DISEASE:
Pontocerebellar hypoplasia type 1

NAME:	Pontocerebellar hypoplasia type 1
DESCRIPTION:	A severe, genetic form of pontocerebellar hypoplasia (PCH) characterized by spinal cord anterior horn cell degeneration in addition to pontocerebellar hypoplasia. Clinically, patients manifest with a severe global development deficit that is evident early on from difficulties in feeding and swallowing
ORPHACODE:	2254
SYNONYMS:	Norman disease PCH1
XREF(S):	Orphanet OMIM OMIM OMIM MeSH ICD-10 OMIM OMIM OMIM

ANALYTE(S):	AGTPBP1 VRK1 EXOSC3 EXOSC8 SLC25A46 EXOSC9
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP/GTP binding carboxypeptidase 1
- exosome component 3
- exosome component 8
- exosome component 9
- solute carrier family 25 member 46
- VRK serine/threonine kinase 1

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB
