# DISEASE:
# ALG2-CDG

| | |
|---|---|
| **NAME:** | ALG2-CDG |
| **DESCRIPTION:** | A form of congenital disorders of N-linked glycosylation characterized by iris coloboma, cataract, infantile spasms, developmental delay and abnormal coagulation factors. The disease is caused by loss-of-function mutations in the gene ALG2 (9q31.1). Transmission is autosomal recessive. |
| **ORPHACODE:** | 79326 |
| **SYNONYMS:** | CDG syndrome type Ii<br>CDG-Ii<br>CDG1I<br>Carbohydrate deficient glycoprotein syndrome type Ii<br>Congenital disorder of glycosylation type 1i<br>Congenital disorder of glycosylation type Ii<br>Mannosyltransferase 2 deficiency |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | ALG2 |
| **CREATED:** | 13 May 2019 - 01:02 |

| | |
|---|---|
| **CHANGED:** | 22 Jun 2023 - 16:14 |

---

Source URL: http://gentest.healthdata.be/disease/3289

# RELATED CONTENT

## Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)

## Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- ALG2 alpha-1,3/1,6-mannosyltransferase

## Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL

Source URL: http://gentest.healthdata.be/disease/3289