DISEASE:
Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome

NAME:	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome
DESCRIPTION:	A rare mitochondrial disease characterized by neonatal onset of severe cardiac and/or neurologic signs and symptoms mostly associated with a fatal outcome in the neonatal period or in infancy, although a milder phenotype with later onset and slowly progressive neurologic deterioration has also been reported. Clinical manifestations are variable and include respiratory insufficiency, hypotonia, cardiomyopathy, and seizures. Serum lactate is elevated in most cases. Brain imaging may show cerebellar atrophy or hypoplasia.
ORPHACODE:	457185
SYNONYMS:	COQ4-related neonatal encephalomyopathy
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	COQ4
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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