

Data updated 1 November 2016

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

Origin	Reported Cases	Cases Examined	Families	Sibships	M	F	Gender n/a	JS Type	Reference (abbreviated) - See last column
Gaza (Gaza A)	31	29	1	18	17	14		A	Jalili and Smith (1989) ¹ , Parry et al (2009) ²
Kosovo*	2	2	1	1	2	0		A	Parry et al (2009) ² , Michaeladis et al ³
Kosovo*	2	2	1	1	1	1		A	Polok et al (2009) ⁴ (Family A)
Gaza (Gaza B)	3	1	1	1	1	2		B	Parry et al (2009) ² , Jalili IK (2010) ⁵
Turkey	2	1	1	1	2	0		B	Parry et al (2009) ²
Iran	4	1	1	2	3	1		u/r	Parry et al (2009) ²
Guatemala	5	5	1	1	5	0		u/r	Parry et al (2009) ²
Scotland	1	1	1	1	1	0		B	Parry et al (2009) ²
Lebanon	3	3	1	2	2	1		A	Polok et al (2009) ⁴ Family B
Unreported	1	1	1	1	0	1		A	Polok et al (2009) ⁴ Family C
Kosovo*	1	1	1	1	0	1		A	Zobor et al (2012) ⁶
N. America	4	1	1	1	1	3		A	Doucette et al (2013) ⁷
Kosovo*	2	2	1	1	2	0		A	Luder et al (2013) ⁸
Saudi Arabia	2	1	1	1			2	A	Abu-Safieh et al (2013) ⁹
Kosovo*	2	2	1	1	0	2		A	Gerth-Kahlert et al (2013) ¹⁰
Algeria	3	3	1	1	2	1		A	Coppieters et al (2014) ¹¹
Algeria	2	1	1	1	-	-	2	A	Prasad et al (2015) ¹²
Saudi Arabia	2	1	1	1	2	0		A	Lopez Torres et al (2015) (abstract) ¹³
China	1	1	1	1			1	A	Wang H et al (2015) ¹⁴
USA: Portland, Oregon	1	1	1	1	1	0		A	Pennesi M et al (2015) ¹⁵
Kosovo*	1	1	1	1	1	1		A	Kiessling F et al (2016) ¹⁶
Turkey	3	3	1	1	0	3		B	Topçu et al (2016) ¹⁷
Iran	24	4	1	14	12	12		A	Rahimi-Aliabadi et al (2016) ¹⁸
Morocco	3	3	1	1	0	3		A	Jaouad CI, et al ²²
Poland	3	3	1	1	3	0		n/a	Wawrocka A et al ²³
Mutations reported in									
China (carrier)									
-									
Huang et al (2012), ¹⁹ carrier state									
Unreported Mutaions									

Gaza (Family Gaza C)	1	1	1	1	1	0	A	Jalili (2010) ³ - Gaza C
India	4	1	4	10	5	5	A	Purwar et al ²⁰
Pakistan	1	1	1	1	1	0	A	Malik et al (2016) ²¹
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Total per column	114	77	31	69	65	51	5	including unreported mutations
Total per column with reported mutations	108	74	25	57	58	46	5	
Total per column with unreported mutations	6	3	6	12	7	5	0	
Kosovan mutations	10	10	6	6	6	5	0	

Families with type 'A' (macular dystrophy)	22
Families with type 'B' (macular dystrophy)	4
Families with undocumented retinal phenotype	2

* Mutations published as at 31 August 2017

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

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