Type III Acrocephalosyndactyly

National Cancer Institute

Source

National Cancer Institute. Type III Acrocephalosyndactyly. NCI Thesaurus. Code C75034.

A rare autosomal dominant syndrome caused by mutations in the TWIST1 gene. It is characterized by premature closure of skull bones resulting in abnormally shaped head, high forehead, hypertelorism, and facial asymmetry. It may be associated with fusion of certain fingers or toes.