Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome. ORPHA:1555

Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome, also known as Beare-Stevenson syndrome (BSS), is a severe form of syndromic craniosynostosis, characterized by a variable degree of craniosynostosis, with cloverleaf skull reported in over 50% of cases, cutis gyrata, corduroy-like linear striations in the skin, acanthosis nigricans, skin tags, and choanal stenosis or atresia). Additional features include facial features similar to Crouzon disease, ear defects (conductive hearing loss, posteriorly angulated ears, stenotic auditory canals, preauricular furrows, and narrow ear canals), hirsutism, a prominent umbilical stump, and genitorurinary anomalies (anteriorly placed anus, hypoplasic labia, hypospadias). BSS is associated with a poor outcome as patients present an elevated risk for sudden death in their first year of life. Significant developmental delay and intellectual disability are observed in most patients who survive infancy.