

DISEASE:
Argininosuccinic aciduria

NAME:	Argininosuccinic aciduria
DESCRIPTION:	A rare, genetic disorder of urea cycle metabolism typically characterized by either a severe, neonatal-onset form that manifests with hyperammonemia accompanied with vomiting, hypothermia, lethargy and poor feeding in the first few days of life, or late-onset forms that manifest with stress- or infection-induced episodic hyperammonemia or, in some, behavioral abnormalities and/or learning disabilities, or chronic liver disease. Patients often manifest liver dysfunction.
ORPHACODE:	23
SYNONYMS:	ASA deficiency ASL deficiency Argininosuccinase deficiency Argininosuccinatelyase deficiency Argininosuccinic acid lyase deficiency
XREF(S):	Orphanet MeSH ICD-10 OMIM MedDRA
ANALYTE(S):	ASL

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