Alazami Syndrome

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Alazami syndrome is a rare autosomal recessive neurodevelopmental disorder due to loss-of-function variants in the La ribonucleoprotein 7 (LARP7) gene. Children with Alazami syndrome are most often affected by a combination of primordial dwarfism, intellectual disability, and distinctive facial features. Previous cases have been primarily found in consanguineous families from the Middle East, Asia, and North Africa. We present a 21-month-old Caucasian male from the Midwest United States with nonconsanguineous parents who presented with frequently reported findings of unusual facial features, poor growth, cardiac and genitourinary findings, and developmental delay; less-frequently reported findings, including transient erythroblastopenia of childhood (TEC) and immune deficiency; and never-before reported findings of periventricular nodular heterotopia and stroke. He developed stroke during a hospitalization for Hemophilus influenzae meningitis. The possible contributions of LARP7 to TEC, immune deficiency, brain malformation, and stroke are discussed. Guidelines for the care of Alazami patients are proposed.