

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