# ANALYTE:
# ATP7B

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|---|---|
| **NAME:** | ATPase copper transporting beta |
| **SYMBOL:** | ATP7B |
| **VERSION OF ORPHANET:** | 2023-06-22 14:14:43 |
| **SYNONYMS:** | Wilson disease<br>copper pump 2<br>copper-transporting ATPase 2 |
| **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 |

# RELATED CONTENT

## Related Genetic Tests

- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Ataxia Spasticity (gene panel)
- Cerebral palsy (gene panel)
- Dystonia (gene panel)
- Hepatology (gene panel)
- Hereditary Spastic Paraplegia (gene panel)
- Inherited Kidney Diseases (Gene Panel)
- Intellectual disability (virtual gene panel)
- Leukodystrophy (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)
- Movement Disorders (gene panel)
- Movement Disorders (gene panel)
- Nephrogenetics / Nephropathy (gene panel)
- Tubulopathy (gene panel)
- Wilson Disease

## Related Diseases

- Wilson disease

## Related Gene Panels

- Ataxia (348 genes) - ULB
- Ataxia Spasticity - UGent

- Cerebral palsy (212 genes) - UZA
- Dystonia (68 genes) - KUL
- Hepatology panel - UGent
- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Intellectual disability (gene panel)
- Leukodystrophy - UGent
- Metabolic diseases with hepatic disorders (20 genes) - UCL
- Movement Disorders - UGent
- Movement Disorders - ULG
- Nephropathy panel - UGent
- Panel Nephro-ULG-V1
- Tubulopathy/Nephrolithiasis (106 genes) - IPG
- Wilson Disease (1 gene) - UCL

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