# DISEASE:
# Nijmegen breakage syndrome

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| **NAME:** | Nijmegen breakage syndrome |
| **DESCRIPTION:** | A rare, genetic chromosomal instability syndrome presenting at birth with microcephaly, dysmorphic facial features which become more noticeable with age, growth delay, recurring sinopulmonary infections and extremely high frequency of malignancies. |
| **ORPHACODE:** | 647 |
| **SYNONYMS:** | AT V1<br>Ataxia-telangiectasia, variant 1<br>Berlin breakage syndrome<br>Immunodeficiency-microcephaly-chromosomal instability syndrome<br>Microcephaly-immunodeficiency-lymphoid malignancy syndrome<br>NBS<br>Seemanova syndrome type 2 |
| **XREF(S):** | Orphanet<br>MeSH<br>OMIM<br>ICD-10<br>MeSH<br>MedDRA |

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| **ANALYTE(S):** | NBN |
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# RELATED CONTENT

## Related Genetic Tests

- Hereditary cancer (Breast, ovary, colon) (26 genes)
- Hereditary cancer (gene panel)
- Nijmegen Breakage Syndrome
- Nijmegen breakage syndrome

## Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- nibrin

## Related Gene Panels

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