Abstract

BEST1-mediated retinal disease in Norway

Kuopio, July 6th 2024

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Inherited retinal disease (IRD) is a leading cause of debilitating low vision in children and adolescents. In 2018 Holtan et al. found *BEST1* to be the third most common gene associated with IRD in Norway. The purpose of this study was to clinically and genetically characterize patients with a *BEST1*-mediated retinal dystrophy registered in the registry of IRD, Oslo University Hospital. In total, 78 patients were suspected of having a BEST1-related dystrophy, while 60 were genetically confirmed cases and two patients had a confirmed first-degree relative and were therefore included. The prevalence in South-Eastern Norway was 1: 53 000, while the prevalence is reported to be around 1 : 127 000 in other populations. Of the 62 patients (of 49 families), the clinical distribution demonstrated that 69% had dominant Best's vitelliform macular dystrophy (BVMD), 6% had AD vitreoretinochoroidopathy (ADVIRC) and 24% had AR bestrophinopathy (ARB). Mean age of first symptoms was 20 with a range from 1 to 70 years. In total 17 distinct pathogenic variants in the *BEST1* were identified of which *c.403G>A* was the most numerous variant identified in 17 of 62 patients. We report three novel variants. The study includes the largest data collection of *BEST1*-mediated retinal dystrophy in Norway, providing knowledge paramount for future inclusion in clinical treatment studies.