 8 9 10

- Dificultad respiratoria, taquicardias

1 2 3 4 5 6 7 8 9 10

- Síndrome del túnel carpiano, pérdida de destreza manual

1 2 3 4 5 6 7 8 9 10

- Otros:

Tratamientos a los que ha recurrido el paciente

Señale únicamente aquellos tratamientos a los que ha recurrido el paciente al que Vd. cuida y el grado de satisfacción que ha obtenido el paciente con ellos en el último año (1= nada satisfecho, 10= muy satisfecho).

- Tratamiento del cansancio

1 2 3 4 5 6 7 8 9 10

- Tratamiento de los síntomas digestivos

1 2 3 4 5 6 7 8 9 10

- Tratamiento del dolor neuropático

1 2 3 4 5 6 7 8 9 10

- Tratamiento de la ansiedad/depresión

1 2 3 4 5 6 7 8 9 10

- Tratamiento del insomnio

1 2 3 4 5 6 7 8 9 10

- Tratamiento de la función cardíaca (p. ej., diuréticos)

1 2 3 4 5 6 7 8 9 10

- Tratamiento de la presión arterial

1 2 3 4 5 6 7 8 9 10

- Tratamiento de las alteraciones visuales

1 2 3 4 5 6 7 8 9 10

Impacto de la AhTTR en la vida del cuidador

Califique del 1 al 10 la carga que ha supuesto para Vd. en el último año el haber cuidado de un paciente con AhTTR en los aspectos que se indican (1= nada problemático, 10= muy problemático).

- Su salud física

1 2 3 4 5 6 7 8 9 10

- En caso afirmativo, concrete de qué manera

_____

- ¿Ha recibido medicación por este motivo? ¿Cuál?

_____

- Su bienestar emocional

1 2 3 4 5 6 7 8 9 10

- En caso afirmativo, concrete de qué manera

- ¿Ha recibido medicación por este motivo? ¿Cuál?

- Sus planes de estudios/laborales

1 2 3 4 5 6 7 8 9 10

- En caso afirmativo, concrete de qué manera

- Sus planes familiares/de ocio

1 2 3 4 5 6 7 8 9 10

- En caso afirmativo, concrete de qué manera

- Sus ingresos

1 2 3 4 5 6 7 8 9 10

- ¿Cuenta con algún tipo de ayuda estatal debido a tarea de cuidador?
  - Sí
  - -No

- En caso afirmativo, ¿cuál?

- ¿Pertenece Ud. a alguna asociación relacionada con la AhTTR?
  - Sí
  - No

- En caso afirmativo, ¿Cuál?

5.1 Accessibility questionnaire

Cuestionario Terapeuta Ocupacional

El presente cuestionario tiene como objetivo desarrollar una investigación relacionada con la percepción de los pacientes sobre los beneficios de la Terapia Ocupacional, en pacientes con diagnóstico de Amiloidosis hereditaria por Transtirretina.

* Indica que la pregunta es obligatoria

Consentimiento Informado y Hoja de Información al Paciente

Confirmo que: He leído la hoja de información sobre el estudio que figura a * continuación. He podido hacer preguntas acerca del estudio. He recibido información suficiente sobre el estudio.• He hablado con el médico del estudio.• Entiendo que mi participación es voluntaria.• Entiendo que puedo retirarme del estudio: Cuando quiera, sin tener que dar explicaciones y sin que dicha retirada afecte a la asistencia médica que reciba. Otorgo libremente mi consentimiento expreso para participar en el estudio.

5.2 Hoja de información al paciente para la realización de ensayos clínicos, proyectos de investigación y estudios genéticos

(Eliminar el que no proceda)

TÍTULO DEL ESTUDIO:

CÓDIGO DEL PROTOCOLO:

PROMOTOR:

INVESTIGADOR PRINCIPAL: (médico del estudio, o en su caso otro profesional)

CARGO, UNIDAD, CENTRO:

TELÉFONO:

CORREO ELECTRÓNICO:

INTRODUCCIÓN

Nos dirigimos a usted para informarle sobre un estudio en el que se le invita a participar. El estudio ha sido aprobado por el Comité de Ética de la Investigación de las Islas Baleares, de acuerdo a la legislación vigente, y se lleva a cabo con respeto a los principios enunciados en la declaración del Helsinki y a las normas de buena práctica clínica.

Nuestra intención es tan solo que usted reciba la información correcta y suficiente para que pueda evaluar y juzgar si quiere o no participar en este estudio. Para ello lea esta hoja informativa con atención y nosotros le aclararemos las dudas que le puedan surgir después de la explicación. Además, puede consultar con las personas que considere oportuno. Si tiene alguna duda diríjase a (investigador principal.).

DESCRIPCIÓN GENERAL

(Este apartado no debería exceder de una página. La información contenida debe ser relevante, expresada en términos claros y comprensibles para los sujetos, no se deben usar tecnicismos)

Se debe explicar en qué consiste y qué objetivo persigue el estudio al cuál se le invita a participar, así como la metodología (ej. cuando el estudio sea doble ciego, se debe decir que "ni el médico ni el paciente sabrán cuál es el tratamiento que va a recibir"), cuánto tiempo dura, los inconvenientes y riesgos derivados del estudio (número de visitas y pruebas complementarias a las que se someterá, reflejando claramente cuales se van a hacer de forma extraordinaria a su asistencia habitual por su participación en el estudio).

Debe explicarse el procedimiento de "asignación al azar" (cuando proceda), y los criterios generales por el que el sujeto es seleccionado.

Se debe indicar el número total de sujetos que se van a incluir.

BENEFICIOS Y RIESGOS DERIVADOS DE SU PARTICIPACIÓN EN EL ESTUDIO

Se deben comentar los beneficios esperados para el sujeto y la sociedad y añadir, obligatoriamente, que es posible que no obtenga ningún beneficio para su salud por participar en este estudio. No se puede inducir a la participación.

Se debe explicar brevemente la experiencia previa referente al estudio, los posibles acontecimientos adversos (en términos que el sujeto pueda comprender y de forma concisa, si se conocen porcentajes se deben incluir) (si procede).

Incluir cualquier tipo de riesgo debido a las pruebas que se realizan como consecuencia del estudio.

La realización gratuita de las pruebas y el seguimiento estricto no deben ser incluidas como beneficio de la participación al paciente.

Explicar los posibles inconvenientes vinculados con la donación y obtención de la muestra, incluida la posibilidad de ser contactado con posterioridad con el fin de recabar nuevos datos y/o obtener otras muestras (si procede).

En el caso de participación de mujeres en edad fértil debería existir un apartado específico sobre el embarazo o lactancia (si procede).

Se deben incluir los riesgos conocidos del fármaco sobre el feto o advertir que se desconocen.

Se debe indicar que, en el caso de producirse un embarazo, se solicitará la recogida de datos y los datos de salud del recién nacido. Se debe informar del periodo en el que se recoge la información y que se garantizará el cumplimiento del Reglamento General de Protección de Datos.

Si se considera necesario evitar el embarazo, debe precisarse qué método(s) anticonceptivos se recomiendan. Se aconseja que, si se tiene que utilizar un método anticonceptivo doble, el coste asociado al segundo método anticonceptivo será reembolsado por el promotor.

Efectos sobre la pareja del sujeto participante, si está en edad fértil, se debe especificar la obligación de informar a su pareja sobre cualquier tipo de contraindicación.

En el caso de embarazo de la pareja de un participante masculino, esta información se debe solicitar a través de un consentimiento específico.

SEGURO

Cuando proceda: la realización de una investigación que comporte un procedimiento invasivo en seres humanos exigirá el concierto de una póliza de seguros que cubra los posibles daños y perjuicios que puedan derivarse para la persona participante en el estudio.

En el caso de ensayos clínicos de baja intervención (art.2j del RD 1090/2015, por el que se regulan los ensayos clínicos con medicamentos) y siempre que la póliza del centro de investigación cubra la investigación clínica no será necesario otro tipo de seguro.

Se recomienda que si el paciente tiene otros seguros, debe ponerse en contacto con la compañía para determinar si su participación en un estudio clínico puede afectar a su póliza.

El promotor del estudio ha concertado una póliza de seguros (número de póliza) con la compañía (nombre compañía) que se ajusta a la legislación vigente y que cubre todos los perjuicios que pudieran producirse en relación con su participación en el estudio.

CONFIDENCIALIDAD

Responsable del tratamiento: (investigador y datos de contacto)

Finalidad de la recogida de datos: (investigación biomedica)

Destinatarios de la información: (indicar si habrá o no cesión a terceros)

Plazo máximo de conservación de los datos: (indicar el plazo máximo de conservación de los datos)

El tratamiento, la comunicación y la cesión de los datos de carácter personal de todos los sujetos participantes se ajustará a lo dispuesto en la Ley Orgánica 3/2018, de 5 de diciembre, de protección de datos de carácter personal y garantía de los derechos digitales.

De acuerdo a lo que establece la legislación mencionada, usted puede ejercer los derechos de acceso, rectificación, supresión, oposición, limitación del tratamiento de datos, incluso a trasladar sus datos a un tercero autorizado (portabilidad), para lo cual deberá dirigirse al investigador principal responsable de tratamiento en las siguientes direcciones (Se debe incluir dirección física y electrónica)

Sus datos serán tratados informáticamente y se incorporarán a un sistema automatizado de datos de carácter personal que cumple con todas las medidas de seguridad de acceso restringido al objetivo descrito en este documento.

Para garantizar la confidencialidad de la información obtenida, (a continuación se dan varios supuestos posibles, elija el que corresponda a su estudio)

1) anonimización: Sus datos y la muestra recogida serán sometidos a un proceso de anonimización, con lo que será irreversiblemente disociada de sus datos personales, de tal manera que será imposible identificar a quién pertenecen posteriormente.

2) codificación o pseudoanonimización: Sus datos y la muestra estarán identificados mediante un código y solo el médico del estudio y colaboradores podrán relacionar dichos datos con usted y con su historia clínica. Por lo tanto, su identidad no será revelada a persona alguna salvo en caso de urgencia médica, requerimiento de la Administración sanitaria o requerimiento legal.

Sólo se transmitirán a terceros y a otros países los datos imprescindibles necesarios para poder realizar el estudio, y que en ningún caso contendrán información que le pueda identificar directamente, como nombre y apellidos, iniciales, dirección, nº de la Seguridad Social, etc. En el caso de que se produzca esta cesión, será para los mismos fines del estudio descrito y garantizando la confidencialidad como mínimo con el nivel de protección de la legislación vigente en nuestro país.

El acceso a su información personal quedará restringido al médico del estudio/colaboradores, autoridades sanitarias, al Comité de Ética de la Investigación de las Islas Baleares y personal autorizado, cuando lo precisen para comprobar los datos y procedimientos del estudio, pero siempre manteniendo la confidencialidad de los mismos de acuerdo a la legislación vigente.

(Si se trata de una investigación del Servicio de Salud,) Igualmente se le informa que usted podrá realizar cualquier consulta sobre este tratamiento ante la Delegación de Protección de Datos del Servicio de Salud de las Islas Baleares la cual tiene su sede en la calle de la Reina Esclarmunda, 9, de Palma (Islas Baleares) y su correo electrónico de contacto es dpd@ibsalut.es.

(Si no se trata de una investigación del Servicio de Salud) Igualmente se le informa que usted podrá realizar cualquier consulta sobre este tratamiento ante la Delegación de Protección de Datos de organismo promotor de la investigación, la cual tiene su sede en la calle dirección postal, de localidad y su correo electrónico de contacto es correoelectronico@correo.com.

En cualquier caso, usted puede dirigirse a la Agencia Española de Protección de Datos para cualquier reclamación derivada del tratamiento de sus datos personales.

COMPENSACIÓN ECONÓMICA

Su participación en el estudio no le supondrá ningún gasto y le serán reintegrados los gastos extraordinarios (por ejemplo: comidas y traslados) (en los casos en los que se haya previsto por las características del estudio compensar a los pacientes por el tiempo dedicado al estudio o por las molestias que se le ocasionen).

Su médico (indicar si recibirá o no una compensación económica por su participación en este estudio) y ha declarado (si existe o no) conflicto de intereses.

PARTICIPACIÓN VOLUNTARIA

Este apartado se redactará según el modo de protección de los datos de los individuos y el modo de tratamiento de las muestras.

En el supuesto de datos (y muestras si procede) anónimos o anonimizados se redactará como sigue:

Debe saber que su participación en este estudio es voluntaria y que puede decidir no participar en él, sin que ello tenga ninguna influencia en la relación con su médico o el tratamiento que debe usted recibir y sin dar ningún tipo de explicación.

En el supuesto de datos (y muestras si procede) codificados o pseudoanonimizados se redactará como sigue:

Debe saber que su participación en este estudio es voluntaria y que puede decidir no participar o cambiar su decisión y retirar el consentimiento en cualquier momento, sin dar ningún tipo de explicación, así como solicitar la destrucción de la muestra (si hay recogida de muestras), sin que por ello se altere la relación con su médico o el tratamiento que debe usted recibir.

Si usted decide revocar su consentimiento, no se recogerán nuevos datos, ni se realizarán nuevos análisis de la muestra, pero esta revocación no afectará a las investigaciones realizadas hasta el momento.

AGRADECIMIENTO

Sea cual sea su decisión, tanto el promotor como el equipo investigador quieren agradecer su tiempo y atención. Usted está contribuyendo al mejor conocimiento y cuidado de su enfermedad, lo que en el futuro puede beneficiar a multitud de personas.

Consiento participar en el presente estudio.

Marca solo un óvalo.

Sí

No

Edad:

_____

Sexo

Marca solo un óvalo.

- ◯ Hombre
- ◯ Mujer
- ◯ Prefiero no decirlo

Edad de debut de la enfermedad

_____

Edad del diagnóstico

_____

Estadio de la enfermedad

Marca solo un óvalo.

- ◯ 0 No tengo síntomas
- ◯ I Alteraciones sensitivas pero capacidad de caminar conservada
- ◯ II Alteración para caminar pero sin precisar ayuda
- ◯ III A Camino con ayuda de un bastón
- ◯ IIIB Camino con ayuda de dos bastones
- ◯ IV No puedo caminar

Otro:

_____

¿Cuáles son sus principales síntomas? *

Selecciona todos los que correspondan.

- Dolor o pérdida de sensibilidad en pies/piernas
- Llagas en los pies
- Debilidad en las piernas
- Impotencia (disfunción eréctil, en varones)
- Dificultad con las habilidades motoras finas (abrocharse los botones de una camisa)
- Depresión
- Pérdida de sensibilidad (frío-calor)
- Estreñimiento o diarrea
- Falta de apetito o anorexia
- Pérdida de peso
- Sudoración anormal
- Dificultad en la visión
- Mareo o sensación de mareo
- Disminución de la tolerancia al ejercicio
- Dificultad respiratoria al estar tumbado
- Cansancio o malestar
- Palpitaciones
- Ansiedad

En relación a su desempeño ocupacional

Indique el nivel de dificultad para realizar las Actividades básicas de la vida diaria (aseo, vestirse, comer, movilidad funcional para realizar tareas básicas). (1= Totalmente en desacuerdo, 5= totalmente de acuerdo)

Marca solo un óvalo.

1	2	3	4	5
◯	◯	◯	◯	◯

Indique el nivel de dificultad para realizar las Actividades Instrumentales de la vida diaria (cocinar, lavar ropa, manejo medicación, compras, uso transporte público). (1= Totalmente en desacuerdo, 5= totalmente de acuerdo)

Marca solo un óvalo.

1 2 3 4 5

Indique el nivel de dificultad para realizar Actividades Avanzadas de la vida diaria (trabajo, viajes, participación social activa). (1= Totalmente en desacuerdo, 5= totalmente de acuerdo)

Marca solo un óvalo.

1 2 3 4 5

Indique brevemente sus ocupaciones significativas actuales (trabajo, ocio y tiempo libre)

Cuáles cree que son los posibles beneficios de la Terapia Ocupacional

¿Sabe qué función tiene el profesional en Terapia Ocupacional?

Marca solo un óvalo.

Sí

No

¿Ha utilizado los servicios de un Terapeuta Ocupacional?

Marca solo un óvalo.

Sí

No

Si ha utilizado los servicios de un Terapeuta Ocupacional, ¿Cuándo empezó a utilizarlos?

Marca solo un óvalo.

- Antes del diagnóstico de su enfermedad Al diagnóstico de su enfermedad
- Menos de dos años después del diagnóstico de su enfermedad
- Más de dos años después del diagnóstico de su enfermedad

Si ha utilizado los servicios de un Terapeuta Ocupacional ¿Qué tipo de intervenciones le han sido aplicadas?

Marca solo un óvalo.

- Programa de ejercicio físico y movilidad para mejorar la fuera, la motricidad, postura y equilibrio
- Programa de educación para adaptar sus tareas y simplificarlas
- Programa de educación para conservar su energía y poder desarrollar sus tareas diarias
- Recomendaciones para adaptar su hogar y facilitar su vida diaria y uso de productos de apoyo
- Programa de motivación ocupacional

Si ha utilizado los servicios de un Terapeuta Ocupacional ¿Cuántos años hace que utilizó los Servicios de un Terapeuta Ocupacional?

Marca solo un óvalo.

- Los estoy utilizando actualmente Los he utilizado en el último año
- Los utilicé hace más de un año
- Los utilicé hace más de tres años
- Los utilicé hace más de cinco años años

Si no ha utilizado nunca los servicios de un Terapeuta Ocupacional y conociendo que un Terapeuta Ocupacional es el encargado de: promover la máxima independencia y calidad de vida de la persona mediante la ocupación. Indique del 1 al 5 si está de acuerdo o no con las siguientes afirmaciones.

Considera que la inclusión de un Terapeuta Ocupacional como parte del equipo multidisciplinar, podría ser beneficioso para el manejo de su patología (1= Totalmente en desacuerdo, 5= totalmente de acuerdo)

Marca solo un óvalo.

1 2 3 4 5

Si ya ha utilizado los servicios de un Terapeuta Ocupacional, ¿considera que el haber recurrido a un Terapeuta Ocupacional fue beneficioso para usted? (1= Totalmente en desacuerdo, 5= totalmente de acuerdo)

Marca solo un óvalo.

1 2 3 4 5

5.2 Semi-structured Interview

FICHA DE DATOS

Nº:

Nombre del Paciente (Iniciales):

En caso de ser portador/a, dejar los huecos en blanco que considere pertinentes.

VARIABLE	RESPUESTA
EDAD	
EDAD DE DEBUT	
SEXO	
ESTADO CIVIL	
DESCRIPCIÓN UNIDAD FAMILIAR (Nº PERSONAS, PARENTESCO)	
ENFERMEDAD HEREDADA (PADRE/MADRE)	
NÚMERO DE FAMILAIRES AFECTADOS	
DESCENDENCIA	
ESTADIO PATOLOGÍA	
AÑO DIAGNÓSTICO	
TRATAMIENTO FARMACOLÓGICO:	
TRASPLANTE HEPÁTICO:	
INTERVENCIONES PREVIAS DE: PSICOLOGÍA/ T.O/ FISIOTERAPIA/NUTRICIÓN/PODOLOGÍA	
OPINIÓN PERSONAL RESPECTO A LAS INTERVENCIONES REFERIDAS	
NIVEL EDUCATIVO/ FORMACIÓN:	
SITUACIÓN LABORAL PREVIA:	
TIEMPO LIBRE/ HOBBIES PREVIOS:	
SITUACIÓN LABORAL ACTUAL:	
TIEMPO LIBRE/ HOBBIES ACTUALES:	
COMENTARIOS/ APORTACIONES:	

Explique cómo valora el impacto de los síntomas en su día a día:

5.3 Scales

Barthel Índex (Spanish version)

Parámetro	Situación del paciente	Puntuación
Total		
Comer	Totalmente independiente	10
	Necesita ayuda para cortar carne, el pan, etc.	5
	Dependiente	0
Lavarse	Independiente: entra y sale solo del baño	5
	Dependiente	0
Vestirse	Independiente: capaz de ponerse y de quitarse la ropa, abotonarse, atarse los zapatos	10
	Necesita ayuda	5
	Dependiente	0
Arreglarse	Independiente para lavarse la cara, las manos, peinarse, afeitarse, maquillarse, etc.	5
	Dependiente	0
Deposiciones (valórese la semana previa)	Continencia normal	10
	Ocasionalmente algún episodio de incontinencia, o necesita ayuda para administrarse supositorios o lavativas	5
	Incontinencia	0
Micción (valórese la semana previa)	Continencia normal, o es capaz de cuidarse de la sonda si tiene una puesta	10
	Un episodio diario como máximo de incontinencia, o necesita ayuda para cuidar de la sonda	5
	Incontinencia	0
Usar el retrete	Independiente para ir al cuarto de aseo, quitarse y ponerse la ropa…	10

Parámetro	Situación del paciente	Puntuación
Uso retrete	Necesita ayuda para ir al retrete, pero se limpia solo	5
	Dependiente	0
Trasladarse	Independiente para ir del sillón a la cama	15
	Mínima ayuda física o supervisión para hacerlo	10
	Necesita gran ayuda, pero es capaz de mantenerse sentado solo	5
	Dependiente	0
Deambular	Independiente, camina solo 50 metros	15
	Necesita ayuda física o supervisión para caminar 50 metros	10
	Independiente en silla de ruedas sin ayuda	5
	Dependiente	0
Escalones	Independiente para bajar y subir escaleras	10
	Necesita ayuda física o supervisión para hacerlo	5
	Dependiente	0

Máxima puntuación: 100 puntos (90 si va en silla de ruedas)

Resultado	Grado de dependencia
< 20	Total
20-35	Grave
40-55	Moderado
≥ 60	Leve
100	Independiente

Lawton and Brody Scale. Instrumental activities of daily life (Spanish version)

Item	Aspecto a evaluar	Puntos
Capacidad para usar el teléfono:	Utiliza el teléfono por iniciativa propia	1
	Es capaz de marcar bien algunos números familiares	1
	Es capaz de contestar al teléfono, pero no de marcar	1
	No es capaz de usar el teléfono	0
Hacer compras	Realiza todas las compras necesarias independientemente	1
	Realiza independientemente pequeñas compras	0
	Necesita ir acompañado para hacer cualquier compra	0
	Totalmente incapaz de comprar	0
Preparación de la comida	Organiza, prepara y sirve las comidas por sí solo adecuadamente	1
	Prepara adecuadamente las comidas si se le proporcionan los ingredientes	0
	Prepara, calienta y sirve las comidas, pero no sigue una dieta adecuada	0
	Necesita que le preparen y sirvan las comidas	0
Cuidado de la casa	Mantiene la casa solo o con ayuda ocasional (para trabajos pesados)	1
	Realiza tareas ligeras, como lavar los platos o hacer las camas	1
	Realiza tareas ligeras, pero no puede mantener un adecuado nivel de limpieza	1
	Necesita ayuda en todas las labores de la casa	1
	No participa en ninguna labor de la casa	0
Lavado de la ropa	Lava por sí solo toda su ropa	1
	Lava por sí solo pequeñas prendas	1
	Todo el lavado de ropa debe ser realizado por otro	0
Uso de medios de transporte	Viaja solo en transporte público o conduce su propio coche	1
	Es capaz de coger un taxi, pero no usa otro medio de transporte	1
	Viaja en transporte público cuando va acompañado por otra persona	1
	Sólo utiliza el taxi o el automóvil con ayuda de otros	0
	No viaja	0
Responsabilidad respecto a su medicación	Es capaz de tomar su medicación a la hora y con la dosis correcta	1
	Toma su medicación si la dosis le es preparada previamente	0
	No es capaz de administrarse su medicación	0
Manejo de sus asuntos económicos	Se encarga de sus asuntos económicos por sí solo	1
	Realiza las compras de cada día, pero necesita ayuda en las grandes compras, bancos	1
	Incapaz de manejar dinero	0

Norfolk questionnaire

Cuestionario sobre la calidad de vida (QOL-DN)

Parte I: Síntomas

¿Ha tenido alguno de los siguientes síntomas en las últimas 4 semanas? Marque todas las casillas que precise.

- Entumecimiento
- Pinchazos
- Calambres eléctricos
- Otras sensaciones no habituales
- Dolor superficial
- Dolor profundo
- Debilidad

Indique si le sucede en: pies, piernas, manos, brazos.

Parte II: Actividades cotidianas

Valore del 0 al 4 (donde 0 es no es un problema y 4 problema intenso)

- En las 4 últimas semanas, ¿el dolor le ha impedido dormir o le ha despertado por la noche?

  1 2 3 4

  ☐ ☐ ☐ ☐

- En las 4 últimas semanas, ¿le ha molestado el tacto de las sábanas, de la ropa o del calzado?

  1 2 3 4

  ☐ ☐ ☐ ☐

- En las 4 últimas semanas, ¿se ha quemado o autolesionado y no se ha dado cuenta?

  1 2 3 4

- En las 4 últimas semanas, ¿ha habido algún síntoma que le haya impedido llevar a cabo sus actividades cotidianas durante el día?

  1 2 3 4

- En las 4 últimas semanas, ¿ha tenido dificultad para hacer movimiento con los dedos como abotonarse la ropa, pasar las páginas de un libro o coger monedas de una mesa?

  1 2 3 4

- En las 4 últimas semanas, ¿se ha notado inestable al caminar?

  1 2 3 4

- En las 4 últimas semanas, ¿ha tenido problemas para incorporarse de una silla sin empujar con las manos?

  1 2 3 4

- En las 4 últimas semanas, ¿ha tenido problemas para bajar escaleras?

  1 2 3 4

- En las últimas 4 semanas ¿ha dejado de sentir los pies al caminar?

  1 2 3 4

- En las últimas 4 semanas, ¿Ha sido incapaz de distinguir el agua fría de la caliente con las manos?

  1 2 3 4

- En las últimas 4 semanas, ¿ha sido incapaz de distinguir el agua fría de la caliente con los pies?

  1 2 3 4

- En las últimas 4 semanas ¿ha tenido problemas de vómitos, sobre todo después de las comidas (pero no debidos a la gripe o a otras enfermedades)?

  1 2 3 4

- En las últimas 4 semanas ¿ha tenido problemas de diarrea o de pérdida del control intestinal?

  1 2 3 4

- En las últimas 4 semanas ¿ha tenido problemas de desvanecimiento o mareo estando de pie?

  1 2 3 4

Responda a estas preguntas del 0 al 4 (siendo 0 sin dificultad y 4 dificultad total).

En las 4 últimas semanas. ¿qué grado de dificultad ha encontrado para llevar a cabo las siguientes actividades?

	0	1	2	3	4
Bañarse o ducharse					
Vestirse					
Caminar					
Sentarse o levantarse del inodoro					
Uso de cubiertos					

Responda a estas preguntas con arreglo a la siguiente escala (0, 1, 2, 3, 4).

En las últimas 4 semanas, ¿ha tenido alguno de los siguientes problemas con su trabajo u otras actividades habituales a consecuencia de su salud física o emocional?

- Ha disminuido el tiempo dedicado al trabajo u otras actividades

  1 2 3 4

- Ha hecho menos de lo que le hubiera gustado

- 1 2 3 4

- Se ha visto limitado en su trabajo u actividades

  1 2 3 4

- Ha tenido dificultad para llevar a cabo el trabajo u otras actividades (esfuerzo adicional)

- 1 2 3 4

- Ha tenido dificultad para llevar a cabo el trabajo u otras actividades (esfuerzo adicional)

1 2 3 4

- En términos generales cómo definiría su salud:
  - Excelente
  - Muy buena
  - Buena
  - Regular
  - Mala

- Como definiría su salud comparada con la de hace tres meses:
  - Mucho mejor
  - Algo mejor
  - Igual
  - Algo peor
  - Mucho peor

SF – 36 Scale

Cuestionario de Salud SF-36 (versión 2)

Su Salud y Bienestar

Por favor conteste las siguientes preguntas. Algunas preguntas pueden parecerse a otras pero cada una es diferente.

Tómese el tiempo necesario para leer cada pregunta, y marque con una la casilla que mejor describa su respuesta.

Marque una sola respuesta:

- En general, usted diría que su salud es:
  - Excelente
  - Muy buena
  - Buena
  - Regular
  - Mala

- ¿Cómo diría que es su salud actual, comparada con la de hace un año?
  - Mucho mejor ahora que hace un año
  - Algo mejor ahora que hace un año
  - Más o menos igual que hace un año
  - Algo peor ahora que hace un año
  - Mucho peor ahora que hace un año

Las siguientes preguntas se refieren a actividades o cosas que usted podría hacer en un día normal

- Su salud actual, ¿le limita para hacer esfuerzos intensos, tales como correr, levantar objetos pesados, o participar en deportes agotadores?
  - Sí, me limita mucho
  - Sí, me limita un poco
  - No, no me limita nada

- Su salud actual, ¿le limita para hacer esfuerzos moderados, como mover una mesa, pasar la aspiradora, jugar a los bolos o caminar más de una hora?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - , no me limita nada

- Su salud actual, ¿le limita para coger o llevar la bolsa de la compra?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - No, no me limita nada

- Su salud actual, ¿le limita para subir varios pisos por la escalera?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - No, no me limita nada

- Su salud actual, ¿le limita para subir un solo piso por la escalera?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - No, no me limita nada

- Su salud actual, ¿le limita para agacharse o arrodillarse?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - No, no me limita nada

- Su salud actual, ¿le limita para caminar un kilómetro o más?
    - Sí, me limita mucho
    - Sí, me limita un poco
    - No, no me limita nada

- Su salud actual, ¿le limita para caminar varias manzanas (varios centenares de metros)?
    - Sí, me limita mucho
    - Sí, me limita un poco

- No, no me limita nada

- Su salud actual, ¿le limita para caminar una sola manzana (unos 100 metros)?
  - Sí, me limita mucho
  - Sí, me limita un poco
  - No, no me limita nada

- Su salud actual, ¿le limita para bañarse o vestirse por sí mismo?
  - Sí, me limita mucho
  - Sí, me limita un poco
  - No, no me limita nada

Las siguientes preguntas se refieren a problemas en su trabajo o en sus actividades diarias

- Durante las últimas 4 semanas, ¿tuvo que reducir el tiempo dedicado al trabajo o a sus actividades cotidianas a causa de su salud física?
  - Sí
  - No

- Durante las últimas 4 semanas, ¿hizo menos de lo que hubiera querido hacer, a causa de su salud física?
  - Sí
  - No

- Durante las últimas 4 semanas, ¿tuvo que dejar de hacer algunas tareas en su trabajo o en sus actividades cotidianas, a causa de su salud física?
  - Sí
  - No

- Durante las últimas 4 semanas, ¿tuvo dificultad para hacer su trabajo o sus actividades cotidianas (por ejemplo, le costó más de lo normal), a causa de su salud física?
  - Sí
  - No

- Durante las últimas 4 semanas, ¿tuvo que reducir el tiempo dedicado al trabajo o a sus actividades cotidianas a causa de algún problema emocional (como estar triste, deprimido, o nervioso)?
    - Sí
    - No

- Durante las últimas 4 semanas, ¿hizo menos de lo que hubiera querido hacer a causa de algún problema emocional (como estar triste, deprimido, o nervioso)?
    - Sí
    - No

- Durante las últimas 4 semanas, ¿no hizo su trabajo o sus actividades cotidianas tan cuidadosamente como de costumbre, a causa de algún problema emocional (como estar triste, deprimido, o nervioso)?
    - Sí
    - No

- Durante las últimas 4 semanas, ¿hasta qué punto su salud física o los problemas emocionales han dificultado sus actividades sociales habituales con la familia, los amigos, los vecinos u otras personas?
    - Nada
    - Un poco
    - Regular
    - Bastante
    - Mucho

- ¿Tuvo dolor en alguna parte del cuerpo durante las 4 últimas semanas?
    - No, ninguno
    - Sí, muy poco
    - Sí, un poco
    - Sí, moderado
    - Si, mucho
    - Sí, muchísimo

- Durante las últimas 4 semanas, ¿hasta qué punto el dolor le ha dificultado su trabajo habitual (¿incluido el trabajo fuera de casa y las tareas domésticas?
    - Nada
    - Un poco
    - Regular
    - Bastante
    - Mucho

Las siguientes preguntas se refieren a cómo se ha sentido y como le han ido las cosas durante las 4 últimas semanas. En cada pregunta, responda lo que se parezca más a cómo se ha sentido usted.

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió lleno de vitalidad?
    - Siempre
    - Casi siempre
    - Muchas veces
    - Algunas veces
    - Sólo alguna vez
    - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo estuvo muy nervioso?
    - Siempre
    - Casi siempre
    - Muchas veces
    - Algunas veces
    - Sólo alguna vez
    - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió tan bajo de moral que nada podía animarle?
    - Siempre
    - Casi siempre
    - Muchas veces
    - Algunas veces
    - Sólo alguna vez

- Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió calmado y tranquilo?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo tuvo mucha energía?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió desanimado y triste?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió agotado?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió feliz?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿Cuánto tiempo se sintió cansado?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

- Durante las 4 últimas semanas, ¿con qué recurrencia la salud física o los problemas emocionales le han dificultado sus actividades sociales (como visitar a amigos o familiares)?
  - Siempre
  - Casi siempre
  - Muchas veces
  - Algunas veces
  - Sólo alguna vez
  - Nunca

Por favor, diga si le parece cierta o falsa cada una de las siguientes frases

- Creo que me pongo enfermo más fácilmente que otras personas
  - Totalmente cierta
  - Bastante cierta
  - No lo sé
  - Bastante falsa
  - Totalmente falsa

- Estoy tan sano como cualquiera
  - Totalmente cierta
  - Bastante cierta
  - No lo sé
  - Bastante falsa
  - Totalmente falsa

- Creo que mi salud va a empeorar
  - Totalmente cierta
  - Bastante cierta
  - No lo sé
  - Bastante falsa
  - Totalmente falsa

- Mi salud es excelente
  - Totalmente cierta
  - Bastante cierta
  - No lo sé
  - Bastante falsa
  - Totalmente falsa

El cuestionario de salud SF-36 está compuesto por 36 ítems que pretenden recoger todos los aspectos relevantes para caracterizar la salud de un individuo. Con estas preguntas se trata de cubrir, al menos, 8 aspectos o dimensiones: Función Física, Rol Físico; Dolor Corporal; Salud General; Vitalidad; Función Social; Rol Emocionay Salud Mental. Para cada una de estas dimensiones se pueden computar escalas de puntuación, fácilmente interpretables, caracterizadas todas ellas por encontrarse ordenadas, de tal suerte que cuanto mayor es el valor obtenido mejor es el estado de salud.

Escala de Bienestar Mental de Warwick - Edimburgo (EBMWE)

A continuación, aparecen algunas afirmaciones sobre sentimientos y pensamientos. Por favor, señale la casilla que mejor describa cómo se ha sentido durante las últimas 2 semanas.

Nº	Afirmaciones	Nunca	Muy pocas veces	Algunas veces	A menudo	Siempre
1.	Me he sentido optimista respecto al futuro					
2.	Me he sentido útil (*)					
3.	Me he sentido relajado/a					
4.	He sentido interés por los demás (**)					
5.	He tenido energía de sobra para mis actividades					
6.	He enfrentado bien los problemas					
7.	He podido pensar con claridad					
8.	Me he sentido bien conmigo mismo/A					
9.	Me he sentido cercano/a los demás (**)					
10.	Me he sentido seguro/a (con confianza)					
11.	He sido capaz de tomar mis propias decisiones					
12.	Me he sentido querido/a valorado/a					
13.	Me he interesado por cosas nuevas					
14.	Me he sentido alegre					

(*) se refiere a todos los aspectos de la vida.

(**) por aquellas personas con las que comparto mis días.

5.5 Interview OT with Iberian visión

Ficha de Informação do Estudo de Investigação

Compreender a prática da Terapia Ocupacional em pacientes diagnosticados com ATTRv em Portugal: a abordagem de equipa multidisciplinar.

O meu nome é Aina Gayà Barroso, sou Terapeuta Ocupacional e estou actualmente a fazer um doutoramento juntamente com a Dra. Inés Losada, Dr. Juan González, Eugenia Cisneros e Adrián Rodríguez, do Hospital Universitário Son Llàtzer em Palma de Maiorca. O meu trabalho centra-se na investigação do papel da Terapia Ocupacional em doentes diagnosticados com Transthyretin Amiloidose Hereditária. Graças a uma bolsa Erasmus+ iniciei um estágio clínico no Centro de Genética Preditiva e Preventiva do IBMC, sob a coordenação de Milena Paneque Herrera onde pretendemos explorar a realidade ocupacional dos pacientes portugueses, a maior coorte de pacientes ATTRv a nível mundial.

De que se trata este estudo?

O objectivo deste estudo é investigar a utilização de serviços de Terapia Ocupacional em doentes com doença de amiloidose transtiretina hereditária numa região endémica do norte de Portugal, para melhor compreender se os serviços estão a ser prestados, os tipos de intervenções que estão a ser realizadas e o conhecimento sobre a Ocupação do ponto de vista dos profissionais. Como resultado, teremos uma visão geral da Península Ibérica, integrando os nossos estudos anteriores no foco endémico de Palma de Maiorca (Mallorca, Illes Baleares, Espanha).

O que me estão a pedir para fazer?

Este trabalho é um estudo qualitativo exploratório que procurou compreender o ponto de vista dos profissionais sobre os serviços ocupacionais e o papel da ocupação na vida quotidiana do paciente.

Se é um profissional de saúde experiente e lida com pacientes com Transthyretin Amiloidose Hereditária, valorizamos a sua ajuda neste estudo.

Gostaríamos de realizar uma entrevista semi-estruturada que incluísse a recolha de informações gerais e perguntas sobre o campo ocupacional. Dividimos a entrevista em cinco secções que incluem entre duas a cinco perguntas cada uma.

Se eu mudar de ideias sobre estar envolvido?

Mesmo se concordar em ajudar, e começar o questionário, pode mudar de ideias em qualquer fase sem dar um motivo, basta deixar o sítio web do inquérito. Ficaria muito grato por responder a quaisquer outras perguntas que possa ter sobre o estudo antes de decidir sobre o seu envolvimento. Se desejar fazer uma pergunta, por favor contacte-me por e-mail para: ainabarroso@gmail.co.

5.6 Entrevista Semi-estruturada Terapia Ocupacional (Portugal)

Inquéritos gerais:

Antes de mais, vamos fazer-lhe quatro perguntas sobre generalidades relevantes para este estudo.

Profissão:

Localização de trabalho:

Experiência com doentes ATTRv (anos):

Número médio de pacientes com ATTRv por semana:

Contexto dos cuidados de saúde

Em segundo lugar, gostaríamos de lhe fazer algumas perguntas sobre o contexto geral dos cuidados de saúde.

Que serviços psicossociais são actualmente oferecidos no sistema de saúde português?

Que serviços psicossociais são actualmente oferecidos na sua instituição?

Quais são as principais intervenções psicossociais oferecidas aos pacientes ATTRv?

Abordagem ocupacional:

A fim de obter informações sobre o papel do terapeuta ocupacional, colocar-lhe-emos cinco questões relacionadas com o tema.

No que diz respeito ao seu ponto de vista, o que faz o Terapeuta Ocupacional nas suas intervenções?

Que papel tem a prática profissional nos serviços de saúde oferecidos em Portugal?

E no departamento onde trabalha?

Qual é a sua perspectiva sobre o papel do terapeuta ocupacional com pacientes diagnosticados (ATTRv)?

Já teve oportunidade de trabalhar com um terapeuta ocupacional?

Se a resposta for sim, explique brevemente a experiência:

Recomendaria aos pacientes a utilização do serviço de terapia ocupacional? Em que situações o recomendaria?

O que pensa sobre os possíveis benefícios da Terapia Ocupacional?

Papel ocupacional em intervenções psicossociais:

(Tendo em conta que na Terapia Ocupacional se distinguem três tipos de actividades: Básica, Instrumental e Avançada):

De acordo com a sua experiência clínica, está consciente de que os pacientes com quem trabalha têm dificuldades nas suas actividades básicas da vida quotidiana (preparação, vestir-se, comer, mobilidade funcional para realizar tarefas básicas)?

De acordo com a sua prática clínica, está consciente de que os pacientes têm dificuldades em realizar actividades instrumentais da vida diária (cozinhar, lavar roupa, manusear medicamentos, fazer compras, utilizar transportes públicos)?

De acordo com a sua prática clínica, está consciente de que os pacientes têm dificuldades em realizar actividades avançadas da vida diária (trabalho, lazer e tempo livre, família)?

Como são abordados estes aspectos mencionados nas perguntas anteriores (instalações, ferramentas, apoio extra?

Acesso ao terapeuta ocupacional:

Finalmente, é essencial saber como os pacientes chegam ao terapeuta ocupacional, por isso vamos fazer-lhe algumas perguntas em relação ao assunto.

Tem a percepção de que os pacientes com quem trabalha estão conscientes dos serviços de terapia ocupacional?

Se a resposta for não, explicar possíveis causas:

Se a resposta for sim, explique como é que eles estão conscientes destes serviços:

Acha que eles têm acesso a serviços de terapia ocupacional?

Em que situações são os pacientes encaminhados para o terapeuta ocupacional?

Quem o encaminha (médicos, psicólogos, fisioterapeutas, assistentes sociais, etc.)?

Percebe algum impacto da terapia ocupacional na vida do paciente?

Considera que a inclusão de um terapeuta ocupacional como parte da equipa multidisciplinar poderia ter um valor extra para a gestão adequada da ATTRv?

Once again, thank you.

The following steps

After three years of work with a focus on patients diagnosed with ATTRv, the developed occupational intervention model will be presented to other groups and associations in order to raise awareness of occupational therapy and offer specific interventions to rare disease patients.

The organisations involved are: Asociación Costarricense de Amiloidosis, Asociación Española de Amiloidosis, Asociación de Paraparesia Espástica Familiar, Asociación Síndrome Alcohólico Fetal, Asociación Piel de Mariposa, Asociación de Esclerosis tuberosa and Asociación Nacional del Síndrome de Ehlers Danlos, Hiperlaxitud y colagenopatías.

People doing things...