DISEASE:
Pyruvate carboxylase deficiency, severe neonatal type

NAME:	Pyruvate carboxylase deficiency, severe neonatal type
DESCRIPTION:	Severe neonatal pyruvate carboxylase (PC) deficiency (Type B) is a rare, extremely severe form of PC deficiency characterized by severe, early-onset metabolic acidosis, and a generally fatal outcome in early infancy.
ORPHACODE:	353314
SYNONYMS:	Pyruvate carboxylase deficiency type B
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PC
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- pyruvate carboxylase