

DISEASE:
Adenylosuccinate lyase deficiency

NAME:	Adenylosuccinate lyase deficiency
DESCRIPTION:	A disorder of purine metabolism characterized by intellectual disability, psychomotor delay and/or regression, seizures, and autistic features.
ORPHACODE:	46
SYNONYMS:	ADSL deficiency Adenylosuccinase deficiency
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	ADSL
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Source URL: <http://gentest.healthdata.be/disease/405>