

DISEASE:**Adenylosuccinate synthetase-like 1-related distal myopathy**

NAME:	Adenylosuccinate synthetase-like 1-related distal myopathy
DESCRIPTION:	A rare autosomal recessive distal myopathy characterized by slowly progressive diffuse muscle weakness in childhood, followed by predominantly distal muscle weakness in adolescence, and quadriceps muscle weakness in the fourth decade. Facial muscle weakness is commonly reported. Muscle biopsy shows fiber size variation, increased internal nuclei, fiber splitting, rimmed vacuoles, and focal endomysial fibrosis.
ORPHACODE:	482601
SYNONYMS:	ADSSL1-related distal myopathy
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>ADSS1</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/2694>

RELATED CONTENT

Related Analytes

- [adenylosuccinate synthase 1](#)

Source URL: <http://gentest.healthdata.be/disease/2694>