
DISEASE:
XYLT1-CDG

NAME:	XYLT1-CDG
DESCRIPTION:	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, synophrys and deep nasolabial ridges. Skeletal features include broad ribs, stocky long bones, short femoral necks with coxa valga, clinodactyly and broad thumbs.
ORPHACODE:	370930
XREF(S):	<u>Orphanet</u> <u>ICD-10</u>
ANALYTE(S):	<u>XYLT1</u>
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