

CNNM4 mutations, Jalili syndrome - References

References

- 1 Jalili IK, Smith NJD. A progressive cone-rod dystrophy and amelogenesis imperfecta: a new syndrome. *J Med Genet.* 1988;25:738-40.
- 2 Parry DA, Mighell AJ, El-Sayed W, Shore RC, Jalili IK, Dollfus H, Bloch-Zupan A, Carlos R, Carr IM, Downey LM, Blain KM, Mansfield DC, Shahrabi M, Heidari, M, Aref P, Abbasi M, Michaelides M, Moore AT, Kirkham J, Inglehearn CF. Mutations in CNNM4 cause Jalili syndrome, consisting of autosomal-recessive cone-rod dystrophy and amelogenesis. *Am. J. Hum. Genet.* 2009; 84: 266-73.
- 3 Michaelides M, Bloch-Zupan A, Holder GE, Hunt DM, Moore AT. An autosomal recessive cone-rod dystrophy associated with amelogenesis imperfecta. *J Med Genet.* 2004;41(6):468-73.
- 4 Polok B, Escher P, Ambresin A, Chouery E, Bolay S, Meunier I, Nan F, Hamel C, Munier FL, Thilo B, Mégarbané A, Schorderet DF. Mutations in CNNM4 cause recessive cone-rod dystrophy with amelogenesis imperfecta. *Am J Hum Genet.* 2009; 84(2):259-65.
- 5 Jalili IK. Cone-rod dystrophy and amelogenesis imperfecta (Jalili syndrome): phenotypes and environs. *Eye* 2010; 24, 1659-68.
- 6 Zobor D, Kaufmann DH, Weckerle P, Sauer A, Wissinger B, Wilhelm H, Kohl S. Cone-rod dystrophy associated with amelogenesis imperfecta in a child with neurofibromatosis type 1. *Ophthalmic Genet.* 2012;33(1):34-8.
- 7 Doucette L, Green J, Black C, Schwartzentruber J, Johnson GJ, Galutira D, Young TL. Molecular genetics of achromatopsia in Newfoundland reveal genetic heterogeneity, founder effects and the first cases of Jalili syndrome in North America. *Ophthalmic Genet.* 2013;34(3):119-29.
- 8 Luder HU, Gerth-Kahlert C, Ostertag-Benzinger S, Schorderet DF. Dental phenotype in Jalili syndrome due to a c.1312 dupC homozygous mutation in the CNNM4 gene. *PLoS One.* 2013;23;8(10):e78529.
- 9 Abu-Safieh L, Alrashed M, Anazi S, Alkuraya H, Khan AO, Al-Owain M, Al-Zahrani J, Al-Abdi L, Hashem M, Al-Tarimi S, Sebai MA, Shamia A, Ray-Zack MD, Nassan M, Al-Hassnan ZN, Rahbeeni Z, Waheeb S, Alkharashi A, Abboud E, Al-Hazaa SA, Alkuraya FS. Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. *Genome Res.* 2013;23(2):236-47.
- 10 Gerth-Kahlert C, Seebauer B, Dold S, Hanson JV, Wildberger H, Spörri A, van Waes H, Berger W. Intra-familial phenotype variability in patients with Jalili syndrome. *Eye (Lond).* 2015;29(5):712-6.
- 11 Coppieters F, Van Schil K, Bauwens M, Verdin H, De Jaegher A, Syx D, Sante T, Lefever S, Abdelmoula NB, Depasse F, Casteels, de Ravel T, Meire F, Leroy BP, De Baere E. Identity-by-descent-guided mutation analysis and exome sequencing in consanguineous families reveals unusual clinical and molecular findings in retinal dystrophy. *Genet Med.* 2014;16(9):671-80.
- 12 Prasad MK, Geoffroy V, Vicaire S, Jost B, Dumas M, Le Gras S, et al. A targeted next-generation sequencing assay for the molecular diagnosis of genetic disorders with orodental involvement. *J Med Genet.* 2016;53(2):98-110.
- 13 Lopez Torres LT, Schorderet D, Valmaggia C, Todorova M. A novel mutation in CNNM4 (G492C) associated with Jalili Syndrome. *Acta Ophthalmologica*, 2015;93(S255). doi: 10.1111/j.1755-3768.2015.0606.
- 14 Wang H, Wang X2, Zou X, Xu S, Li H, Soens ZT, Wang K, Li Y, Dong F, Chen R, Sui R. Comprehensive Molecular Diagnosis of a Large Chinese Leber Congenital Amaurosis Cohort. *Invest Ophthalmol Vis Sci.* 2015;56(6):3642-55.
- 15 Pennesi M, Thomas A, Zhongqi GE, Che R. Meeting of the International Society for Genetic Eye Diseases & Retinoblastoma ISGEDR Halifax, Nova Scotia, Canada August 6-8, 2015
- 16 Kiessling F, Mitter D, Langmann T, Müller D, Tegetmeyer H. Novel Deletion in the CNNM4 Gene in Siblings with Jalili Syndrome. *Kiessling et al. Int J Ophthalmol Clin Res* 2016, 3(1):046
- 17 Topçu V, Alp MY, Alp CK, Bakır A, Geylan D, Yılmazoğlu MÖ. A new familial case of Jalili syndrome caused by a novel mutation in CNNM4. *Ophthalmic Genet.* 2016;12:1-6.
- 18 Rahimi-Aliabadi S, Daftarian N, Ahmadi H, Emamalizadeh B, Jamshidi J, Tafakhori A, et al. A Novel Mutation and Variable Phenotypic Expression in a Large Consanguineous Pedigree with Jalili Syndrome: *Eye (Lond).* 2016;30, 1424–32.
- 19 Huang L, Xiao X, Li S, Jia X, Wang P, Guo X, Zhang Q. CRX variants in cone-rod dystrophy and mutation overview. *Biochem Biophys Res Commun.* 2012;426(4):498-503.
- 20 Purwar P, Sareen S, Bhartiya K, Sayed Inayatullah SR, Bansal M, Chahal V, Gupta SK, Dixit J, Sheel V, Rai P. Jalili syndrome presenting with situs inversus totalis and keratoconus: the first case in Indian subcontinent. *Oral Surg Oral Med Oral Pathol Oral Radiol.* 2015;120(5):e210-8.
- 21 Malik TG, Khalil M, Shah SA, Shafiq MM. Jalili Syndrome (case report). *Pak J Ophthalmol* 2016;32(1):56-59.
- 22 Jaouad CI, Lyahyai J, Guaoua S, El Alloussi M, Zrhidri A, Doubaj Y, Boulanouar A, Sefiani A. Novel splice site mutation in CNNM4 gene in a family with Jalili syndrome. *Eur J Med Genet.* 2017; 60(5):239-244.

- 23** Wawrocka A, Walczak-Sztulpa J, Badura-Stronka M, Owecki M, Kopczynski P, Mrukwa-Kominek E, Skorczyk-Werner A, Gasperowicz P, Ploski R, Krawczynski MR. Am J Med Genet A. 2017; 173(8): 2280-2283.

Ref: www.jalili.co.uk/cnm4/cnm4-Jalilisynndrome-ref.xlsx