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Cataract-nephropathy-encephalopathy syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Cataract-nephropathy-encephalopathy syndrome. ORPHA:1380*

Cataract - nephropathy - encephalopathy syndrome describes a lethal combination of manifestations including short stature, congenital cataracts, encephalopathy with epileptic fits, and postmortem confirmation of nephropathy (renal tubular necrosis). The combination of cataract - nephropathy - encephalopathy has been described in 2 female infant children of first cousin parents. The infants did not survive beyond 4 and 8 months respectively. There have been no further descriptions in the literature since 1963.