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WES reveals Novel Heterozygous NBAS Gene Mutations in a patient with SOPH syndrome associated with Fanconi Syndrome: A Case Report

Thong Jia Yean

University Malaya Medical Centre, Malaysia

Variations in the NBAS gene is known to cause a spectrum of phenotypes ranging from isolated recurrent acute liver failure (RALF) to a multisystem presentation known as SOPH syndrome. Patients with SOPH present with optic atrophy, acute liver failure, short stature, and Pelger-Huet anomaly. We report the presence of a novel pair of biallelic heterozygous mutation (c.5139-5T>G and c.2203-2A>G) in the NBAS gene of a patient with SOPH syndrome. A 9-year-old patient was clinically diagnosed with SOPH following clinical laboratory analyses. Current interventions for managing the disease encompass IVIG, methylprednisolone, calcium, and vitamin D administration. Whole-exome sequencing (WES) results showed two mutations: c.5139-5T>G and c.2203-2A>G, in the NBAS gene, which had not been previously reported. Notably, we hypothesize that NBAS mutations could potentially contribute to the development of Fanconi syndrome, a clinical finding reported in our patient. Our study also supports the renaming of SOPH to SOPHIA to allow early detection and effective treatment.

Keywords: Short Stature, Optic Atrophy, Pelger-Huet Anomaly, Neuroblastoma Amplified Sequence, Whole-Genome Sequencing, Myopia, Case Report, Acute Liver Failure, Fanconi Syndrome.

Biography

Thong Jia Yean completed my MBBS degree from Shanghai Medical College, Fudan University in 2020. At the present moment, Thong Jia Yean serve as a house officer at University Malaya Medical Centre, a university hospital located in Kuala Lumpur, Malaysia.