DISEASE:
Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease

NAME:	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
DESCRIPTION:	A rare glycogen storage disease characterized by fetal or neonatal onset of severe cardiomyopathy with non-lysosomal glycogen accumulation and fatal outcome in infancy. Patients present with massive cardiomegaly, severe cardiac and respiratory complications, and failure to thrive. Non-specific facial dysmorphism, bilateral cataracts, macroglossia, hydrocephalus, enlarged kidneys, and skeletal muscle involvement have been reported in some cases.
ORPHACODE:	439854
SYNONYMS:	Fatal congenital hypertrophic cardiomyopathy due to GSD Fatal congenital hypertrophic cardiomyopathy due to glycogenosis
XREF(S):	Orphanet OMIM
ANALYTE(S):	PRKAG2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Cardiopathies, hereditary (gene panel)
- Hypertrophic cardiomyopathy (gene panel)

Related Laboratories

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

Related Analytes

- protein kinase AMP-activated non-catalytic subunit gamma 2

Related Gene Panels

- Cardiopathies, hereditary (102 genes) - KUL
- Congenital heart disease (29 genes) - VUB
- Hypertrophic cardiomyopathy (75 genes) - IPG