DISEASE:
5p13 microduplication syndrome

NAME:	5p13 microduplication syndrome
DESCRIPTION:	A rare partial autosomal trisomy/tetrasomy characterized by global developmental delay, intellectual disability, autistic behavior, muscular hypotonia, macrocephaly and facial dysmorphism (frontal bossing, short palpebral fissures, low set, dysplastic ears, short or shallow philtrum, high arched or narrow palate, micrognathia). Other associated clinical features include sleep disturbances, seizures, aplasia/hypoplasia of the corpus callosum, skeletal abnormalities (large hands and feet, long fingers and toes, talipes).
ORPHACODE:	329802
SYNONYMS:	Dup(5)(p13) Trisomy 5p13
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	NIPBL
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- NIPBL cohesin loading factor

Source URL: http://gentest.healthdata.be/disease/1646