Epidemiology and molecular characterization of adult genetic myopathies in a southeastern region of Spain

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Introduction. Genetic myopathies constitute a collection of rare diseases that significantly impact patient functionality and quality of life. Early diagnosis of genetic myopathies can prevent future complications and provide families with genetic counselling. Despite the substantial impact of genetic myopathies on the adult population, the global epidemiology of these disorders is inadequately addressed in the literature.

Aims. To enhance understanding of both the epidemiology and genetics of these disorders within the province of Alicante, situated in southeastern Spain.

Material and methods. Between 2020 and 2022, a prospective observational study was conducted at the Alicante Health Area-General Hospital, enrolling patients aged 16 years or older with suspected genetic myopathies. Sociodemographic, clinical, and genetic data were collected. The reference date for prevalence calculation was established as December 31, 2022. Official demographic data of the health area were used to set the population at risk.

Results. In total, 83 patients were identified with confirmed genetically related myopathy, resulting in an overall prevalence of 29.59 cases per 100,000 inhabitants. The diagnostic yield for molecular genetic testing was found to be 69.16%. The most prevalent genetic myopathies identified included myotonic dystrophy (27.5%), dystrophinopathies (15.7%), and facioscapulohumeral dystrophy (15.7%).

Conclusion. The prevalence of genetic myopathies can vary considerably depending on the geographical region and the studied population. The analysis of diagnostic yield suggests that genetic studies should be considered useful in the diagnosis of genetic myopathies.

Key words. Epidemiology. Genetics. Muscular dystrophies. Myopathies. Prevalence. Spain.

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Introduction

Genetic myopathies are classified as rare diseases due to their low prevalence. They impact muscular tissue to varying degrees via distinct molecular pathways, including direct structural damage, dysregulation of muscular metabolism, and alterations in ionotropic channels [1]. These conditions can manifest with symptoms such as muscle weakness, cramps, stiffness, contractures, pain, and fatigue and can also affect other organ systems, such as cardiovascular function, vision, or even cognition [2].

Despite their low prevalence, genetic myopathies collectively impose a substantial burden of disability, exerting widely variable effects on patients' quality of life [3,4]. The incidence of these diseases varies in different geographical areas, underscoring the necessity of ascertaining local prevalence to devise effective prevention and treatment approaches. Timely diagnosis can prevent future complications and provide affected families with appropriate environmental adaptations and genetic counselling. From a public health perspective, comprehending the prevalence of distinct genotypes within the served population remains crucial.

Most epidemiological studies on myopathies tend to focus on specific types or subtypes of muscular diseases, with a limited number addressing genetic myopathies as a unified group. The scarcity of studies exploring the worldwide epidemiology of adult genetic myopathies is particularly pronounced in Spain.

Studying the genetic alterations associated with genetic myopathies can provide important insights into the underlying causes of these diseases. This study contributes to a better understanding of the

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epidemiology and genetics of these conditions in the Alicante healthcare area, a region in southeastern Spain. This knowledge is crucial for developing more effective interventions and treatments for individuals affected by genetic myopathies, as well as improving the accuracy of diagnoses and genetic counselling.

Material and methods

Prospective observational study based on the identification of patients with genetic myopathies from multiple data sources in a healthcare area in Alicante (southeastern Spain). A prospective follow-up of these patients was conducted from 2020 to 2022. In some cases, genetic segregation studies were performed, identifying newly affected patients. Demographic, clinical, and genetic data were collected. This study was granted ethical approval by the ethical research committee of the hospital, and all procedures adhered rigorously to the principles outlined in the Helsinki Declaration of 1975, as revised in 2000. All patients gave their informed consent for inclusion before they participated in the study.

Data sources

Data were collected from electronic medical records of different computerized registries within the healthcare area. In the healthcare area under investigation, every individual within the population possesses an electronic medical record. Records from the specialized neuromuscular clinic of the neurology department at the hospital were also reviewed. Patients from other healthcare areas were excluded from these registries. Diagnostic codes from the tenth revision (ICD-10) of the international classification of diseases related to muscle disorders were employed for the identification of patients with genetic myopathies, facilitated by the capability of electronic medical records to screen specific ICD-10 diagnoses. The following codes were searched: A36.81, G13.0, G71, G71.0, G71.00, G71.01, G71.02, G71.11, G71.2, G71.20, G71.21, G71.22, G71.220, G71.228, G71.29, G71.3, G72, G72.0, G72.1, G72.2, G72.4, G72.49, G72.8, G72.81, G72.89, and G72.9.

Selection criteria

Patients aged 16 years and above presenting with the ICD-10 diagnostic codes specified above and evaluated within the neuromuscular clinic for suspected myopathy at the Alicante - General Hospital healthcare area were included in the study for comprehensive assessment. Patients with a final diagnosis of genetic myopathies were included in the study.

Patient follow-up

Patients received ongoing monitoring within the specialized neuromuscular clinic. This included confirmation of a consistent phenotype and validation of sociodemographic patient data. Additionally, relevant complementary tests were meticulously reviewed and, if necessary, requested. In certain cases, family members were also investigated, and segregation studies were undertaken if not previously conducted.

Genetic studies

Genetic studies were conducted in accredited genetics laboratories within the Spanish healthcare system using standard procedures on peripheral blood samples extracted from patients, with prior informed consent. Different molecular techniques were employed based on clinical suspicion (see supplementary material). Family segregation studies were performed in families of patients with previously undescribed likely pathogenic single nucleotide variants, following the American College of Medical Genetics and Genomics [5].

Genetic myopathy diagnosis

The diagnosis of genetic myopathies was based on the presence of a confirmed molecular defect through blood extraction or a compatible phenotype and genetic defect pedigree within the family. For specific instances involving mitochondrial myopathy, congenital muscular dystrophy, and calpain deficiency, the diagnosis was accepted upon observing a compatible phenotype and a muscle biopsy defect. Cases of probable genetic myopathies were defined as cases with well-founded suspicion based on phenotype and clinical context, lacking genetic confirmation, as described by Harris et al [6].

Prevalence calculation and statistical analysis

The date selected for prevalence estimation was December 31, 2022. The at-risk population encompassed individuals enrolled within the General Hospital healthcare department area of Alicante,

totaling 280,535 inhabitants as of the prevalence calculation date. The statistical analysis was carried out using R software version 4.0.5 through the RStudio user interface version 1.4. Qualitative variables were described by their frequency distribution and expressed as percentages. Quantitative variables that followed a normal distribution (Shapiro test) were described based on the mean and standard deviation (SD). These results were expressed as mean ± SD. An inferential estimation of prevalences was performed using confidence intervals based on the population of the Alicante general health department, and the result was expressed along with the 95% confidence interval (95% CI).

Results

Our search strategy yielded 383 potential cases after eliminating duplicates. Among these, 145 cases fulfilled the eligibility criteria. Subsequent comprehensive evaluations conducted during multiple visits to the neuromuscular diseases clinic led to the identification of 83 cases with genetically confirmed genetic myopathies. Of these, 75 cases were pinpointed through genetic studies, while 8 cases were revealed via muscle biopsy or detection of mitochondrial DNA alterations. The remaining patients were categorized as follows: 4 patients had passed away before the prevalence calculation date, 5 patients received diagnoses differing from myopathy, 15 patients were diagnosed with myopathy stemming from nongenetic factors, and 37 cases exhibited a phenotype and clinical history indicative of genetic myopathy but lacked genetic validation.

The mean age of 83 confirmed cases at the time of the study was 49.7 years (SD = 16.9; range 17-82), with a slight male prevalence of 54.2%. The overall prevalence of genetically confirmed genetic myopathies within the healthcare region was 29.59 cases per 100,000 inhabitants (95% CI: 23.71-36.87). A breakdown of prevalence across groups and subgroups, in addition to other sociodemographic data, is presented in table I. Considering both the genetically confirmed cases and potential genetic myopathies instances lacking genetic confirmation at the time of the study, the estimated total prevalence was 42.78 cases per 100,000 inhabitants (95% CI: 35.61-51.34).

Molecular diagnostic assessments were carried out for 112 patients, with 75 of them receiving a confirmed diagnosis, yielding a diagnostic success rate of 66.96%. Table II displays all the genetic alterations identified in the patients with genetic myopathies. Notably, three of these molecular anomalies were previously unreported and were categorized as variants of uncertain significance according to the American College of Medical Genetics criteria [5]. Nevertheless, the alignment of clinical phenotype, in silico molecular function predictions, and results from segregation studies strongly pointed towards a correlation between the pathological condition and the genetic alteration.

Discussion

Global prevalence of genetic myopathies

The study area exhibited a genetic myopathies prevalence of 29.59 cases per 100,000 inhabitants on the selected date. In Spain, another singular study investigating genetic myopathies prevalence, executed in Navarra (a northern region), unveiled a prevalence of 59 per 100,000 inhabitants [7].

When compared to other European areas, the prevalence reported in our study is akin to the figure calculated in a study from northern England, which identified a prevalence of 37 cases per 100,000 inhabitants [8]. In contrast, it diverges from the prevalence noted in another study conducted in northern Norway, where genetic myopathies prevalence reached 67,7 (95% CI: 60.8-75.4) [8]. On a global scale, a study conducted in New Zealand identified a prevalence of 22.7, mirroring our findings [9].

Only a limited number of studies assess prevalence across diverse geographical regions worldwide. In a comprehensive systematic review that amalgamated the prevalence of muscular dystrophies on a global scale, the combined prevalence was observed to be 16.14 cases per 100,000 inhabitants (95% CI: 11.21-23.23) [10].

Prevalence of genetic myopathy subgroups

Upon meticulous analysis of the estimated prevalences within distinct subgroups, no significant deviations are observed compared to those reported in specific meta-analyses. In our study, the estimated prevalence of dystrophinopathies was 4.63 cases per 100,000 inhabitants, closely aligning with the range elucidated in a systematic review focusing on the worldwide prevalence of Duchenne muscular dystrophy, which reported a prevalence of 7.1 cases per 100,000 inhabitants [11]. A similar pattern emerges with the estimated prevalence of

Table I. Demographics and Prevalence of Genetic Myopathies (GMs) in the healthcare area of Alicante.

	N (%)	Men:women	Mean age (SD)	Prevalence[a]	95% CI
Total	**83 (100%)**	**45:38**	**49.70 (16.9)**	**29.59**	**23.71-36.87**
Muscular dysthrophies	**72 (86.7%)**	**38:34**	**47 (16.99)**	**23.88**	**20.22-32.51**
MD-1	22 (26.5%)	12:10	42.33 (12.69)	7.84	5.03-12.09
Dystrophinopathies	13 (15.7%)	9:4	36.31 (16.32)	4.63	2.58-8.15
• Duchenne	8 (9.6%)	8:0	29 (10.99)	2.85	1.33-5.86
• Becker	2 (2.4%)	1:1	51.50 (23.33)	0.71	0.12-2.88
• Symptomatic carrier	3 (3.61%)	0:3	52.33 (3.78)	1.07	0.28-3.41
FSHD	13 (15.7%)	6:7	51.67 (17.46)	4.63	2.58-8.15
• FSHD 1	10 (12.04%)	4:6	53.78 (18.12)	3.2	1.57-6.33
• FSHD 2	3 (3.61%)	2:1	45.33 (16.86)	1.07	0.28-3.41
OPMD	8 (9.6%)	5:3	61.62 (13.88)	2.85	1.33-5.86
LGMD	7 (8.4%)	3:4	50.14 (16.65)	2.5	1.09-5.39
• LGMD 1B	2 (2.4%)	1:1	39 (18.38)	0.71	0.12-2.88
• LGMD 2A	1 (1.2%)	0:1	53	0.35	0.02-2.32
• LGMD 2J	3 (3.6%)	1:2	48 (13.89)	1.07	0.28-3.41
• LGMD 2L	1 (1.2%)	1:0	76	0.35	0.02-2.32
CMD	4 (4.8%)	0:5	58.75 (14.22)	1.43	0.46-3.92
• Central core	1 (1.2%)	0:1	68	0.35	0.02-2.32
• Centronuclear	1 (1.2%)	0:1	61	0.35	0.02-2.32
• Minicore	1 (1.2%)	0:1	68	0.35	0.02-2.32
• UCMD	1 (1.2%)	0:1	38	0.35	0.02-2.32
EDMD	2 (2.4%)	1:1	44.50 (33.23)	0.71	0.12-2.88
MD-2	1 (1.2%)	1:0	62	0.35	0.02-2.32
MFM3	1 (1.2%)	1:0	64	0.35	0.02-2.32
MFM5	1 (1.2%)	1:0	51	0.35	0.02-2.32
Mitochondrial	**7 (8.4%)**	**4:3**	**54.67 (19.78)**	**2.49**	**1.09-5.39**
Complex III deficiency	2 (2.4%)	1:1	68.5 (7.78)	0.71	0.12-2.88
PDCD	1 (1.2%)	0:1	44	0.35	0.02-2.32
nDNA mutation	1 (1.2%)	0:1	75	0.35	0.02-2.32
mDNA deletion	1 (1.2%)	1:0	70	0.35	0.02-2.32
Other	2 (2.4%)	2:0	36 (16.97)	0.71	0.12-2.88
GSD	**2 (2.4%)**	**2:0**	**56.5 (21.9)**	**0.71**	**0.12-2.88**
Pompe	1 (1.2%)	1:0	72	0.35	0.02-2.32
McArdle	1 (1.2%)	1:0	41	0.35	0.02-2.32
Channelopathies	**2 (2.4%)**	**1:1**	**37.50 (12.02)**	**0.71**	**0.12-2.88**
CACNA1S	1 (1.2%)	1:0	46	0.35	0.02-2.32
CLCN1	1 (1.2%)	0:1	29	0.35	0.02-2.32

95% CI: confidence Interval of the prevalence calculation; CACNA1S: calcium voltage-gated channel subunit alpha1 S; CLCN1: chloride voltage-gated channel 1; CMD: congenital muscular dystrophy; EDMD: Emery-Dreifuss muscular dystrophy; FSHD: facioscapulohumeral muscular dystrophy; GSD: glycogen storage disease; LGMD: limb girdle muscular dystrophy; nDNA: nuclear DNA; MD-1: myotonic dystrophy type 1; MD-2: myotonic dystrophy type 2; MFM: myofibrillar myopathy; mDNA: mitochondrial DNA; OPMD: oculopharyngeal muscular dystrophy; PDCD: pyruvate dehydrogenase complex deficiency; SD: Standard deviation; UCMD: Ullrich congenital muscular dystrophy. [a] Cases per 100,000 inhabitants.

Table II. Molecular defects found in patients with genetic myopathy.

	Gene	Mutation type	Sequence variation	Position	Zigosis	*n*
MD-1	*DMPK*	CTG expansion			Het	22
Dystrophinopathies	*DMD*				Het	13
		deletion	–	–		10
				exon 1 to 5		1
				exon 3 to 7		1
				exon 45		1
				exon 47 to 48		2
				exon 48		2
				exon 53		2
				exon 59		1
		duplication		exon 21 to 62		1
		nonsense	–	–		2
			c.5530C>T	exon 39		1
			c.4084C>T	exon 30		1
FSHD 1	*DUX*	deletion Chr. 4		D4Z4 region	Het	10
FSHD 2	*SMCHD1*		–	–	Het	3
		SNV	c.3802-2A>G			1
		deletion	c.1131+1delG			2
OPMD	*PAPBN1*	GCN expansion			Het	8
LGMD						6
• LGMD 1B	*LMNA*	SNV	c.1130G>A		Het	2
• LGMD 2J	*TTN*	SNV	c.21088C>T	–	Het	3
• LGMD 2L	*ANO5*	insertion	c.1622_1623insA	–	Hom	1
CMD						3
UCMD	*COL6A2*	SNV	c.801+2T>C		Het	1
• Centronuclear	*BIN1*	SNV	c.700C>T		Hom	1
• Central core	*RYR1*	SNV	c.6207A>G		Het	1
EDMD	*SYNE2*	SNV	c.14518T>C		Het	2
MD-2	*CNPB*	CCTG expansion			Het	1
MFM3	*MYOT*	SNV	c.179C>T		Het	1

Table II. Molecular defects found in patients with genetic myopathy *(cont.)*.

	Gene	Mutation type	Sequence variation	Position	Zigosis	*n*
MFM5	*FLNC*	SNV	c.755C>T		Het	1
Mitocondrial	*TWNK*	SNV	c.1106C>T	exon 1	Het	1
GSD						2
• Pompe	*GAA*	unknown	–	–	–	1
• McArdle	*PYGM*	SNV	c.148C>G		Hom	1
Channelopathies						2
	CLCN1	SNV	c.712A>T		Het	1
	CACNA1S	SNV	c.1583G>A		Het	1

CMD: congenital muscle dystrophy; EDMD: Emery-Dreifuss muscular dystrophy; FSHD: facioscapulohumeral muscular dystrophy; GSD: glycogen storage disease; Het: heterozygous; Hom: homozygous; LGMD: limb girdle muscular dystrophy; MD-1: myotonic dystrophy type 1; MD-2: myotonic dystrophy type 2; MFM: myofibrillar myopathy; OPMD: oculopharyngeal muscular dystrophy; SNV: single nucleotide variant; UCMD: Ullrich congenital muscular dystrophy.

congenital myopathies, which was determined to be 1.43 (95% CI: 0.46-3.92), akin to the figure derived from a meta-analysis encompassing global congenital myopathy prevalence, where they reported a prevalence of 1.62 (95% CI: 1.13-2.11) [12].

Spain's epidemiological investigations primarily delve into the prevalence and/or incidence of specific genetic myopathies types. One particularly extensively studied type is type 1 myotonic dystrophy. In various regions, the reported data include an incidence of 20.61 cases per million person-years in Aragon, a prevalence of 26.5 per 100,000 inhabitants in Guipuzcoa, and a prevalence of 10.8 per 100,000 inhabitants in Mallorca [13-15]. In contrast, our study reveals a prevalence of 7.84 myotonic dystrophy type 1 cases per 100,000 inhabitants (95% CI: 5.03-12.09). While the case numbers are relatively modest and observed variations might be attributed to methodological considerations, this investigation suggests a higher prevalence of genetic myopathies in northern Spain compared to the southern regions.

Diagnostic rate of genetic studies

The diagnostic rate of genetic studies in myopathies exhibits broad variability, spanning from 16% to 65% depending on the specific cohort under investigation and the NGS approach used [16-20]. Regarding molecular diagnosis, our study identified genetic diagnosis for 75 out of the 112 patients examined, constituting a diagnostic yield of 66.96%. This closely mirrors the 64% achievement at a reference center in Germany [21]. These outcomes underscore the pivotal role of genetic testing in diagnosing genetic myopathies patients and validate the congruence of our findings with those reported by other reference centers worldwide.

Conclusions

The prevalence of genetic myopathies exhibits notable variability contingent upon geographic location and the population under investigation. In the Alicante healthcare area, the observed prevalence closely aligns with the reported global figures. Our diagnostic yield analysis underscores the significance of genetic studies as a valuable diagnostic tool in the realm of genetic myopathies. The imperative to persist in investigating the epidemiology of these conditions remains paramount, alongside the facilitation of streamlined access to specialized population-based registries catering to neuromuscular disorders and other rare afflictions, all while conscientiously considering the inherent diversity within these disorders.

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Annex. Molecular diagnostic techniques.

Molecular diagnostic techniques were requested as part of the standard clinical practice according to the considerations of the attending physician. Blood samples were taken from the patients following standard peripheral blood sampling procedures, sent to reference centers, and the relevant studies were carried out in each case by these centers. Different sequencing techniques were performed:

- Next generation sequencing (NGS) techniques using different NGS platforms (mainly based on Illumina HiSeq®), for the identification of specific genes or variants by groups or panels of genes related to neuromuscular diseases or genetic origin muscle myopathies (Table annex).

- Direct Sanger sequencing techniques in family segregation studies aimed at finding the variant of interest or in cases directed at the most common variants of interest of a specific gene and for confirmation of pathogenic variants or variants of interest detected in NGS techniques.

Screening studies for copy number variations (CNVs) were also conducted in some cases. For this, analyses based on read depth, comparison of the analyzed samples against reference samples, and copy number calling, annotation of candidate variables, selection of genes of clinical interest, and evaluation of CNVs in the selected genes were performed.

Other molecular diagnostic studies were conducted in a targeted manner for the detection of specific genetic defects:

- Screening for dystrophin gene deletions/duplications using MLPA (multiplex ligation-dependent probe amplification) to detect the copy number of all exons of the dystrophin gene, which causes dystrophinopathies such as Duchenne and Becker muscular dystrophy.

- Measurement of the CTG trinucleotide sequence expansion in the 3′ region of the DMPK gene using RP-PCR (repeat-primed PCR) and analysis of fragment length. The expansion of (CTG)n repeats is responsible for type I myotonic dystrophy or Steinert's disease.

- For the detection of D4Z4 copy number loss at the chromosome 4 telomeric region, responsible for facioscapulohumeral muscular dystrophy

Annex table. Genetic panels of interest in neuromuscular diseases.

Neuromuscular diseases panel used
*ABHD5, ACADL, ACADM, ACADS, ACADVL, ACE, ACTA1, ACTN3, AGK, AGL, AGRN, ALDOA, ALG2, ALG13, ALG14, AMPD1, ANO5, ATP2A1, B3GALNT2, B3GNT1, BAG3, BIN1, C10ORF2, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL6A1, COL6A2, COL6A3, COL9A2, COL9A3, COL12A1, COL13A1, COLQ, COMP, COX15, CPT1B, CPT2, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FAM111B, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GBE1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADHA, HADHB, HNRNPDL, HNRPDL, IGHMBP2, ISCU, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL9, KLHL40, KLHL41, LAMA2, LAMB2, LAMP2, LARGE, LDB3, LDHA, LIMS2, LMNA, LPIN1, LRP4, MATR3, MEGF10, MSTN, MTM1, MTMR14, MTTP, MUSK, MYBPC3, MYF6, MYH2, MYH3, MYH7, MYH14, MYOT, NEB, OPA1, ORAI1, PABPN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPARGC1A, PREPL, PRKAG2, PTPLA, PTRF, PYGM, RAPSN, RRM2B, RYR1, SCN4A, SEPN1, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A4, SLC25A20, SLC52A3, SMCHD1, SNAP25, STAC3, STIM1, STIM2, SUCLA2, SYNE1, SYNE2, SYT2, TARDBP, TAZ, TCAP, TIA1, TK2, TMEM5, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, UBA1, VAPB, VCP, VMA21, YARS2***
Mitochondrial DNA maintenance-related nuclear DNA genes panel involved in mitochondrial diseases
DGUORK, MNF2, MPV17, OPA1, POLG, POLG2, RRM2B, SLC25A4 (ANT1), SUCLA2, SUCLG1, TK2, TWINKLE (C10orf2) y TYMP

type 1 (FSHD1), a specific probe is used after enzymatic DNA digestion and subsequent fragment length analysis. A length of less than 38kb was considered pathological, although in cases with lengths between 34 and 38kb, the 4QA161 haplotype was sought, a condition necessary for the development of the disease. In some cases, in laboratories outside the Valencian community, the length of the D4Z4 region was analyzed by Southern blot.

Epidemiología y caracterización molecular de las miopatías genéticas en adultos en una región del sureste de España

Introducción. Las miopatías genéticas constituyen un conjunto de enfermedades raras que impactan significativamente en la funcionalidad y la calidad de vida del paciente. Un diagnóstico temprano de las miopatías genéticas puede prevenir complicaciones futuras y proporcionar a las familias asesoramiento genético. A pesar del impacto sustancial de las miopatías genéticas en población adulta, la epidemiología global de estos trastornos está inadecuadamente abordada en la bibliografía.

Objetivos. Mejorar el entendimiento tanto de la epidemiología como de la genética de estos trastornos en la provincia de Alicante, situada en el sureste de España.

Material y métodos. Entre 2020 y 2022, se llevó a cabo un estudio observacional prospectivo en el área de salud Alicante-Hospital General, que incluyó a pacientes de 16 años o más con sospecha de miopatías genéticas. Se recopilaron datos sociodemográficos, clínicos y genéticos. La fecha de referencia para el cálculo de la prevalencia se estableció el 31 de diciembre de 2022. Se utilizaron datos demográficos oficiales del área de salud para establecer la población en riesgo.

Resultados. En total, se identificó a 83 pacientes con miopatía genéticamente confirmada, lo que dio lugar a una prevalencia total de 29,59 casos por cada 100.000 habitantes. El rendimiento diagnóstico de las pruebas genéticas moleculares fue del 69,16%. Las miopatías genéticas más frecuentes incluyeron la distrofia miotónica (27,5%), las distrofinopatías (15,7%) y la distrofia facioescapulohumeral (15,7%).

Conclusión. La prevalencia de las miopatías genéticas puede variar considerablemente dependiendo de la región geográfica y la población estudiada. El análisis del rendimiento diagnóstico sugiere que los estudios genéticos deberían considerarse útiles en el diagnóstico de las miopatías genéticas.

Palabras clave. Distrofia muscular. Epidemiologia. España. Genética. Miopatías. Prevalencia.