Dr Schaida Schirwani selected publications

1. Cragg, A., et al., **Schirwani, S., et al.**, *Early onset basal cell carcinoma: Consider Bazex-Dupre-Christol syndrome.* Eur J Med Genet, 2025. **75**: p. 105004.
2. **Schirwani, S., et al.**, *Coexistence of multiple self-healing squamous epithelioma and features of Loeys-Dietz syndrome caused by a pathogenic missense variant in the kinase domain of TGFBR1.* Clin Exp Dermatol, 2024. **49**(6): p. 665-669.
3. O'Brien, A., et al., **Schirwani, S., et al.**, *Mosaic Muir Torre Syndrome: Keratoacanthoma as a Piece of the Puzzle.* Am J Dermatopathol, 2024. **46**(3): p. 162-166.
4. **Schirwani, S., et al.**, *Familial Bainbridge-Ropers syndrome: Report of familial ASXL3 inheritance and a milder phenotype.* Am J Med Genet A, 2023. **191**(1): p. 29-36.
5. Williams, S.T., et al., **Schirwani, S., et al.**, *SDHC phaeochromocytoma and paraganglioma: A UK-wide case series.* Clin Endocrinol (Oxf), 2022. **96**(4): p. 499-512.
6. **Schirwani, S., et al.**, *Amniotic band sequence in vascular Ehlers-Danlos Syndrome (EDS): Experience of the EDS National Diagnostic Services in the UK.* Eur J Med Genet, 2022. **65**(10): p. 104592.
7. **Schirwani, S., et al.**, *Homozygous intronic variants in TPM2 cause recessively inherited Escobar variant of multiple pterygium syndrome and congenital myopathy.* Neuromuscul Disord, 2021. **31**(4): p. 359-366.
8. **Schirwani, S., et al.**, *Homozygosity for the pathogenic RET hotspot variant p.Cys634Trp: A consanguineous family with MEN2A.* Eur J Med Genet, 2021. **64**(2): p. 104141.
9. **Schirwani, S., et al.**, *Expanding the phenotype of ASXL3-related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in ASXL3.* Am J Med Genet A, 2021. **185**(11): p. 3446-3458.
10. Jalal, M., **S. Schirwani**, and K.D. Bardhan, *The Imbalance in Medico-Legal Cover Awareness and Uptake Between Overseas Junior Doctors and Local Graduates in the NHS.* Cureus, 2021. **13**(2): p. e13336.
11. **Schirwani, S., et al.**, *DSE associated musculocontractural EDS, a milder phenotype or phenotypic variability.* Eur J Med Genet, 2020. **63**(4): p. 103798.
12. **Schirwani, S., et al.**, *Mosaicism in ASXL3-related syndrome: Description of five patients from three families.* Eur J Med Genet, 2020. **63**(6): p. 103925.
13. **Schirwani, S.** and J. Campbell, *Genetics for paediatric radiologists.* Pediatr Radiol, 2020. **50**(12): p. 1680-1690.
14. **Schirwani, S., et al.**, *Duplications of GPC3 and GPC4 genes in symptomatic female carriers of Simpson-Golabi-Behmel syndrome type 1.* Eur J Med Genet, 2019. **62**(4): p. 243-247.
15. **Schirwani, S., et al.**, *Exploring the association between SRPX2 variants and neurodevelopment: How causal is it?* Gene, 2019. **685**: p. 50-54.

16. Balasubramanian, M., et al., **Schirwani, S., et al.**, *Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature*. Bone, 2019. **121**: p. 191-195.
17. **Schirwani, S., et al.**, *Expanding the molecular basis and phenotypic spectrum of ZDHHC9-associated X-linked intellectual disability.* Am J Med Genet A, 2018. **176**(5): p. 1238-1244.
18. **Schirwani, S.**, K. Smith, and M. Balasubramanian, *Clinical and molecular characterization of the first familial report of 1p32 microdeletion*. Clin Dysmorphol, 2018. **27**(2): p. 36-41.
19. **Schirwani, S., et al.**, *Carbamazepine Improves Apneic Episodes in Congenital Central Hypoventilation Syndrome (CCHS) With a Novel PHOX2B Exon 1 Missense Mutation*. J Clin Sleep Med, 2017. **13**(11): p. 1359-1362.
20. Balasubramanian, M. and **S. Schirwani**, *ASXL3-Related Disorder*, in *GeneReviews((R))*, M.P. Adam, et al., Editors. 1993: Seattle (WA).