

DISEASE:
Senior-Boichis syndrome

NAME:	Senior-Boichis syndrome
DESCRIPTION:	A rare ciliopathy characterized by the association of nephronophthisis and liver fibrosis. Renal manifestations include chronic renal failure, polyuria, polydipsia, anemia, as well as increased echogenicity on renal ultrasound and interstitial fibrosis and tubular dilation on biopsy. Hepatic involvement manifests as hepatosplenomegaly with extensive fibrosis, destruction of the bile ducts, and cholestasis. Mild psychomotor retardation and ocular symptoms, such as strabismus, nystagmus, retinal degeneration, and anisocoria, have been reported in some patients.
ORPHACODE:	84081
SYNONYMS:	Boichis disease Nephronophthisis-hepatic fibrosis syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	DCDC2 TMEM67
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14
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