DISEASE:
Deletion 5q35

NAME:	Deletion 5q35
DESCRIPTION:	Deletion 5q35 refers to the different congenital malformation syndromes resulting from deletions of variable extent of the terminal part of the long arm of chromosome 5 (5q), spanning the region from 5q35.1 to 5q35.3 . The most significant anomaly is a recurring deletion in 5q35.2 comprising the NSD1 gene that causes Sotos syndrome that is characterized by cardinal features including excessive growth during childhood, macrocephaly, distinctive facial gestalt and various degrees of learning difficulty. Subtelomeric deletions of the terminal 3.5 Mb region on 5q35.3 are very rare, characterized by prenatal lymphedema with increased nuchal translucency, pronounced muscular hypotonia in infancy, borderline intelligence, postnatal short stature due to growth hormone deficiency, and a variety of minor anomalies such as mildly bell-shaped chest, minor congenital heart defects and a distinct facial gestalt. Larger deletions including bands 5q35.1, 5q35.2 and 5q35.3 cause a more severe phenotype that associates severe developmental delay with microcephaly, and significant cardiac defects (e.g. atrial septal defect with/without atrioventricular conduction defects, Ebstein anomaly, tetralogy of Fallot) linked to haploinsufficiency of NKX2.5 (5q35.1). Various combinations of signs may result from deletions of variable extent depending on the genes comprised in the deleted segment.
ORPHACODE:	1627

SYNONYMS:	Del (5)(q35) Del (5)(qter) Distal 5q deletion Monosomy 5q35 Telomeric deletion 5q
XREF(S):	Orphanet ICD-10
ANALYTE(S):	NKX2-5 NSD1
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