

## Opis choroby \*

### Definicja

A form of ectodermal dysplasia syndrome characterized by a short stature of prenatal onset, alopecia, ichthyosis, photophobia, ectrodactyly, seizures, scoliosis, multiple contractures, fusions of various bones (particularly elbows, carpals, metacarpals, and spine), intellectual disability, and facial dysmorphism (microdolichocephaly, madarosis, large ears and long nose). ACD syndrome overlaps with ichthyosis follicularis-alopecia-photophobia syndrome.

### Dane

#### Klasyfikacja

#### Synonimy

Zespół wad wrodzonych ACD-intellectual disability syndrome

#### Kod ORPHA

1005

#### Kod OMIM

203550

#### Kod ICD10

Q87.8

#### Kod ICD11

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#### \*Źródło

orphanet