DISEASE:
Autosomal recessive dopa-responsive dystonia

NAME:	Autosomal recessive dopa-responsive dystonia
DESCRIPTION:	A very rare neurometabolic disorder characterized by a spectrum of symptoms ranging from those seen in dopa-responsive dystonia (DRD) to progressive infantile encephalopathy.
ORPHACODE:	101150
SYNONYMS:	Autosomal recessive Segawa syndrome DYT5b Tyrosine hydroxylase deficiency Tyrosine hydroxylase-deficient dopa-responsive dystonia
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TH TSPOAP1
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RELATED CONTENT

Related Genetic Tests

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- Segawa syndrome (TH gene)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
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Related Analytes

- tyrosine hydroxylase
- TSPO associated protein 1

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Source URL: http://gentest.healthdata.be/disease/3679