Beare-Stevenson Cutis Gyrata Syndrome

National Cancer Institute

Source

National Cancer Institute. Beare-Stevenson Cutis Gyrata Syndrome. NCI Thesaurus. Code C123813.

A rare, autosomal dominant inherited disorder caused by mutations in the FGFR2 gene. It is characterized by the premature fusion of the bones of the skull (craniosynostosis) and a skin abnormality called cutis gyrata. The craniosynostosis results in a cloverleaf-shaped skull, wide-set eyes, ear abnormalities, underdeveloped upper jaw, and developmental delays. Cutis gyrata is characterized by a wrinkled skin appearance, especially on the face, near the ears, and on the palms and soles.