TMEM165-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. TMEM165-CDG. ORPHA:314667

TMEM165-CDG is a form of congenital disorders of N-linked glycosylation characterized by a psychomotor delay-dysmorphism (pectus carinatum, dorsolumbar kyphosis and severe sinistroconvex scoliosis, short distal phalanges, genua vara, pedes planovalgi syndrome) with postnatal growth deficiency and major spondylo-, epi-, and metaphyseal skeletal involvement. Additional features include facial dysmorphism (midface hypoplasia, internal strabism of the right eye, low-set ears, moderately high arched palate, small teeth), nephrotic syndrome, cardiac defects, and feeding problems. The disease is caused by mutations in the gene TMEM165 (4q12).