Severe SOPH syndrome due to a novel NBAS mutation in a 27-year-old woman-Review of this pleiotropic, autosomal recessive disorder: Mystery solved after two decades

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Abstract

Autosomal recessive SOPH syndrome was first described in the Yakuts population of Asia by Maksimova et al. in 2010. It arises from biallelic pathogenic variants in the NBAS gene and is characterized by severe postnatal growth retardation, senile facial appearance, small hands and feet, optic atrophy with loss of visual acuity and color vision, and normal intelligence (OMIM #614800). The presence of Pelger-Huet anomaly in this disorder led to its name as an acronym for Short stature, Optic nerve atrophy, and Pelger-Huet anomaly. Recent publications have further contributed to the characterization of this syndrome through additional phenotype-genotype correlations. We review the clinical features described in these publications and report on a 27-year-old woman with dwarfism with osteolysis and multiple skeletal problems, minor anomalies, immunodeficiency, diabetes mellitus, and multiple secondary medical problems. Her condition was considered an unknown autosomal recessive disorder for many years until exome sequencing provided the diagnosis by revealing a founder disease-causing variant that was compound heterozygous with a novel pathogenic variant in NBAS. Based on the major clinical features of this individual and others reported earlier, a revision of the acronym is warranted to facilitate clinical recognition.

Keywords

Author Keywords: combined immunodeficiency; diabetes mellitus; dwarfism and osteolysis; long survival; novel NBAS variant; SOPH syndrome
KeyWords Plus: ACUTE LIVER-FAILURE; AMPLIFIED SEQUENCE GENE; DEFICIENCY; ONSET; BONE

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