

Dr. Shehla Mohammed

Relevant key publications:

- Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Bengani H, et al **Genet Med**. 2017 Feb 2. doi: 10.1038/gim.2016.211. [Epub ahead of print]
- Prevalence and architecture of de novo mutations in developmental disorders. Deciphering Developmental Disorders Study **Nature**. 2017 Feb 23;542(7642):433-438. doi: 10.1038/nature21062. Epub 2017 Jan 25.
- The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Redin C **Nat Genet**. 2017 Jan;49(1):36-45. doi: 10.1038/ng.3720. Epub 2016 Nov 14.
- Risk Factors for Severe Renal Disease in Bardet-Biedl Syndrome. **J Am Soc Nephrol**. Forsyth et al 2017 Mar;28(3):963-970. doi: 10.1681/ASN.2015091029. Epub 2016 Sep 22.
- Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL) Sahar Elouej DOI: 10.1016/j.**Metabolism** 2017.03.011
- An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Lopez RJ **Sci Signal**. 2016 Jul 5;9(435):ra68. doi: 10.1126/scisignal.aad9813.
- Advances in Genetic Testing for Hereditary Cancer Syndromes. Thomas E, Mohammed S. **Recent Results Cancer Res**. 2016;205:1-15. doi: 10.1007/978-3-319-29998-3_1.
- Deep phenotyping of 89 xeroderma pigmentosum patients reveals unexpected heterogeneity dependent on the precise molecular defect. Fassihi et al **Proc Natl Acad Sci U S A**. 2016 Mar 1;113(9):E1236-45. doi: 10.1073/pnas.1519444113. Epub 2016 Feb 16
- A Distinct Genotype of XP Complementation group A: Surprisingly Mild Phenotype Highly Prevalent in Northern India/ Pakistan/Afghanistan. Journal of Investigative Dermatology: Sethi et al and XP team 2016 **J Invest Dermatol**. 2016 Apr;136(4):869-72. doi: 10.1016/j.jid.2015.12.031. Epub 2015 Dec 29.
- EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Byrn S et al: **Brain** 2016 Mar;139(Pt 3):765-81. doi: 10.1093/brain/awv393.
- A de novo, heterozygous, loss-of-function mutations in *SYNGAP1* cause a syndromic form of intellectual disability Parker MJ et al, **AJMG Part A** Volume 167, Issue 10, 2231–2237, October 2015
- Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Tummala H **J Clin Invest**. 2015 May;125(5):2151-60. doi: 10.1172/JCI78963. Epub 2015 Apr 20.

- Large-scale discovery of novel genetic causes of developmental disorders. The Deciphering Developmental Disorders Study. **Nature**. 2014 Dec 24. doi: 10.1038/nature14135.
- A new direction for prenatal chromosome microarray testing: software-targeting for detection of clinically significant chromosome imbalance without equivocal findings. Ahn JW, et al. *PeerJ*. 2014 Apr 22;2:e354. doi: 10.7717/peerj.354. eCollection 2014.
- Genotype-phenotype correlation in Bardet-Biedl Syndrome Forsythe et al (2012)**J Med Genet** 49, S71
- Genetic predictors of cardiovascular morbidity in Bardet-Biedl syndrome. Forsythe E, **Clin Genet**. 2014 Mar 10. doi: 10.1111/cge.12373.
- Realising Genomics in Clinical Practice. **PHG Foundation** (2014). ISBN 978-1-907198-15-1
- Array CGH testing for learning disability- when is it worth it? Gurdeep Sagoo, Shehla Mohammed **PHG publication** (2014) <http://www.phgfoundation.org/file/16397/>
- Reduced dosage of ERF causes complex craniosynostosis in humans and mice and links ERK1/2 signaling to regulation of osteogenesis. Twigg SR et al **Nat.Genet**. 2013 Mar;45(3):308-13.
- Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Cullup T, et al **Nat Genet**. 2013 Jan;45(1):83-7. doi: 10.1038/ng.2497. Epub 2012 Dec 9.
- Mutations in the gene PRRT2 cause paroxysmal kinesigenic dyskinesia with infantile convulsions. Lee HY et al **Cell Rep**. 2012 Jan 26;1(1):2-12. doi: 10.1016/j.celrep.2011.11.001. Epub 2011
- Mutation of the RAD51C gene in a Fanconi anemia-like disorder. Fiona Vaz et al **Nature Genetics** 2010 May 42(5):368-9
- Congenital leptin deficiency is associated with severe early-onset obesity in humans Montague et al **Nature** 387(6636):903-8 July 1997

85 key publications including original articles in: Nature, PNAS, British Medical Journal, American Journal of Medical Genetics, Journal of Medical Genetics, European Journal of Medical Genetics, Human Mutation, British Journal of Cancer, Genes Chromosome and Cancer, Disease Markers as well as book chapters

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