DISEASE:
Usher syndrome type 2

NAME:	Usher syndrome type 2
DESCRIPTION:	A rare ciliopathy characterized by congenital moderate-to-severe deafness, retinitis pigmentosa developing in the first or second decade, and normal vestibular function. Congenital bilateral sensorineural hearing loss is mild to moderate in the low frequencies and severe to profound in the higher frequencies. Additional manifestations include night blindness, constricted visual field (tunnel vision), and later on decreased visual acuity sometimes ending with bare light perception.
ORPHACODE:	231178
SYNONYMS:	USH2
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10
ANALYTE(S):	USH2A ADGRV1 MYO7A WHRN PDZD7

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

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Related Laboratories

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- adhesion G protein-coupled receptor V1
- myosin VIIA
- PDZ domain containing 7
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Related Gene Panels

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