# DISEASE:
# COG2-CDG

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| **NAME:** | COG2-CDG |
| **DESCRIPTION:** | A rare, congenital disorder of glycosylation caused by mutations in the COG2 gene and characterized by normal presentation at birth, followed by progressive deterioration with postnatal microcephaly, developmental delay, intellectual disability, seizures, spastic quadriplegia, liver dysfunction, hypocupremia and hypoceruloplasminemia in the first year of life. Diffuse cerebral atrophy and thin corpus callosum may be observed on brain MRI. |
| **ORPHACODE:** | 435934 |
| **SYNONYMS:** | COG2-related congenital disorder of glycosylation |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | COG2 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)

## Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- component of oligomeric golgi complex 2

## Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL