DISEASE:
Huntington disease-like syndrome due to C9ORF72 expansions

NAME:	Huntington disease-like syndrome due to C9ORF72 expansions
DESCRIPTION:	A rare, genetic neurodegenerative disease characterized by movement disorders, including dystonia, chorea, myoclonus, tremor and rigidity. Associated features are also cognitive and memory impairment, early psychiatric disturbances and behavioral problems.
ORPHACODE:	401901
SYNONYMS:	C9ORF72-related Huntington disease phenocopy C9ORF72-related Huntington disease-like syndrome Huntington disease phenocopy due to C9ORF72 expansions
XREF(S):	Orphanet
ANALYTE(S):	C9ORF72
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RELATED CONTENT

Related Genetic Tests

- Amyotrophic lateral sclerosis (GGGGCC repeat expansion in the C9ORF72 gene)
- Neurodegeneration (gene panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- C9orf72-SMCR8 complex subunit

Related Gene Panels

- Neurodegeneration (99 genes) - IPG
- Neuropathy (148 genes) - IPG