CONCLUSIONS

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-Mitjançant l'anàlisi de lligament s'han pogut excloure les 5 formes de LGMD autosòmiques dominants prèviament descrites, com a possibles responsables de les manifestacions de la malaltia en el nostre pedigrí.

-També mitjançant l'anàlisi de lligament s'ha pogut mapar el gen responsable de la malaltia a 7q31-q32.

-A partir de l'anàlisi dels individus recombinants s'ha pogut delimitar la regió candidata final entre els marcadors D7S680 (centromèric) i D7S2544 (telomèric), abastant una distància d'unes 3,7 Megabases.

-Després d'escollir-la com a principal gen candidat donada la seva funció d'unió a actina i la seva expressió a múscul, s'ha exclòs la Filamina C com a gen responsable de la malaltia, no trobant-se cap mutació patogènica a la seva seqüència nucleotídica ni cap alteració a nivell d'expressió en individus afectes respecte dels sans. Tampoc s'ha trobat cap expansió en regions de seqüències repetitives del gen que poguessin suposar una causa de la malaltia i a la vegada una explicació del fenomen d'anticipació observat a la família estudiada. S'ha trobat un polimorfisme, el qual s'ha utilitzat com a marcador per acotar la regió candidata amb més precisió.

-Un cop exclosa la Filamina C, s'han identificat una sèrie de gens candidats a la regió lligada que podrien estar implicats en la malaltia, en base a la seva expressió a teixit muscular i/o funció (ARF5, Fascina 3, Calumenina, LOC63220, Transportina, UBE2H, HSPC216 i KIAA0265). Actualment s'està efectuant la seqüenciació d'alguns d'aquests gens, utilitzant-se també alguns polimorfismes puntuals trobats en ells com a marcadors adicionals per tal de delimitar encara més la regió on es troba el gen responsable de la malaltia. La utilització d'aquests nous polimorfismes és de gran utilitat, donada la dificultat de trobar nous marcadors microsatèl.lits polimòrfics amb prou grau d'heterozigositat perque siguin informatius.

-La continua actualització de les diferents bases de dades contribuirà amb tota seguretat a delimitar definitivament les posicions i localitzacions exactes tant de marcadors com

de gens descrits i predits, permetent l'abordatge més precís dels possibles gens candidats.

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