Type I Acrocephalosyndactyly

National Cancer Institute

Source

National Cancer Institute. Type I Acrocephalosyndactyly. NCI Thesaurus. Code C99099.

An autosomal dominant inherited type of acrocephalosyndactyly caused by mutations in the FGFR2 gene. It is characterized by early closure of the sutures between the skull bones, bulging eyes, low-set ears, fusion of the second, third, and forth fingers, and fusion of the toes.