DISEASE:
Combined oxidative phosphorylation defect type 8

NAME:	Combined oxidative phosphorylation defect type 8
DESCRIPTION:	Combined oxidative phosphorylation defect type 8 is a mitochondrial disease due to a defect in mitochondrial protein synthesis resulting in deficiency of respiratory chain complexes I, III and IV in the cardiac and skeletal muscle and brain characterized by severe hypertrophic cardiomyopathy, pulmonary hypoplasia, generalized muscle weakness and neurological involvement.
ORPHACODE:	319504
SYNONYMS:	COXPD8
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	AARS2
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