# DISEASE:
# Homocystinuria due to methylene tetrahydrofolate reductase deficiency

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| **NAME:** | Homocystinuria due to methylene tetrahydrofolate reductase deficiency |
| **DESCRIPTION:** | Homocystinuria due to methylene tetrahydrofolate reductase (MTHFR) deficiency is a metabolic disorder characterised by neurological manifestations. |
| **ORPHACODE:** | 395 |
| **SYNONYMS:** | MTHFR deficiency<br>Methylene tetrahydrofolate reductase deficiency |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | MTHFR |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Homocystinuria (hot spot mutation - c.1298A>C)
- Homocystinuria (hot spot mutation - c.677C>T)

## Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

## Related Analytes

- methylenetetrahydrofolate reductase