Biallelic Optic Atrophy (*OPA1*) Related Disorder – Case Report and Literature Review

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Introduction: The most common hereditary optic neuropathy, dominant optic atrophy (DOA), is inherited in an autosomal dominant pattern. Clinically, it presents as a progressive decrease in central visual acuity, central and later peripheral visual field defects, and retinal ganglion cells loss. Biallelic inheritance leads to a more severe disease usually referred to as Behr syndrome.

Methods: This is a case report focuses on a family with Biallelic Optic Atrophy 1 (*OPA1*).

Results/case report: The proband is a 17-month-old child with a severe phenotype and two variants in the *OPA1* gene. The symptoms that he presented with were progressive vision loss, congenital nystagmus, progressive ataxia, and optic atrophy. Genetic testing showed two likely pathogenic variants in his *OPA1* gene: one of which was inherited maternally, c.2287del (p.Ser763Valfs*15), and the other was inherited paternally, c.1311A>G (p.Ile437Met). The first variant, c.2287del, is predicted to be pathogenic and likely to cause DOA. In contrast, c.1311A>G, on its own, is considered asymptomatic but has been reported in patients with the DOA phenotype and is presumed to act as a phenotypic modifier. On follow-up, the child developed a multitude of symptoms including profound vision impairment, metabolic strokes, and intractable seizures. Upon literature review, twenty-one cases of biallelic *OPA1*-related Behr syndrome have been previously reported.

Conclusion: An early-onset, severe ocular phenotype and associated systemic features, seem to be hallmarks of the disease.