

DISEASE:
Waardenburg-Shah syndrome

NAME:	Waardenburg-Shah syndrome
DESCRIPTION:	Waardenburg-Shah syndrome (WSS), also known as Waardenburg syndrome type 4 (WS4) is characterized by the association of Waardenburg syndrome (sensorineural hearing loss and pigmentary abnormalities) and Hirschsprung disease (aganglionic megacolon).
ORPHACODE:	897
SYNOMYS:	Shah-Waardenburg syndrome WS4 Waardenburg syndrome type 4 Waardenburg-Hirschsprung syndrome
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10
ANALYTE(S):	SOX10 EDN3 EDNRB MITF

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