DISEASE:
Nijmegen breakage syndrome-like disorder

NAME:	Nijmegen breakage syndrome-like disorder
DESCRIPTION:	Nijmegen breakage syndrome-like disorder is a rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by growth retardation, short stature, developmental delay, intellectual disability, craniofacial dysmorphism (i.e. severe microcephaly, sloping forehead, prominent eyes, broad nasal ridge, hypoplastic nasal septum, epicanthal folds), spontaneous chromosomal instability, cellular hypersensitivity to ionizing radiation and radioresistant DNA synthesis, without severe infections, immunodeficiency or cancer predisposition. Additional reported features include mild spasticity, slight and nonprogressive ataxia, hyperopia, multiple pigmented nevi, widely spaced nipples, and clinodactyly.
ORPHACODE:	240760
SYNONYMS:	Microcephaly and chromosomal instability without immunodeficiency NBS-like disorder NBSLD RAD50 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MRE11 RAD50

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1632

RELATED CONTENT

Related Genetic Tests

- Hereditary cancer (Breast, ovary, colon) (26 genes)
- Hereditary cancer (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- MRE11 homolog, double strand break repair nuclease
- RAD50 double strand break repair protein

Related Gene Panels

- Cancer (Breast, ovary, colon,…) (26 genes) - ULG
- Hereditary predisposition to cancer (47 genes) - IPG