DISEASE:
Partington syndrome

NAME:	Partington syndrome
DESCRIPTION:	Partington syndrome is a form of syndromic X-linked mental retardation (S-XLMR) characterised by the association of mild to moderate intellectual deficit, dysarthria and dystonic hand movements. So far, less than 20 cases have been described in the literature. The syndrome is caused by mutations in the Aristaless-related homeobox (ARX) gene (Xp22.13). Transmission is X-linked recessive.
ORPHACODE:	94083
SYNONYMS:	Partington-Mulley syndrome X-linked intellectual disability-dystonia-dysarthria syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ARX
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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