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SUPPLEMENTAL DATA

DYSF gene variant spectrum in Arab populations across eight countries: A systematic review

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Full article is available at the following link: DYSF gene variant spectrum in Arab populations across eight countries: A systematic review

Table S1. Caracteristics of the included studies in a systematic review of *DYSF* gene mutations in Arab populations

Authors/Year	Country	Study Type	Population	Patients Arab with genetic mutation	Age at diagnosis	Mean age at onset	Gender	Clinical presentation	Genetics test	Mutation of DYS gene found in arabe population
Argov Z et al., 2000 [30]	Libyan	Descriptive observational (case series)	29 patients	29	38,30,28	20.3±5.5 years	9M/20F	LGMDR2	DNA Extraction, PCR,SSCP,BmyI and AvaII or Sau96I restriction enzyme	c.4872delG (p.Glu1624Aspfs*10)
Aoki M et al., 2001 [19]	Mumulti-country (Libyan)	Descriptive observational (case series)	21 patients	1	NR	19.0 ±3.9 years	NR	LGMDR2	SSCP and direct sequencing	c.5434ins (5410–5433 tandem repeat at 5434)
Nguyen K et al., 2005 [47]	Marseille (Morocco)	Descriptive observational (case	34 patients	2	NR	NR	21M/13F	LGMDR2/PD phenotype	Western-blotting,immunohistochemistry,SSCP or Dhplc and	c.2858dupT (p.Phe954Valfs*2) and c.5594delG (p.Gly1865Alafs*101)

		series)							sequencing	
Nguyen K et al., 2007 [38]	Marseille/Paris (Morocco,Algeria,Tunisia)	Retrospective (case series)	40 patients	14	NR	39.1±11.5 years	21M/19F	LGMDR2/PD phenotype/MM	Immunohistochemistry,Western blot	c.2858dupT (p.Phe954Valfs*2),c.4200delC (p.Ile1401Serfs*47),c.5594delG (p.Gly1865Alafs*101),c.1795_1799dup (p.Ala601Thrfs*28),c.1663C>T (p.Arg555Trp),c.3035G>A (p.Trp1012*),c.1834C>T (p.Gln612*),c.3967C>T (p.Gln1323*),c.4876delG (p.Val1626Tyrfs*8),c.1392dupA (p.Asp465Argfs*9)
Rajab A et al., 2015 [48]	Oman	Descriptive observational (case series)	NR	1	NR	NR	NR	LGMDR2	NR	c.526C>T (p.Gln176*)
Nouioua S., 2017 [40]	Algeria	Descriptive observational (case report)	2 patients	2	37.48	NR	1M/1F	LGMDR2	NGS	c.2643+1G>A (p.Tyr838_Thr881del)
Algahtani H., 2018	Saudi Arabia	Descriptive	1 patien	1	41 years	NR	1F	LGMDR2	NGS	c.164dupA (p.Ile57Hisfs*8)

[42]		observational (case report)	t							
Okubo M., 2018 [46]	Japan (Egypt)	Descriptive observational (case series)	3 Patients	1	33 years(Egyptian)	NR	1F	NR	NGS and direct sequencing	c.4431G > T (p.Trp1477Cys)
Rekik S., 2020 [39]	Tunisia	Descriptive observational (case report)	3 Patients	3	55,53,48	NR	2M/1F	LGMDR2,DMAT,PD phenotype	NGS	c.4597-2A>G
Aldosari KH., 2020 [41]	Saudi Arabia	Descriptive observational (case report)	1 patient	1	26 years	NR	NR	LGMDR2	Exome sequencing	c.166dupA (p.L55fs)
Megarbane A., 2022 [44]	France , Lebanon	Retrospective observa	506 patients	2	21 years of 2 patients	8 years	291M/215F	LGMDR2	Multiplex PCR, Southern blot,MLPA,Sa	c.5438T >C (p.Leu1813Pro)

		tional (case series)							nger sequencing,NGS	
El Sherif R., 2020 [45]	Egypt	Descriptive observational (case series)	24 patients	24	NR	NR	NR	LGMDR2	NR	c.342+1G>A, c.755C>T (p.Thr252Met),c.1958delG(p.Gly653Valfs*3),c.2190dupA(p.Pro731Thrfs*23) c.3597G>A (p.Trp1199*),c.3944_3948delinsG(p.Glu1315Glyfs*29),c.4101G>A,c.6124C>T (p.Arg2042Cys),c.4431G>T(p.Trp1477Cys), c.5983_5984del(p.Ser1995*)
Alharbi N., 2022 [31]	Saudi Arabia	Descriptive observational (case series)	53 patients	53	22–61(39.55 ± 8.82)	17.79 ± 3.48	28M/25F	LGMDR2, MM, hyperCKemia,PD phenotype	Whole Exome Sequencing	c.89-1G>A, c.631A>G (p.Lys211Glu),c.164_165insA,c.1351A>G (p.Met451Val),c.5264T>C (p.Leu1755Pro), c.3702+6T>A,c.1422C>T,c.359A>C (p.Gln120Pro),c.1433delC,c.4210_4211insC(p.Lys1404Thrfs*5),c.5201A>T (p.Tyr1734Phe), c.5907G>C(p.Trp1969Cys),c.755C>T(p.Thr252(Met)
Belhassen I., 2023 [29]	Tunisia	Descriptive observational (case	20 patients	20	50.5 years(median age)	17 years old (IQR = [16,8–28,4]	10M/10F	LGMDR2, MM, hyperCKemia,PD phenotype,DMAT	NGS	c.1392dupA(p.Asp465Argfs*9),c.4200dupC (p.Ile1401Hisfs*8),c.4597-2A>G,c.5525G>A,c.1392dupA (p.Asp465Argfs*9)

		series)								

Abbreviations: MLPA: Multiplex ligation-dependent probe amplification; F: Female; M: Male; LGMDR2: Limb-girdle muscular dystrophy, recessive type 2; SSCP: Single-strand conformational polymorphism; NR: Not reported; MM: Miyoshi myopathy; PD: Proximodistal; NGS: Next-generation sequencing.

JBI Critical Appraisal Checklist for case reports

Table S2. Quality assessment of included studies utilizing the JBI critical appraisal checklist for case reports

Case report	Q1	Q2	Q3	Q4	Q5	Q6	Q7	Q8	Overall
Algahtani.,2018	Yes	Yes	Yes	Yes	No	Yes	Not Applicable	Yes	Include
Aldosari KH.,2020	Yes	Unclear	Yes	Yes	No	Yes	Not Applicable	Yes	Include
Nouioua S.,2017	Yes	Unclear	Yes	Yes	Not Applicable	Not Applicable	Not Applicable	Yes	Include
Rekik S.,2020	Yes	Yes	Yes	Yes	Not Applicable	Not Applicable	Not Applicable	Yes	Include

Abbreviations: Q: Question; JBI: Joanna Briggs Institute

Overall appraisal: Include, Exclude, Seek further info.

Questions

1. Were patient's demographic characteristics clearly described?
2. Was the patient's history clearly described and presented as a timeline?
3. Was the current clinical condition of the patient on presentation clearly described?
4. Were diagnostic tests or methods and the results clearly described?
5. Was the intervention(s) or treatment procedure(s) clearly described?

6. Was the post-intervention clinical condition clearly described?
7. Were adverse events (harms) or unanticipated events identified and described?
8. Does the case report provide takeaway lessons?

JBI Critical Appraisal Checklist for case series

Table S3. Quality assessment of included studies using the JBI critical appraisal checklist for case series

Case series	Q1	Q2	Q3	Q4	Q5	Q6	Q7	Q8	Q9	Q10	Overall
Argov Z et al.,2000	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	Yes	Yes	Not Applicable	Include
Aoki M et al.,2001	Yes	Yes	Yes	No	No	Yes	Yes	Unclear	Unclear	Not Applicable	Include
Nguyen K et al.,2005	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	Unclear	Unclear	Not Applicable	Include
Nguyen K et al.,2007	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Unclear	Yes	No	Include
Rajab A et al.,2015	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	No	Yes	Not Applicable	Include
Okubo M.,2018	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	No	Yes	Not Applicable	Include
Megarbane A.,2022	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	Yes	Yes	Not Applicable	Include
El Sherif R.,2020	Yes	Unclear	Unclear	Unclear	Unclear	Yes	Yes	No	No	Not Applicable	Seek further info
Alharbi N.,2022	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	No	Yes	No	Include
Belhassen I.,2023	Yes	Yes	Yes	Unclear	Unclear	Yes	Yes	Unclear	Yes	No	Include

Abbreviations: Q: Question; JBI: Joanna Briggs Institute.

Overall appraisal: Include, Exclude, Seek further info.

Questions

1. Were there clear criteria for inclusion in the case series?

2. Was the condition measured in a standard, reliable way for all participants included in the case series?
3. Were valid methods used for identification of the condition for all participants included in the case series?
4. Did the case series have consecutive inclusion of participants?
5. Did the case series have complete inclusion of participants?
6. Was there clear reporting of the demographics of the participants in the study?
7. Was there clear reporting of clinical information of the participants?
8. Were the outcomes or follow-up results of cases clearly reported?
9. Was there clear reporting of the presenting site(s)/clinic(s) demographic information?
10. Was statistical analysis appropriate?

Table S4. Summary of genomic information for *DYSF* gene variants

Variants	Transcript ID	Genomic coordinates	Genome assembly	ACMG classification	ACMG criteria	LOVD ID
c.89-1G>A	NM_003494.3	g.71708012G>A	GRCh37/hg19	Pathogenic	PVS1, PP1-S, PM2, PM3, PM4, PP1-M, PP1, PP3, PP4	0000597599
c.164dupA (p.Ile57Hisfs*8)	NM_003494.3	g.71709028dup	GRCh37/hg19	Pathogenic	NR	0000449649
c.631A>G (p.Lys211Glu)	NM_003494.4	g.71513889A>G	GRCh38/hg38	Pathogenic	NR	NR
c.164_165insA	NM_003494.4	NR	GRCh38/hg38	Pathogenic	NR	NR
c.166dupA (p.L55fs)	NM_001130455	NR	NR	Pathogenic	NR	NR
c.1351A>G (p.Met451Val)	NM_003494.3	g.71762217A>G	GRCh37/hg19	VUS	NR	0000462992
c.5264T>C (p.Leu1755Pro)	NM_003494.4	g.71667439T>C	GRCh38/hg38	NR	NR	NR
c.3702+6T>A	NM_003494.4	g.71598751T>A	GRCh38/hg	NR	NR	NR

			38			
c.1422C>T	NM_003494.4	g.71539181C>T	GRCh38/hg 38	Likely pathogenic	NR	NR
c.359A>C (p.Gln120Pro)	NM_001130987 .2	NR	NR	Likely pathogenic	NR	NR
c.1433delC	NM_001130976	NR	NR	Pathogenic	NR	NR
c.4210_4211insC (p.Lys1404Thrfs*5)	NM_003494.4	g.71612683_71612684insC	GRCh38/hg 38	Pathogenic	NR	NR
c.4873C>T (p.Arg1625*)	NM_001130987 .2	g.71658995C>T	GRCh38/hg 38	Pathogenic	NR	NR
c.5201A>T (p.Tyr1734Phe)	NM_001130985 .2	g.71665251A>T	GRCh38/hg 38	NR	NR	NR
c.5907G>C (p.Trp1969Cys)	NM_003494.4	g.71679196G>C	GRCh38/hg 38	VUS	NR	NR
c.755C>T (p.Thr252Met)	NM_003494.3	g.71742844C>T	GRCh37/hg 19	VUS	NR	0000462990
c.342+1G>A	NM_003494.3	g.71730450G>A	GRCh37/hg 19	Pathogenic	PVS1, PM2, PP1 moderate, PP4 supporting,	0000712919

c.755C>T (p.Thr252Met)	NM_003494.3	g.71742844C>T	GRCh37/hg19	Pathogenic	PM3 supporting PM1 PM2 PP2 PP3 PP5/S	0000936143
c.1958delG (p.Gly653Valfs*3)	NM_003494.3	g.71780964del	GRCh37/hg19	Pathogenic	NR	0000632263
c.2190dupA (p.Pro731Thrfs*23)	NM_003494.4	g.71561779dup	GRCh38/hg38	NR	NR	NR
c.3597G>A (p.Trp1199*)	NM_003494.3	g.71825770G>A	GRCh37/hg19	Pathogenic	PVS1, PM2, PP4 moderate	0000712116
c.3944_3948delinsG (p.Glu1315Glyfs*29)	NM_003494.3	g.71611285_71611289delinsG	GRCh38/hg38	NR	NR	NR
c.4101G>A	NM_003494.4	g.71611560G>A	GRCh38/hg38	NR	NR	NR
c.6124C>T (p.Arg2042Cys)	NM_003494.3	g.71909727C>T	GRCh37/hg19	Pathogenic	PM3 very strong, PM2, PP4 moderate, PP1	0000450483

					supporting, PP3	
c.4431G>T (p.Trp1477Cys)	NM_003494.3	g.71643985G>T	GRCh38/hg 38	Pathogenic	NR	0000595803
c.5983_5984del (p.Ser1995*)	NM_003494.3	g.71681037_71681038del	GRCh38/hg 38	NR	NR	NR
c.457+2T>G (p.Ala115_Asp153)	NM_003494.3	g.71739053T>G	GRCh37/hg 19	Pathogenic	NR	0000449977
c.1663C>T (p.Arg555Trp)	NM_003494.3	g.71551631C>T	GRCh38/hg 38	Pathogenic	PM3 very strong, PM2, PP1 supporting, PP3, PP4 moderate	0000449995
c.1795_1799dup (p.Ala601Thrfs*28)	NM_003494.3	g.71553053_71553057dup	GRCh38/hg 38	Pathogenic	PVS1, PM2, PP4 moderate, PP1 supporting, PM3 supporting	0000449655

c.1834C>T (p.Gln612*)	NM_003494.3	g.71780222C>T	GRCh37/hg 19	Pathogenic	PVS1, PM3 strong, PM2, PP4 moderate, PP1 supporting	0000450495
c.3967C>T (p.Gln1323*)	NM_003494.3	g.71838438C>T	GRCh37/hg19	Pathogenic	PVS1, PM2, PP4 moderate, PM3 supporting	0000450017
c.1834C>T (p.Gln612*)	NM_003494.3	g.71780222C>T	GRCh37/hg19	Pathogenic	PVS1, PM3 strong, PM2, PP4 moderate, PP1 supporting	0000450016
c.4876delG (p.Val1626Tyrfs*8)	NM_003494.3	g.71887771del	GRCh37/hg19	Pathogenic	NR	0000450009
c.2643+1G>A (p.Tyr838_Thr881del)	NM_003494.3	g.71795213G>A	GRCh37/hg19	Pathogenic	PM3 very strong,	0000450005

c.3035G>A (p.Trp1012*)	NM_003494.3	g.71797732G>A	GRCh37/hg19	Pathogenic	PVS1 moderate, PP4 moderate, PP1 moderate	0000449720
c.4201dupA (p.Ile1401Asnfs*8)	NM_003494.3	g.71839804dup	GRCh37/hg19	Pathogenic	PVS1, PM2, PP1 supporting, PP4 supporting, PM3 supporting	0000449778
c.5296G>T (p.Glu1766*)	NM_003494.3	g.71894601G>T	GRCh37/hg19	Pathogenic	PVS1, PM2, PP4 moderate,	0000449812

c.1392dupA (p.Asp465Argfs*9)	NM_003494.3	g.71762436dup	GRCh37/hg19	Pathogenic	PM3 supporting PVS1, PM2, PP1 moderate, PP4 supporting, PM3 supporting	0000449641
c.3113G>A (p.Arg1038Gln)	NM_003494.3	g.71797810G>A	GRCh37/hg19	Likely pathogenic	NR	0000116997
c.4200dupC (p.Ile1401Hisfs*8)	NM_003494.3	g.71839803dup	GRCh37/hg19	Pathogenic	PVS1, PM3 strong, PM2, PP1 moderate, PP4 moderate	0000450246
c.4597-2A>G	NM_001130976.2	g.71886006A>G	GRCh37/hg19	Pathogenic	NR	NR
c.5525G>A (p.Gly1842Asp)	NM_003494.3	g.71896337G>A	GRCh37/hg19	Pathogenic	PP3, PM2, PM3	0000789499
c.2858dupT (p.Phe954Valfs*2)	NM_003494.3	g.71796997dup	GRCh37/hg19	Pathogenic	PVS1, PM2,	0000449692

c.4200delC (p.Ile1401Serfs*47)	NM_003494.3	g.71839803del	GRCh37/hg19	Pathogenic	PM3, PP4 supporting PVS1, PM2, PP4 moderate, PM3 supporting	0000450608
c.5594delG (p.Gly1865Alafs*101)	NM_003494.3	g.71896803del	GRCh37/hg19	Pathogenic	PVS1, PM3 strong, PM2, PP4 moderate, PP1 moderate	0000449605
c.4872delG (p.Glu1624Aspfs*10)	NM_003494.3	g.71887767del	GRCh37/hg19	Pathogenic	NR	0000449792
c.5434ins (5410–5433 tandem repeat at 5434)	NR	NR	NR	Pathogenic	NR	NR
c.5438T>C (p.Leu1813Pro)	NM_003494.3	g.71896250T>C	GRCh37/hg19	Pathogenic	NR	0000670484
c.5555T>C (p.Lys1852Pro)	NM_001130977.2	g.71896743T>C	GRCh37/hg19	Likely pathogenic	NR	NR

c.526C>T (p.Gln176*)	NM_003494.3	g.71740914C>T	GRCh37/hg19	Pathogenic	PVS1, PM3, PM2, PP4_mod	0000872016

Genomic coordinates are reported according to the GRCh37 and GRCh38 reference genome assemblies. **Abbreviations**: NR: Not reported; ID: Identifier; ACMG: American College of Medical Genetics and Genomics; PVS: Pathogenic very strong; PM: Pathogenic moderate; PP: Pathogenic supporting.