Severe achondroplasia-developmental delay-acanthosis nigricans syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Severe achondroplasia-developmental delay-acanthosis nigricans syndrome. ORPHA:85165

Severe achondroplasia-developmental delay-acanthosis nigricans syndrome is characterised by the association of severe achondroplasia with developmental delay and acanthosis nigricans. It has been described in four unrelated individuals. Structural central nervous system anomalies, seizures and hearing loss were also reported, together with bowing of the clavicle, femur, tibia and fibula in some cases. The syndrome is caused by a Lys650Met substitution in the kinase domain of fibroblast growth factor receptor 3 (encoded by the FGFR3 gene; 4p16.3).