

Data updated 1 November 2016

CNNM4 mutations, Jalili syndrome type, and statistics (including cases with unreported mutations): statistics on sibships and patients.*

Origin	Total Cases Reported	Cases Examined	Families	Sibships	M	F	Gender n/a	Phenot ype	Mutations
MENA - Easter Medeterranean (including Iran and Turkey)									
Gaza Strip (GS)									
GS South (Gaza A)	31	29	1	18	17	14		A	c.599C>A;p.S200Y / ditto
Gaza City (Gaza B)	3	3	1	1	1	2		B	c.1813C>T;p.R605X / ditto
GS South (Rafah) (Gaza C)	1	1	1	1	1	0		A	-
Lebanon	3	3	1	2	2	1		A	c.707G>A;p.R236Q / ditto
Saudi Arabia	2	1	1	1			2	A	c.1484C>T; p.T495Ile / ditto
	2	1	1	1	2	0		A	c.[1474G>T]; p. C492 / ditto
Turkey	2	1	1	1	2	0		B	c.586T>C;p.S196P / ditto
	3	3	1	1	0	3		B	c.1781A>G (p.N594S) / ditto
Iran	4	4	1	2	3	1		?	c.1-?_1403+?del / ditto
	24	4	1	14	12	12		A	c.1091delG / ditto
MENA - N. Africa									
Algeria	3	3	1	1	2	1		A	c.189del; Asp63GluFs*12 / ditto
	2	1	1	1	-	-	2	A	c.1495G>A; p.[V499M] / ditto
Morocco	3	3	1	1	0	3	0	A	c.1682-1G>C; p.Glu561Glyfs*5. / ditto
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Total MENA	83	57	13	45	42	37	4		
Indo China									
China	1	1	1					?	c.896_897insT; p.A300CfsX22 / ditto
China (carrier)	1	1	1	1			1	A	c.47G>A;p.Arg16His
India	4	4	4	10	5	5		A	n/a
Pakistan	1	1	1	1	1	0		A	n/a
Europe									

Scotland	1	1	1	1	1	0	B	c.971T>C;p.L324P / c.1690T>C;p.O564*
Unreported (reviewed in Swizerland)	1	1	1	1	0	1	A	c.971T>C;p.L324P / ditto
Kosovan mutations	2	2	1	1	2	0	A	c.1312dupC;p.L438ProfX9 / ditto
	2	2	1	1	1	1	A	
	1	1	1	1	0	1	A	
	2	2	1	1	2	0	A	
	2	2	1	1	0	2	A	
	1	1	1	1	1	1	A	
<u>Kosovon mutation total*</u>	10	10	4	4	3	4		
<u>Poland</u>	3	3	1	1	3	0	?	c.1076T>C, p.(Leu359Pro)
Subtotal European Mutations	25	25	13	13	13	10	9	
North and South America								
Canada (New Foundland)	1	1	1	1			1	? c.1555C>T;p.R519* / ditto
USA: Portland, Oregon	1	1	1	1	1	0	A	c.1307delC, p.T436fs / c.C1690T, p.Q564X
Guatemala	5	5	1	1	5	0	?	c.2149C>T;p.Gln717X / c.62_145 del;L21HisfsX185
subtotal Americas	7	7	3	3	6		1	

* Mutations as published by 1 November 2016.

Ref: www.jalili.co.uk/cnm4/cnm4-js-byregion.xlsx

For any query related to this work, please contact: dr@jalili.co.uk

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[References](#)

[Statistics](#)

Reference (abbreviated) - See last column

Jalili and Smith (1989)¹, Parry et al (2009)²

Parry et al (2009)², Jalili IK (2010)⁵

Mutations unreported

Polok et al (2009)⁴ Family B

Abu-Safieh et al (2013)⁹

Lopez Torres et al (2015) (abstract)¹³

Parry et al (2009)²

Topçu et al (2016)¹⁷

Parry et al (2009)²

Rahimi-Aliabadi et al (2016)¹⁸

Coppieters et al (2014)¹¹

Prasad et al (2015)¹²

Jaouad et al²²

Wang et al (2015)¹⁴

Huang et al (2012)¹⁹

Purwar et al (2015)²⁰

Malik et al (2016)²¹

Parry et al (2009) ²

Polok et al (2009) ⁴ Family C

Parry et al (2009) ², Michaeladis et al (2004)
³

Polok et al (2009) ⁴ Family A

Zobor et al (2012) ⁶

Luder et al (2013) ⁸

Gerth-Kahlert et al (2013) ¹⁰

Kiessling et al (2016) ¹⁶

Wawrocka A et al (2017)

Doucette et al (2013) ⁷

Pennesi et al (2015) ¹⁵

Parry et al (2009) ²
