

**ANALYTE:**  
**AP2S1**

|                             |                                                                                                                           |
|-----------------------------|---------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>                | adaptor related protein complex 2 subunit sigma 1                                                                         |
| <b>SYMBOL:</b>              | AP2S1                                                                                                                     |
| <b>VERSION OF ORPHANET:</b> | 2023-06-22 14:14:43                                                                                                       |
| <b>SYNONYMS:</b>            | FBH3<br>FBHOK<br>sigma-2                                                                                                  |
| <b>XREF(S):</b>             | <u>Orphanet</u><br><u>Ensembl</u><br><u>Genatlas</u><br><u>HGNC</u><br><u>OMIM</u><br><u>Reactome</u><br><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

- [Endocrine Disorders - Hyper\(Hypo\)parathyroidism \(gene panel - 24 genes\)](#)
- [Genetic disorders of Calcium and Phosphate metabolism \(gene panel\)](#)
- [Hypocalciuric hypercalcemia, familial type III](#)
- [Inherited Kidney Diseases \(Gene Panel\)](#)
- [Nephrogenetics / Nephropathy \(gene panel\)](#)
- [Nephropathies, hereditary \(gene panel\)](#)
- [Thyroid dysgenesis \(38 genes\)](#)
- [Tubulopathy \(gene panel\)](#)

### Related Diseases

- [Familial hypocalciuric hypercalcemia type 3](#)

### Related Gene Panels

- [Endocrine Disorders - Hyper\(Hypo\)parathyroidism \(24 genes\) - ULB](#)
  - [Genetic disorders of Calcium and Phosphate metabolism \(31 genes\) - KUL](#)
  - [Nephropathies, hereditary \(219 genes\) - KUL](#)
  - [Nephropathy panel - UGent](#)
  - [Panel Nephro-ULG-V1](#)
  - [Thyroid dysgenesis \(38 genes\) - VUB](#)
  - [Tubulopathy/Nephrolithiasis \(106 genes\) - IPG](#)
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