Current Pediatric Research

Genotype and phenotype studies of Lowe syndrome in three families in Taiwan

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Abstract

Lowe syndrome is a rare X-linked disease characterized by the classic triad of congenital cataracts, intellectual disabilities, and renal tubular dysfunction. We reported four patients among three families diagnosed with Lowe syndrome at Changhua Christian Children's Hospital in Taiwan. Age at molecular diagnosis varied between 0.5 and 2.5 years. Three novel mutations (c.1853-1854insA, c.2309-2312delATTG, and c.1501A>T) found in our study were located in different exons. No particular hotspots were found among cases reported in Taiwan. All patients presented with a complete oculocerebrorenal phenotype with decelerated growth after infancy, congenital cataract and hypotonia, psychomotor delay and behavioral problems. However, a higher prevalence of hypercalciuria, potassium and phosphate wasting was observed. All patients developed renal insufficiency and progressive rickets, ended up with multiple fractures since early childhood and kyphoscoliosis before adolescence. Renal biopsy was performed in all patients at the time of diagnosis, which offered valuable information: Atrophic or dilated tubules was the earliest sign noted in all patients whereas interstitial fibrosis occurred later in the elder children. Swollen mitochondria with abnormal cristae were also seen in 75% of the patients, indicating that pleiotropy in Lowe syndrome can be a result of not only single enzymopathy but also secondary mitochondrial dysfunction. Overall, in our study and literature review, individuals in Taiwan are prone to more severe tubulopathy and thus, a shorter stature (height: −6.1 to −8.7 standard deviation) with worsen bone health, limited activities, and poor quality of life.

Biography

Professor Mei-Chyn Chao graduated from Kaohsiung Medical University, Taiwan. She completed residency in Pediatrics in Kaohsiung Medical University Hospital. Besides, she has visited Teikyo University and Prenatal Diagnosis Laboratory of New York City University for medical genetic training. She has been the chief director of Department of Pediatrics, Department of Cytogentics Laboratory, and Genetic Counseling Center in Kaohsiung Medical University Hospital. In her decades of medical career, she is dedicated in the diagnosis and care for families with rare disease. At present, she is the chief director of Pediatric Genetics and Metabolism at Changhua Christian Children's Hospital in Taiwan.

Publication

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