# Belgian Genetic Tests database

## ANALYTE:
## MMACHC

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|---|---|
| **NAME:** | metabolism of cobalamin associated C |
| **SYMBOL:** | MMACHC |
| **VERSION OF ORPHANET:** | 2023-06-22 14:14:43 |
| **SYNONYMS:** | DKFZP564I122<br>cblC |
| **XREF(S):** | Orphanet<br>Ensembl<br>Genatlas<br>HGNC<br>OMIM<br>Reactome<br>SwissProt |
| **CREATED:** | 13 May 2019 - 01:01 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

Source URL: http://gentest.healthdata.be/analyte/3780

# RELATED CONTENT

## Related Genetic Tests

- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Atypical Hemolytic Uremic Syndrome (aHUS) (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Early onset epileptic encephalopathy (gene panel - 845 genes)
- Epilepsy gene panel
- Hereditary Spastic Paraplegia (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Intellectual disability & Epilepsy (gene panel)
- Intellectual disability (gene panel)
- Intellectual disability (virtual gene panel)
- Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders (213 genes)
- Nephrogenetics / Nephropathy (gene panel)
- Nephropathies, hereditary (gene panel)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Neuropathy (gene panel)
- Peripheral neuropathy (gene panel)
- Stroke (gene panel)

## Related Diseases

- Methylmalonic acidemia with homocystinuria, type cblC

## Related Gene Panels

- Ataxia (348 genes) - ULB
- Atypical Hemolytic Uremic Syndrome (aHUS) and Complement disorders (17 genes) - IPG
- Congenital malformation (1721 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Intellectual disability/Epilepsy (1091 genes) - ULG
- Metabolic disorders (213 genes) - VUB
- Nephropathies, hereditary (219 genes) - KUL
- Nephropathy panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Neuropathy (genepanel) - UZA
- Neuropathy panel - UGent
- Panel Nephro-ULG-V1
- Stroke - UGent

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