DISEASE:
Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia

NAME:	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia
DESCRIPTION:	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia is a rare autosomal dominant disease characterized by a complex phenotype including progressive dementia, apraxia, apathy, impaired balance, parkinsonism, spasticity and epilepsy.
ORPHACODE:	313808
SYNONYMS:	ALSP Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia Autosomal dominant leukoencephalopathy with neuroaxonal spheroids FPSG Familial dementia, Neumann type Familial progressive subcortical gliosis GPSC HDLS Hereditary diffuse leukoencephalopathy with spheroids POLD Pigmentary orthochromatic leukodystrophy Subcortical gliosis of Neumann

XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	AARS2 CSF1R
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2039

RELATED CONTENT

Related Genetic Tests

- Mitochondrial disorders (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- alanyl-tRNA synthetase 2, mitochondrial
- colony stimulating factor 1 receptor

Related Gene Panels

- mitochondrial disease, nuclear based (343 genes) - VUB