Bainbridge-Roper Syndrome ASXL3

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De novo truncating and splicing pathogenic variants in the Additional Sex Combs-Like 3 (ASXL3) gene are known to cause neurodevelopmental delay, intellectual disability, behavioral difficulties, hypotonia, feeding problems and characteristic facial features. We previously reported 45 patients with ASXL3-related disorder including three individuals with a familial variant. Here we report the detailed clinical and molecular characteristics of these three families with inherited ASXL3-related disorder. First, a father and son with c.2791_2792del p.Gln931fs pathogenic variant. The second, a mother, daughter and son with c.4534C > T, p.Gln1512Ter pathogenic variant. The third, a mother and her daughter with c.4441dup, p.Leu1481fs maternally inherited pathogenic variant. This report demonstrates intrafamilial phenotypic heterogeneity and confirms heritability of ASXL3-related disorder.