



Pachyonychia Congenita Project

Fighting for a cure. Connecting & helping patients. Empowering research.

Selected Articles for Other Genetic Disorders

There are over 600 articles available in the Published Research Articles section of the website at www.pachyonychia.org. The following select list includes articles on disorders which at times been misdiagnosed as Pachyonychia Congenita. Genetic testing is helping to differentiate and clarify Pachyonychia Congenita.

Cast

[Identification of a CAST Mutation in a Cohort Previously Misdiagnosed as Having Autosomal Recessive Pachyonychia Congenita](#). *JAMA Dermatology*. Dec 1 2015;151(12):1393-1394. Haber RM, Rose TH.

[Loss-of-function mutations in CAST cause peeling skin, leukonychia, acral punctate keratoses, cheilitis, and knuckle pads](#). *American Journal of Human Genetics*. Mar 5 2015;96(3):440-447. Lin Z, Zhao J, Nitoiu D, Scott CA, Plagnol V, Smith FJD, Wilson NJ, Cole C, Schwartz ME, McLean WHI, Wang H, Feng C, Duo L, Zho EY, Ren Y, Dai L, Chen Y, Zhang J, Xu X, O'Toole EA, Kelsell DP, Yan Y.

Olmsted Syndrome

[Recurrent heterozygous missense mutation, p.Gly573Ser, in the TRPV3 gene in an Indian boy with sporadic Olmsted syndrome](#). *British Journal of Dermatology*. 17 Jun 2012;167:440-442. Lai-Cheong JE, Sethuraman G, Ramam M, Stone K, Simpson MA, McGrath JA.

Expanding the Phenotypic Spectrum of Olmsted Syndrome. *Journal of Investigative Dermatology*. Nov 2015;135(11):2879-2883. Wilson NJ, Cole C, Milstone LM, Kiszewski AE, Hansen CD, O'Toole EA, Schwartz ME, McLean WHI, Smith FJD.

Other

[Mutations in GJB6 causing phenotype resembling pachyonychia congenita](#). *British Journal of Dermatology*. May 2015;172(5):1447-1449. Hale GI, Wilson NJ, Smith FJ, Wylie G, Schwartz ME, Zamiri M.

[Recessive mutations in the gene encoding frizzled 6 cause twenty nail dystrophy--expanding the differential diagnosis for pachyonychia congenita](#). *Journal of Dermatological Science*. Apr 2013;70(1):58-60. Wilson NJ, Hansen CD, Azkur D, et al.