

## 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

Choroba

#### Synonimy

ACADS deficiency  
Niedobór ACADS  
Niedobór SCAD  
SCADD  
SCAD deficiency  
SCADD

#### Kod ORPHA

26792

#### Kod OMIM

201470

#### Kod ICD10

E71.3

#### Kod ICD11

5C52.01

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#### \*Źródło

orphanet