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**DISEASE:**  
**Autosomal recessive spastic paraplegia type 56**

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| <b>NAME:</b>        | Autosomal recessive spastic paraplegia type 56                                                                                                                                                                                                                                                                                           |
| <b>DESCRIPTION:</b> | A rare form of hereditary spastic paraplegia characterized by delayed walking, toe walking, unsteady and spastic gait, hyperreflexia of the lower limbs, and extensor plantar responses. Upper limbs spasticity and dystonia, subclinical axonal neuropathy, cognitive impairment and intellectual disability have also been associated. |
| <b>ORPHACODE:</b>   | 320411                                                                                                                                                                                                                                                                                                                                   |
| <b>SYNONYMS:</b>    | SPG56                                                                                                                                                                                                                                                                                                                                    |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                                                                                          |
| <b>ANALYTE(S):</b>  | <u>CYP2U1</u>                                                                                                                                                                                                                                                                                                                            |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                      |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                      |

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## RELATED CONTENT

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### Related Genetic Tests

- [Spastic Paraplegia \(gene panel\)](#)

### Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)

### Related Analytes

- [cytochrome P450 family 2 subfamily U member 1](#)

### Related Gene Panels

- [Spastic Paraplegia \(89 genes\) - IPG](#)

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