DISEASE:
Inherited congenital spastic tetraplegia

NAME:	Inherited congenital spastic tetraplegia
DESCRIPTION:	Inherited congenital spastic tetraplegia is a rare, genetic, neurological disease characterized by non-progressive, variable spastic quadriparesis in multiple members of a family, in the absence of additional factors complicating pregnancy or birth (e.g. perinatal asphyxia, congenital infection). Additional clinical features include congenital hypotonia, intellectual disability, and developmental delay. Dysphagia, dysarthria, exotropia, nystagmus, seizures and brain atrophy with ventriculomegaly may be also present.
ORPHACODE:	210141
SYNONYMS:	Inherited congenital spastic quadriplegia
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	KANK1 GAD1 ADD3
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RELATED CONTENT

Related Genetic Tests

- Spastic Paraplegia (gene panel)

Related Laboratories

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

Related Analytes

- adducin 3
- glutamate decarboxylase 1
- KN motif and ankyrin repeat domains 1

Related Gene Panels

- Spastic Paraplegia (89 genes) - IPG