

Opis choroby *

Definicja

Short-chain acyl-CoA dehydrogenase (SCAD) deficiency is a very rare inborn error of mitochondrial fatty acid oxidation characterized by variable manifestations ranging from asymptomatic individuals (in most cases) to those with failure to thrive, hypotonia, seizures, developmental delay and progressive myopathy.

Dane

Klasyfikacja	Synonimy
Choroba	ACADS deficiency Niedobór ACADS
	Niedobór SCAD
	SCADD
	SCAD deficiency
	SCADD
Kod ORPHA	
26792	Kod OMIM
	201470
Kod ICD10	
	E71.3
Kod ICD11	
5C52.01	

*Źródło

orphanet