1 Functional abilities of children and adults with FOXP1 syndrome - dr. Saskia Koene

1. Aim of this study

To provide parents and practitioners with clear information about functional, neurobehavioral and medical complications of FOXP1 syndrome. (Figure 1)

Figure 1:
Terms in which parents describe their child with FOXP1 syndrome

2. Study procedure

- Receive information about study
- Informed consent
- Verify diagnosis
- Online questionnaire by parents (2-3h)
- Receive study results

Enuresis is present in 45% of individuals.

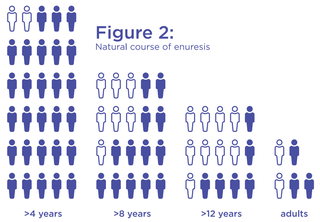


Figure 2:
Natural course of enuresis

Dr. Koene

Want to contribute?

For more information about this study, please send an email to foxp1@lumc.nl (see QR code)

3. Results

Medical phenotype in 40 individuals:
"Symptoms that are quite specific to FOXP1 syndrome include strabismus, hypermetropia, cryptorchidism, enuresis and behavioral problems." (Figure 2)

Will be submitted shortly.

Neurobehavioral phenotype
"The neurobehavioral Gestalt in FOXP1 syndrome is composed of repetitive behavior, including the need for "security objects" in the hands, a complex sensory profile with both easy overstimulation (specifically auditory hyperreactivity) and sensory seeking, attention deficit, and hyperactive, obsessive and compulsive behavior."

Will be submitted end of 2023.

4. Functional abilities for the future **(in preparation)**

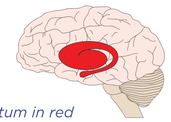
How many individuals are able to...
...be alone for 30 minutes?
...recognize dangerous situations?
...cook a simple meal?
...go on public transportation alone?
...be on time for a meal?
...tie shoelaces?
...make breakfast

Analysis end of 2023.

2 Understanding the molecular mechanism of the FOXP1 syndrome - prof. Willeke van Roon and dr. Elena Daoutsali

1. Aim of this study

To discover what is the mechanism of FOXP1 syndrome.
The FOXP1 gene is very important for the development of a brain area called "striatum". This brain area is important for motor control, learning, action selection and cognitive function.



Striatum in red

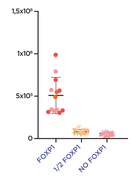
2. Study procedure

To study the function of FOXP1 in the cellular level, we need to remove the FOXP1 gene from the genome and look at how this will affect other genes. We will use cells that have FOXP1, cells that lack half of FOXP1 and cells that lack all FOXP1, like the glasses below.

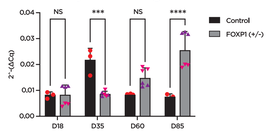
3. Results

We used cells carrying FOXP1, 1/2 FOXP1 and no FOXP1 and we grew cells in a 3D emvironment to better mimic the human brain. This structures are called "brain organoids". We found that lack of FOXP1 affects the size of these organoids and the differentiation of "early" neurons to "mature" neurons.

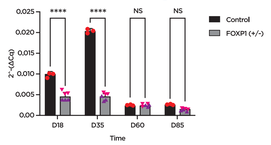
organoid size



"early neuron" gene

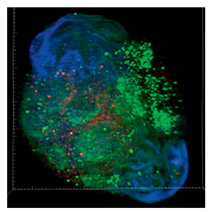


"mature neuron" gene



4. Future studies

In our future studies we will use the FOXP1, 1/2 FOXP1 and no FOXP1 cells, but also cells that have a known mutation in FOXP1. Using these cells we will generate "brain organoids" to further study FOXP1 mechanisms. Using the brain organoids we can visualize different proteins that can be affected by FOXP1.



Dr. Daoutsali

Prof. van Roon

Research initiated in The Netherlands

Kind Beter

(Foundation Child Well)
Two main goals
- Collecting funds for research
- Connecting parents in NL and Belgium

http://www.kindbeter.nl

Other researches where FoxP1 is included:
- Sleep research
- Speech therapy research
- Blatter control research
- Neurological profile research

3 Navigating information on Facebook about FOXP1; balancing between hope and uncertainty - dr. Annemiek Linn

1. Aim:

Caretakers of individuals diagnosed with a rare disease face various obstacles, with a lack of information being the most significant one. As a consequence, caregivers turn to social media to fulfill their informational and social support needs. The aim of this study is to understand how caretakers of individuals diagnosed with FOXP1 experience the use of social media to comprehend and cope with this diagnosis.

2. Method:

Online interviews were conducted with 20 caretakers of individuals diagnosed with FOXP1. Maximum variation was sought concerning the age of the diagnosed individuals, date of diagnosis, and education level. Thematic analysis was used to qualitatively analyze the anonymized transcripts using ATLAST.TI.

3. Results:

Preliminary findings suggest that the use and significance of Facebook evolve over time. Initially, caretakers primarily utilize Facebook to acquire knowledge about FOXP1 and connect with similar caretakers. During this phase, the information and experiences shared by other caretakers offer hope, particularly after receiving negatively biased information from their doctors.
As the years go by, caretakers feel compelled to give back to the community by sharing their experiences, aiming to provide hope to others.

4. Conclusion:

The insights obtained from this study will help highlight important patient values and enable the medical and scientific communities to enhance communication with and understanding of the patients under their care.

Dr. Linn

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4 Mapping the collective wisdom of online rare disease communities: a Patient-driven Algorithm (PIPA project) - dr. Annemiek Linn

1. Aim:

This project aims to collect, bundle, and analyze the information caretakers have about their child, by developing an open-source algorithm that maps the collective wisdom about rare diseases on social media. This project contributes to the knowledge of rare diseases and allows individuals to understand better and manage their rare disease.

2. Method:

The project includes the donation of Facebook usage data by participants exercising their own rights to download their data (GDPR article 15). The PORT software will be used to ensure (1) privacy protection and (2) meaningful data extraction14. Privacy protection is ensured by informed consent after which individuals download their data and decide whether and how to donate their data.

3. Impact:

This project provides an important contribution to the knowledge of a specific RD (FOXP1 and paves the way for more knowledge on other RDs. The data obtained can be used to prepare individuals what they can expect from their RD, practical support, and which treatments have worked. The physician can use the data to inform newly diagnosed individuals and steer future epidemiological analyses. The data can also be used to train physicians with the necessary competencies for adequate care of these individuals. On a societal level, the blueprint can be used to map the collective wisdom of other RDs. In addition, the reuse of the algorithm makes cost-efficient longitudinal research into RDs possible since the model can automatically and consciously be applied to new data. Insights can also lead to better recommendations to strengthen the infrastructure to support and treat individuals with RDs. The results of this study can also be used as a fundamental basis for new grant applications within the domain of RD research.

**Do you want to receive more information on how to donate your Friends and Family of FOXP1 Facebook data?
Scan the QR code to fill out your contact details.**

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