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]
Mutations reported in									
China (carrier)								-	Huang et al (2012),[19] carrier state
<u>Unreported Mutaions</u>									

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]
Total per column	111	74	30	68	62	51	5		including unreported mutations
Total per colomn with reported mutations	105	71	24	56	55	46	5		
Total per colomn 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)	21
Families with type 'B' (macular dystrophy)	4
Families with undocumented retinal phentype	2

* Mutations published as at 1 November 2016.

Ref: www.jalili.co.uk/cnnm4/cnnm4-js-statistics.xlsx

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