DISEASE:
Menkes disease

NAME:	Menkes disease
DESCRIPTION:	A rare congenital disorder of copper metabolism with severe multisystemic manifestations that are primarily characterized by progressive neurodegeneration and marked connective tissue anomalies. A pathognomonic feature is the typical sparse, abnormal steely hair.
ORPHACODE:	565
SYNONYMS:	MD Menkes kinky hair disease Menkes syndrome
XREF(S):	Orphanet MedDRA OMIM ICD-10
ANALYTE(S):	ATP7A
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ichthyosis (gene panel)
- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- ATPase copper transporting alpha

Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL
- Neuropathy (148 genes) - IPG

Source URL: http://gentest.healthdata.be/disease/187