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Excerpta Abstract
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Lowe Syndrome: Neuroimaging Spectrum Including Rare Craniocervical Junction Dysplasia in an Adult Patient

Huda Zoori, The University of Texas Health Science Center Houston

Clinical History

A 42-year-old male with genetically confirmed Lowe syndrome (oculocerebrorenal syndrome) is being followed secondary to to chronic epilepsy and developmental delay. His past medical history includes Fanconi syndrome diagnosed at 6 months of age, chronic kidney disease stage 3, congenital cataracts (removed in infancy), glaucoma (surgery in 2012), and moderate intellectual disability. The patient also has stereotypic behaviors, partial hearing loss, multiple sebaceous cysts, and mild scoliosis. Family history revealed a carrier mother and an affected half-brother, consistent with X-linked inheritance.

Imaging Findings

Brain MRI (non-contrast) demonstrates extensive bilateral periventricular T2/FLAIR hyperintensities with innumerable small cystic spaces, suggestive of chronic leukoencephalopathy and/or dilated Virchow–Robin spaces. The craniocervical junction shows dysplastic dens and flattened clivus with upper cervical canal narrowing and associated CSF effacement.

Discussion

Lowe syndrome is a rare X-linked recessive multisystem disorder caused by pathogenic variants in the OCRL1 gene, encoding the inositol-5-phosphatase OCRL-1. Loss of this enzyme disrupts phosphatidylinositol (4,5)-bisphosphate metabolism, leading to abnormal endosomal trafficking and actin regulation. The prevalence is estimated at 1 in 500,000 in the general population. The classic triad includes dense congenital bilateral cataracts, intellectual impairment, and renal tubular dysfunction with slowly progressive renal failure. Other features include development delay, seizures, glaucoma, keloids, areflexia/hypotonia, dental anomalies, subcutaneous nodules, behavioral abnormalities and arthropathy, which can be observed in about 50 % of adult patients.

Neuroimaging findings typically include ventriculomegaly and periventricular/deep white matter T2 hyperintensities with associated cystic changes, often stable in size and distribution. Other findings more variable and non-specific are tigroid pattern with hypointense radially oriented stripes within the hyperintense cerebral white matter on T2-weighted images, brain atrophy, cerebellar hypoplasia, pachygyria, polymicrogyria, aberrant neuronal migration and subependymal cysts. Musculoskeletal implications are less described and include osteopenia, swollen/enlarged joints, scoliosis rickets, joint laxity, arthropathy and cervical abnormalities.

Teaching Point

Recognition of the oculocerebrorenal triad and distinctive MRI findings—including periventricular cystic leukoencephalopathy, and craniocervical junction dysplasia—broadens the imaging spectrum of Lowe syndrome. Identification of these features, particularly clival and odontoid dysplasia with canal stenosis, highlights the importance of multisystem evaluation and multidisciplinary management to mitigate neurological and renal complications in long-term survivors.

References

None

Figures

None