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Lowe Syndrome (Oculocerebrorenal Syndrome of Lowe): A Case Report of Two Brothers from India

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ABSTRACT Lowe syndrome (oculo-cerebro-renal syndrome of Lowe) is a rare X-linked recessive disorder characterized by the involvement of eyes, brain and kidneys. It is caused by the deficiency of enzyme phosphatidylinositol 4, 5-bisphosphate 5-phosphatase, which is required for the intracellular trafficking, second messengers and for the other aspects of cellular metabolism. The gene coding for this enzyme, OCRL1, has been localised at Xq25-q26.1 and mutations in it are reported to cause Lowe Syndrome. In this article we report two male siblings, from North India, with Lowe syndrome.

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