XYLT1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. XYLT1-CDG. ORPHA:370930

XYLT1-CDG is a rare congenital disorder of glycosylation characterized by moderate intellectual disability, short stature, mild skeletal changes and distinctive facial features with coarse face, synophyrs and deep nasolabial ridges. Skeletal features include broad ribs, stocky long bones, short femoral necks with coxa valga, clinodactyly and broad thumbs.