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Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome. ORPHA:3390*

Proximal tubulopathy-diabetes mellitus-cerebellar ataxia syndrome is characterised by onset of proximal tubulopathy in the first year of life, followed by progressive development during childhood of skin anomalies (erythrocytosis and abnormal pigmentation), blindness, osteoporosis, cerebellar ataxia, mitochondrial myopathy, deafness and diabetes mellitus.