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Clinical Features of Lowe Syndrome Confirmed by a OCRL1 Gene Mutation (c.2083C>T) in a Korean Child

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Lowe syndrome (MIM 309000), also known as oculocerebrorenal syndrome of Lowe (OCRL), is a rare X-linked multi-system disorder characterized by the presence of developmental abnormalities affecting the eye, the nervous system, and the kidney. The syndrome is caused by pathogenic DNA variations in the X-linked OCRL1 gene. Bilateral cataracts and severe hypotonia are present at birth. Lowe syndrome can clinically be suspected in males who show the typical phenotype of deep-set small eyes, frontal bossing, elongated face, bilateral dense congenital cataracts, infantile congenital hypotonia, delayed development, and proximal renal tubular dysfunction. We report the first case of a patient initially characterized by delayed development and proteinuria and genetically diagnosed with OCRL1 gene mutation (c.2083C>T) in Korean children.

Key Words: 로웨 증후군, OCRL1 유전자 변이, 소아
Lowe syndrome, Oculocerebrorenal syndrome, OCRL1 Gene mutation