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Alopecia-contractures-dwarfism-intellectual disability syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Alopecia-contractures-dwarfism-intellectual disability syndrome. ORPHA:1005*

Alopecia-contractures-dwarfism-intellectual disability syndrome (ACD syndrome) is 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-alpecia-photophobia syndrome.