DISEASE:
Usher syndrome type 1

NAME:	Usher syndrome type 1
DESCRIPTION:	A rare ciliopathy characterized by profound congenital deafness, retinitis pigmentosa and vestibular dysfunction. Retinitis pigmentosa results in visual loss and generally manifests as night blindness, progressively constricted visual fields, and impaired visual acuity. Vestibular dysfunction a defining feature of this form, manifests as delayed motor development with affected infants taking longer to sit independently and to walk. Later on, vestibular dysfunction results in difficulty with activities requiring balance.
ORPHACODE:	231169
SYNONYMS:	USH1

XREF(S):	Orphanet OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	USH1K CDH23 USH1C USH1G MYO7A PCDH15 USH1E USH1H CIB2 ESPN
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