How to collaborate?

• ONLINE: www.stxbp1.es

BECOME A MEMBER **MAKE A DONATION**

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• MEMBERSHIP REGISTRATION FORM:

I request to register as a member, in accordance with the Statutes of this Association (available in the web):

First name:_____
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Postcode:_____ Province:_____
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Phone 1:_____ Phone 2:_____
E-mail:_____
Date of Birth:_____
ID/Passport:_____
Fee € (*) _____ Type of fee: ☐ annual ☐ quarterly
IBAN:_____
Account Holder:_____

*Mínium 20€ year o 5€ quarter. Signature:

You can submit this form either:
- By e-mail socios@stxbp1.es
- By post to C/Aprestadora, 114, Esc. E, ATC 3ª, 08902, L'Hospitalet de Llobregat (Barcelona) SPAIN
- Or to any of our collaborators.

The personal data collected will be incorporated in the member identification file, whose purpose is to assist the member in all areas. Such file is under the responsibility of Association Syndrome STXBP1, with registered office at C/ Aprestadora 114, Esc E, Atc 3a, 08902 l'Hospitalet de Llobregat (Barcelona, Spain), where the interested party may exercise the rights of access, rectification, cancellation and opposition to it. No data will be transferred to third parties, except in the case of legal obligation. You can consult the additional and detailed information on data protection at www.stxbp1.es/aviso-legal.

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Solidarity Race APPI 2017 Almussafes

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And many more events...

Asociación Síndrome STXBP1

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Asociación Síndrome STXBP1

What is STXBP1 Syndrome?

STXBP1 encephalopathy is a serious neurological disease caused by a mutation of the ***STXBP1*** gene.

It is considered a rare disease. But in spite of its low prevalence, recent studies indicate that it is one of the main causes of "early epileptic encephalopathy".

Genetic alterations in the STXBP1 gene may lead to different onsets of epilepsy during the first months of life, which usually include Ohtahara syndrome in newborns or West syndrome in infants. Sometimes, an infant may present both syndromes or neither of them and have Dravet syndrome or Rett syndrome instead, usually in their atypical forms.

It is frequently accompanied by neurodevelopmental problems, usually including moderate-severe psychomotor developmental delay and autistic spectrum disorder. Although almost all the information available comes from patients with epilepsy, there have been reports of patients with cognitive delay / learning disorder with STXBP1 mutations who do not suffer from epilepsy.

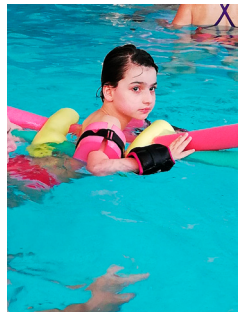
Movement disorders including unsteadiness (ataxia), abnormal muscle function (dystonia), abnormal involuntary movements (dyskinesia) and poor muscle tone (hypotonia) also occur frequently.

Why does this happen?

Genes are DNA fragments that contain instructions for forming a protein. The changes that occur in the genes are called mutations, which may hinder protein formation or lead to an abnormal protein.

The ***STXBP1*** gene contains the information to produce the 'SynTaXin Binding Protein 1'. This protein plays a vital role in releasing chemicals into the brain (neurotransmitters) that are used by brain cells to communicate with each other. A mutation in this gene and the resulting alteration in the protein, leads to abnormalities in brain function causing epilepsy, developmental delay, learning difficulties and behavior problems.

Can it happen again?



Unlike other genetic disorders in which parents are carriers or are also affected by the disease, children with ***STXBP1*** encephalopathy are carriers of "de novo" mutations. That is, the mutation has occurred in a germ cell (egg or sperm) without any of the progenitors being carriers of the mutation. It is therefore very unlikely that it will happen again in future gestations although it is advisable to seek genetic counseling to rule out the possibility of germinal mosaicism.

Can it be cured?

To the present day, there is no cure for this disorder and the treatment is only symptomatic, i.e. limited to treat the manifestations of the disease. However, it is very important to know the diagnosis in order to provide genetic advice, avoid unnecessary explorations, anticipate difficulties and choose the most appropriate medical treatment and pedagogical approach for the child as soon as possible.

Goals

1. Raise awareness about the disease caused by the STXBP1 gene mutation.

2. Provide support, information and advice to affected families.

3. Promote and financially support research projects aimed at finding early diagnosis methods; improving the quality of life and well-being of people affected; developing an effective treatment to mitigate the symptoms and/or cure the disease.

4. Promote and support training of professionals dealing with the STXBP1 syndrome.