DISEASE:
ATP13A2-related juvenile neuronal ceroid lipofuscinosis

NAME:	ATP13A2-related juvenile neuronal ceroid lipofuscinosis
DESCRIPTION:	A rare neuronal ceroid lipofiscinosis disorder characterized by juvenile-onset of progressive spinocerebellar ataxia, bulbar syndrome (manifesting with dysarthria, dysphagia and dysphonia), pyramidal and extrapyramidal involvement (including myoclonus, amyotrophy, unsteady gait, akinesia, rigidity, dysarthric speech) and intellectual deterioration. Muscle biopsy displays autofluorescent bodies and lipofuscin deposits in brain and, occasionally the retina, upon post mortem.
ORPHACODE:	314632
SYNONYMS:	CLN12 disease Juvenile parkinsonism-neuronal ceroid lipofuscinosis
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ATP13A2
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RELATED CONTENT

Related Genetic Tests

- Neurodegeneration (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- ATPase cation transporting 13A2

Related Gene Panels

- Neurodegeneration (99 genes) - IPG