

**ANALYTE:**  
**AP4E1**

<b>NAME:</b>	adaptor related protein complex 4 subunit epsilon 1
<b>SYMBOL:</b>	AP4E1
<b>VERSION OF ORPHANET:</b>	2023-06-22 14:14:43
<b>SYNONYMS:</b>	AP-4-EPSILON SPG51
<b>XREF(S):</b>	<u>Orphanet</u> <u>Ensembl</u> <u>Genatlas</u> <u>HGNC</u> <u>OMIM</u> <u>Reactome</u> <u>SwissProt</u>
<b>CREATED:</b>	13 May 2019 - 01:01
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- [Ataxia Spasticity \(gene panel\)](#)
- [Cerebral palsy \(gene panel\)](#)
- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Early onset epileptic encephalopathy \(gene panel - 845 genes\)](#)
- [Epilepsy gene panel](#)
- [Hereditary Spastic Paraplegia \(94 genes\)](#)
- [Hereditary Spastic Paraplegia \(gene panel\)](#)
- [Hereditary spastic paraplegia \(gene panel - 249 genes\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Malformations of cortical development \(235 genes\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Neuromuscular disorders \(548 genes\)](#)
- [Spastic Paraplegia \(gene panel\)](#)

### Related Diseases

- [Severe intellectual disability and progressive spastic paraplegia](#)

### Related Gene Panels

- [Ataxia Spasticity - UGent](#)
- [Cerebral palsy \(212 genes\) - UZA](#)

- Congenital malformation (1721 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Epilepsy gene panel - VUB
- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Hereditary Spastic Paraplegia (94 genes) - KUL
- Hereditary spastic paraplegia (188 genes) - ULB
- Intellectual disability & Epilepsy - UGent
- Intellectual disability (gene panel)
- Malformations of cortical development (235 genes) - VUB
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Neuromuscular disorders (548 genes) - ULB
- Spastic Paraplegia (89 genes) - IPG

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