DISEASE:
Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome

NAME:	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome
DESCRIPTION:	Congenital cataract - hypertrophic cardiomyopathy - mitochrondrial myopathy (CCM) is a mitochondrial disease (see this term) characterized by cataracts, hypertrophic cardiomyopathy, muscle weakness and lactic acidosis after exercise.
ORPHACODE:	1369
SYNONYMS:	Sengers syndrome
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	TKFC SLC25A4 AGK
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/274

RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- acylglycerol kinase
- solute carrier family 25 member 4
- triokinase and FMN cyclase

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB