Arthrogryposis-ectodermal dysplasia-other anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Arthrogryposis-ectodermal dysplasia-other anomalies syndrome. ORPHA:3200

A rare, genetic developmental defect during embryogenesis syndrome characterized by camptodactyly, joint contractures with amyotrophy, and ectodermal anomalies (oligodontia, enamel abnormalities, longitudinally broken nails, hypohidrotic skin with tendency to excessive bruising and scarring after injuries and scratching), as well as growth retardation, kyphoscoliosis, mild facial dysmorphism, and microcephaly. There have been no further descriptions in the literature since 1992.