CNNM4 mutations, Jalili syndrome type, and statistics (including cases with unreported mutations) (as published by 1 April 2017)

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Year	Origin	novel	zygosity	Mutation 1 / Mutation 2	CF & additional associations	Reference (abbreviated) - See last column
2009	Gaza (Gaza A)	novel	homozygous	c.599C>A;p.S200Y / ditto	JS (CRD & AI)	Jalili and Smith (1989) [1], Parry et al (2009) [2],
2009	Kosovo	novel	homozygous	c.1312dupC;p.L438ProfX9 / ditto	JS (CRD & AI) plus: taurodontism of permanent	Parry et al (2009) [2], Michaeladis et al [3],
2009	Kosovo				JS (CRD & AI)	Polok et al (2009 - Family A) 4
2009	Gaza (Gaza B)	novel	homozygous	c.1813C>T;p.R605X / ditto	JS (CRD & AI)	Parry et al (2009) [2], Jalili (2010) [5]
2009	Turkey	novel	homozygous	c.586T>C;p.S196P / ditto	JS (CRD & AI)	Parry et al (2009) [2]
2009	Iran	novel	homozygous	c.1-?_1403+?del / ditto	JS (CRD & AI)	Parry et al (2009) [2]
2009	Guatemala	novel	compound heterozygous	c.2149C>T;p.Gln717X / c.62_145 del;L21HisfsX185	JS (CRD & AI)	Parry et al (2009) [2]
2009	Scotland	novel	compound heterozygous	c.971T>C;p.L324P / c.1690T>C;p.O564*	JS (CRD & AI)	Parry et al (2009) [2]
2009	Lebanon	novel	homozygous	c.707G>A;p.R236Q / ditto	JS (CRD & AI)	Polok et al (2009) [4]- Family B
2009	Unreported	novel	homozygous	c.971T>C;p.L324P / ditto	JS (CRD & AI)	Polok et al (2009) [4] - Family C
2012	Kosovo	known	homozygous	c.1312dupC;p.L438PfX9 / ditto;. ADDITIONAL: c.2033insC;p.Ile679AsnfsX21 (NF1)	JS (CRD & AI)	Zobor et al (2012) [6]
2013	N. America	novel	homozygous	c.1555C>T;p.R519* / ditto	JS (CRD & AI)	Doucette et al (2013) [7]
2013	Kosovo	known	homozygous	c.1312dupC;p.L438Pfs9X / ditto	JS (CRD & AI)	Luder et al (2013) [8]
2013	Saudi Arabia	novel	homozygous	c.1484C>T; p.T495Ile / ditto	JS (CRD & AI)	Abu-Safieh et al (2013) [9]
2013	Kosovo	known	homozygous	c.1312dupC;p.L438Pfs*9 / ditto	JS (CRD & AI)	Gerth-Kahlert et al (2013) [10]
2014	Algeria	novel	homozygous	c.189del; Asp63Glufs*12 / ditto	JS (CRD & AI)	Coppieters et al (2014) [11]
2015	Algeria	novel	homozygous	c.1495G>A; p.[V499M] / ditto	JS (CRD & AI)	Prasad et al (2015) [12]
2015	Saudi Arabia	novel	homozygous	c.[1474G>T]; p. C492 / ditto	JS (CRD & AI), mental deficiency	Lopez Torres et al (2015) (abstract) [13]

2015	China	novel	homozygous	c.896_897insT; p.A300CfsX22 / ditto	JS (CRD & AI)	Wang H et al (2015) [14]
2015	USA: Portland, Oregon	novel	compound heterozygous	c.1307delC, p.T436fs / c.C1690T, p.Q564X	JS (CRD & AI)	Pennesi M et al (2015) [15]
2016	Kosovo	novel	as compound heterozygou	c.1312dupC;p.L438PfsX9 / c.694_722del;p.lle232ProfsX80	JS (CRD & AI)	Kiessling F et al (2016) [16]
2016	Turkey	novel	homozygous	c.1781A>G (p.N594S) / ditto	JS (CRD & AI)	Topçu et al (2016) [17]
2016	Iran	novel	homozygous	c.1091delG / ditto	JS (CRD & AI)	Rahimi-Aliabadi et al (2016) [18]
2017	Morroco	novel	homozygous	c.1682-1G>C; p.Glu561Glyfs*5. / ditto	JS (CRD & AI)	Jaouad et al [22]

Mutations reported in a carrier status

2013	China	novel	Heterozygous, carrier	c.47G>A;p.Arg16His	JS (CRD & AI)	Huang Li et al (2013),[19] carrier state

Unreported mutations

JS (CRD & AI)

1989	Gaza singleton (Family Gaza C)	n/a		n/a	JS (CRD & AI)	Jalili (2010) [5] - Gaza C
2015	India	n/a	n/a	n/a	JS (CRD & AI) plus other ocular	Purwar et al [20]
2016	Pakistan	n/a	n/a	n/a	JS (CRD & AI)	Malik et al [21]

Number of novel mutations 21

Homozygous mutations 18

* Number of reported 6

** An existing novel mutation as a compound heterzygous

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