

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-aloppecia-photophobia syndrome.

Dane

Klasyfikacja **Synonimy**
Zespół wad wrodzonych ACD-intellectual disability syndrome

Kod ORPHA **Kod OMIM** **Kod ICD10**
1005 203550 Q87.8

Kod ICD11

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*Źródło

orphanet