DISEASE:
TMEM165-CDG

NAME:	TMEM165-CDG
DESCRIPTION:	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).
ORPHACODE:	314667
SYNONYMS:	CDG syndrome type IIk CDG-IIk CDG2K Carbohydrate deficient glycoprotein syndrome type IIk Congenital disorder of glycosylation type 2k Congenital disorder of glycosylation type IIk
XREF(S):	Orphanet OMIM ICD-10

ANALYTE(S):	TMEM165
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