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# Oculocerebrorenal Syndrome

National Cancer Institute

## Source

National Cancer Institute. *Oculocerebrorenal Syndrome*. NCI Thesaurus. Code C84940.

An X-linked inherited multisystem metabolic disorder caused by mutations in the OCRL1 gene. The main manifestations of this disorder result from involvement of the eyes, nervous system and kidneys. Signs and symptoms include congenital cataracts, infantile glaucoma, intellectual disability, hypotonia, aminoaciduria and renal tubular dysfunction.