

DISEASE:
Acute neonatal citrullinemia type I

NAME:	Acute neonatal citrullinemia type I
DESCRIPTION:	A severe form of citrullinemia type 1 characterized biologically by hyperammonemia and clinically by progressive lethargy, poor feeding and vomiting, seizures and possible loss of consciousness, within one to a few days of birth, with variable signs of increased intracranial pressure. The condition can lead to significant neurologic deficits.
ORPHACODE:	247546
SYNOMYS:	Acute neonatal citrullinemia type 1 Early-onset citrullinemia type 1 Early-onset citrullinemia type I
XREF(S):	Orphanet
ANALYTE(S):	ASS1
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Source URL: <http://gentest.healthdata.be/disease/856>