DISEASE:
Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation

NAME:	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation
DESCRIPTION:	A mitochondrial oxidative phosphorylation disorder characterized by hypertrophic and dilated cardiomyopathy, failure to thrive, myopathy with generalized hypotonia and increased creatine kinase, developmental delay and/or regression with cerebral atrophy on brain MRI, renal manifestations including chronic renal failure, renal tubular acidosis and lactic acidosis. Additional clinical features include seizures and respiratory failure.
ORPHACODE:	324525
SYNONYMS:	Hypertrophic cardiomyopathy with kidney anomalies due to mtDNA mutation Hypertrophic cardiomyopathy with renal anomalies due to mitochondrial DNA mutation
XREF(S):	Orphanet
ANALYTE(S):	MT-TL1
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