Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome. ORPHA:79118

A syndrome associating neonatal diabetes, congenital hypothyroidism, congenital glaucoma, hepatopathy evolving to fibrosis and polykystic kidneys has been described in two sibs. Minor facial anomalies were also observed. Two other families presented incomplete forms of this syndrome. Mutations in GLIS3 encoding for the transcription factor GLI similar 3 seem to be responsible of the syndrome.