Alazami Syndrome

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Alazami syndrome (AS) is an autosomal recessive condition characterized by the cardinal features of severe growth restriction, moderate to severe intellectual disability, and distinctive facial features. Biallelic pathogenic variants of the LARP7, encoding a chaperone of 7SK noncoding RNA, is implicated in this disease. There are <35 reported cases in the literature. All reported cases share the same three cardinal features of the syndrome. Herein, we report on 12 patients with a confirmed diagnosis of AS from eight unrelated families. The cohort shares the same key feature of the syndrome. Moreover, we report additional phenotypic features, including genito-renal anomalies, ophthalmological abnormalities, and congenital heart disease. Whole-exome sequencing was used in all reported cases, implicating a clinical under-recognition of the syndrome. This report further expands the clinical and molecular characteristics of Alazami syndrome.