

Yale Center for Mendelian Genomics: List of Publications

1. Sadowski CE, Svjetlana Lovric, Shazia Ashraf, Werner L. Pabst, Heon Yung Gee, Stefan Kohl, Susanne Engelmann, Virginia Vega-Warner, Humphrey Fang, Jan Halbritter, Michael J. Somers, Weizhen Tan, Shirlee Shril, Inès Fessi, Richard P. Lifton, Detlef Bockenhauer, Sherif El-Desoky, Jameela A. Kari, Martin Zenker, Markus J. Kemper, Dominik Mueller, Hanan M. Fathy, Neveen A. Soliman, the SRNS Study Group and Friedhelm Hildebrandt¹, *J Am Soc Nephrol*. 2014 Oct 27. pii: ASN.2014050489. [Epub ahead of print] PMID: 25349199.
2. Boyden LM, Craiglow BG, Zhou J, Hu R, Loring EC, Morel KD, Lauren CT, Lifton RP, Bilguvar K, Paller AS, Choate KA. Dominant De Novo Mutations in GJA1 Cause Erythrokeratoderma Variabilis Et Progressiva, without Features of Oculodentodigital Dysplasia. *J Invest Dermatol*. 2014 Nov 14. doi: 10.1038/jid.2014.485. [Epub ahead of print] PMID:25398053 [PubMed - as supplied by publisher]
3. Vilarinho S, Choi M, Jain D, Malhotra A, Kulkarni S, Pashankar D, Phatak U, Patel M, Bale A, Mane S, Lifton RP, Mistry PK. Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. *J Hepatol*. 2014 Nov; 61(5):1056-63. doi: 10.1016/j.jhep.2014.06.038. Epub 2014 Jul 10. PMID: 25016221.
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5. Romberg N, Al Moussawi K, Nelson-Williams C, Stiegler AL, Loring E, Choi M, Overton J, Meffre E, Khokha MK, Huttner AJ, West B, Podoltsev NA, Boggon TJ, Kazmierczak BI, Lifton RP. Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. *Nat Genet*. 2014 Oct;46(10):1135-9. doi: 10.1038/ng.3066. Epub 2014 Sep 14. PMID: 25217960. PMCID: PMC4177367
6. Caglayan AO, Comu S, Baranoski JF, Parman Y, Kaymakçalan H, Akgumus G, Caglar C, Dolen D, Omay ZE, Harmanci AS, Mishra K, Freeze HH, Yasuno K, Bilguvar K, Gunel M. NGLY1 Mutation Causes Neuromotor Impairment, Intellectual Disability, and Neuropathy. *European Journal of Medical Genetics Eur J Med Genet*. 2014 Sep 9. pii: S1769-7212(14)00168-2. doi: 10.1016/j.ejmg.2014.08.008. PMID: 25220016.
7. Caglayan AO, Jacob F. Baranoski, Fesih Aktar, Wengi Han, Beyhan Tuysuz, Aslan Guzel, Bulent Guclu, Hande Kaymakçalan, Berrin Aktekin, Gozde Tugce Akgumus, Phillip B. Murray, Emine Z. Erson-Omay, Caner Caglar, Mehmet Bakircioglu, Yildirim Bayezit Sakalar, Ebru Guzel, Nihat Demir, Oguz Tuncer, Senem Senturk, Baris Ekici, Frank J. Minja, Nenad Sestan, Katsuhito Yasuno. Brain Malformations Associated With Knobloch Syndrome—Review of Literature, Expanding Clinical Spectrum, and Identification of Novel Mutations. *Pediatric Neurology*. Volume 51, Issue 6, December 2014, Pages 806–813.e8. doi:10.1016/j.pediatrneurol.2014.08.025
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