DISEASE:
Hao-Fountain syndrome due to 16p13.2 microdeletion

NAME:	Hao-Fountain syndrome due to 16p13.2 microdeletion
DESCRIPTION:	A partial deletion of the short arm of chromosome 16 characterized by developmental delay, intellectual disability, speech delay, autism spectrum disorder, epilepsy, hypogonadism, and hypotonia. The behavioral profile includes impulsivity, compulsivity, stubbornness, manipulative behaviors, temper tantrums, and aggressive behaviors.
ORPHACODE:	500055
SYNONYMS:	Chromosome 16p13.2 deletion syndrome Del(16)(p13.2) Monosomy 16p13.2
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	USP7
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- ubiquitin specific peptidase 7

Source URL: http://gentest.healthdata.be/disease/2624