

## Opis choroby \*

### Definicja

Intellectual disability-alacrima-achalasia syndrome is a rare, genetic intellectual disability syndrome characterized by delayed motor and cognitive development, absence or severe delay in speech development, intellectual disability, and alacrima. Achalasia/dysphagia and mild autonomic dysfunction (i.e. anisocoria) have also been reported in some patients. The phenotype is similar to the one observed in autosomal recessive Triple A syndrome, but differs by the presence of intellectual disability in all affected individuals.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

289483

#### Kod OMIM

300858

#### Kod ICD10

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#### Kod ICD11

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### \*Źródło

orphanet