

## Questionnaire

### **Study about hyperekplexia in people with CTNNB1 gene mutation**

You can complete this questionnaire for your child with a mutation on the CTNNB1 gene or do it for yourself if you carry also a such mutation.

If you do not have the information to answer certain questions, please do not hesitate to proceed to the next question.

Thanks for your help,

Dr Vincent Bulot (Neurology intern), Dr Laure Mazzola (Neurologist) et Dr Francis Ramond (Geneticist).

**Initials Name/First name of the person carrying the mutation : \_\_\_\_ / \_\_\_\_**

**Age :**      **Sex :** ☐ Male      ☐ Female

#### **• Pregnancy and birth**

◦ *Were there any difficulties or abnormalities during pregnancy?*

☐ Yes

☐ No

If yes, which ones ?

.....  
.....

◦ *Were there any difficulties at birth or during the first months of life ?*

☐ Yes

☐ No

If yes, which ones?

.....  
.....

#### **• Developpement of your child**

◦ *How is his walking now ?*

☐ Normal

- ☐ Unstable
- ☐ He/She spreads legs when walking to avoid falling
- ☐ Hampered by stiffness of the legs
- ☐ Need of assistance (crutch, wheelchair)

Details, if necessary :

.....  
 .....

◦ *If he/she can walk, at what age did he/she take his/her first steps without help ?*

☐ Age of acquisition de la marche = .....

◦ *At what age did he/she sit alone without assistance ?*

☐ Age of sitting = .....

◦ *How is his/her language now ?*

- ☐ He/She does not talk
- ☐ A few sounds or syllables
- ☐ A few words
- ☐ A few sentences, poorly constructed
- ☐ Normal

Details, if necessary:

.....  
 .....

◦ *If he/she can talk, at what age did he/she say his/her first word ?*

☐ Age of his/her first words = .....

◦ *Did he/she make sentences before going in kindergarten ?*

- ☐ Yes
- ☐ No

◦ *What does he/she understand ?*

☐ Complex orders (to show a color, to give a specific object...)

☐ Simple orders (to raise one's hand, to close the eyes...).

☐ Some simple situations (time to eat, doing a hug...)

◦ *About social interactions :*

☐ He/She plays with other kids

☐ He/she avoid contact with others

☐ He/She does not talk with other kids (brothers and sisters, schoolmate)

☐ An autism spectrum disorder was diagnosed

Details, if necessary :

.....  
.....

◦ *About his/her behaviour :*

☐ He/She is rather quiet.

☐ He/ She easily gets angry by having intense anger attacks and/or crying.

☐ He/She can be aggressive.

☐ He/she has already harmed himself/herself (scratches, bites, etc.).

☐ He/She is interested in very specific things (trains, spinning objects...)

☐ He/She has repetitive behaviors (clapping one's hand, turn on oneself...)

Details, if necessary :

.....  
.....

◦ *At school :*

☐ He/She does not attend school.

☐ He/She goes to an adapted structure (Medical and Education Institute...).

☐ School results are good.

☐ He/She has learning difficulties.

☐ He/She can write.

☐ He/She can read.

☐ He/She can do simple math (addition, subtraction, multiplication).

Details, if necessary:

.....  
.....

• **Ophtalmology**

◦ *Does your child have eye problems ? (here some ophtalmologic troubles described in CTNNB1 syndrome : hypermetropia, strabismus, retinal detachment, retinal hemorrhages, opacities of the lens and vitreous )*

☐ Yes

☐ No

Details, if necessary :

.....  
.....

• **Hyperekplexia**

◦ *Did your child experience in the first few months after birth episodes where he/she became completely stiff up to apnea (transient cessation of breathing)?*

☐ Yes

☐ No

If yes, was it triggered by specific events (sudden noise, touch, bath...)? .....

◦ *Does he jump in an exaggerated way to surprise or noise ?*

☐ Yes

☐ No

If yes, what situations cause these startles ?

☐ Sudden noise

☐ Touch (by surprise)

☐ Emotion (joy, sadness)

☐ Others : .....

◦ *Do the startles decrease or even disappear as the event repeats itself? ( for example, if the noise that triggers the startle repeats itself, does your child end up no longer startling ?)*

☐ Yes

☐ No

◦ *Do these startles provoke falls ?*

☐ Yes

☐ No

If yes, did these falls cause injuries ?

☐ Yes

☐ No

If yes, is your child now very afraid of falling as soon as he walks ?

☐ Yes

☐ No

◦ *Does your child lose consciousness during the startle ?*

☐ Yes

☐ No

◦ *Did you try treatments for these startles ?*

☐ Yes

☐ No

If yes, which ones and were they effective ?

.....

◦ *At what age did these startles appear ? .....*

◦ *How is it now evolving (improvement, worsening, stability) ?*

.....

◦ *Did your child have seizures ?*

☐ Yes

☐ No

**• Medical tests (If you do not have the results of these tests, you can skip this part)**

◦ *Did your child have a MRI (Magnetic Resonance Imagery) ?*

☐ Yes

☐ No

If yes, was it normal ? ☐ Yes ☐ No If no, can you copy the abnormalities found in the report or attach the report .....

◦ *Did your child have an electroencephalogram (EEG) ?*

☐ Yes

☐ No

If yes, was it normal ? ☐ Yes ☐ No If no, can you copy the abnormalities found in the report or attach the report .....

**• Medical follow-up**

◦ *Is your child supervised by a neurologist ?*

☐ Yes

☐ No

If yes, can you write down his/her name +/- contact information so we can get in touch with him/her if we need more information :  
.....

◦ *Is your child supervised by a geneticist or has your child consulted with a geneticist at least once ?*

☐ Yes

☐ No

If yes, can you write down his/her name +/- contact information so we can get in touch with him/her if we need more information :  
.....

Thank you for your participation.

We will inform you of the results of the study as soon as possible. Data collection is spread over 2021 and results are expected to be reported later this year.

Completed questionnaire on \_\_/\_\_/\_\_\_\_