

---

**DISEASE:**  
**X-linked epilepsy-learning disabilities-behavior disorders syndrome**

|                     |                                                                                                                                                                                                                                                                                                                                                                        |
|---------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>        | X-linked epilepsy-learning disabilities-behavior disorders syndrome                                                                                                                                                                                                                                                                                                    |
| <b>DESCRIPTION:</b> | X-linked epilepsy-learning disabilities-behavior disorders syndrome is characterized by epilepsy, learning difficulties, macrocephaly, and aggressive behaviour. It has been described in males from a four-generation kindred. It is transmitted as an X-linked recessive trait and is likely to be caused by mutations in the gene encoding synapsin I (Xp11.3-q12). |
| <b>ORPHACODE:</b>   | 85294                                                                                                                                                                                                                                                                                                                                                                  |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                                                                                                                        |
| <b>ANALYTE(S):</b>  | <u>SYN1</u>                                                                                                                                                                                                                                                                                                                                                            |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                    |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                    |

---

Source URL: <http://gentest.healthdata.be/disease/3330>

## RELATED CONTENT

---

### Related Genetic Tests

- [Epilepsy \(gene panel\)](#)

### Related Laboratories

- [Centrum Medische Genetica - UZ Antwerpen](#)

### Related Analytes

- [synapsin I](#)

### Related Gene Panels

- [Rare epilepsy with developmental delay \(> 240 genes\) - UZA](#)

---

Source URL: <http://gentest.healthdata.be/disease/3330>