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# Hepatic fibrosis-renal cysts-intellectual disability syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Hepatic fibrosis-renal cysts-intellectual disability syndrome. ORPHA:2031*

Hepatic fibrosis-renal cysts-intellectual disability syndrome is a rare, syndromic intellectual disability characterized by early developmental delay with failure to thrive, intellectual disability, congenital hepatic fibrosis, renal cystic dysplasia, and dysmorphic facial features (bilateral ptosis, anteverted nostrils, high arched palate, and micrognathia). Variable additional features have been reported, including cerebellar anomalies, postaxial polydactyly, syndactyly, genital anomalies, tachypnea. There have been no further descriptions in the literature since 1987.