

# NRXN1 Study

NOVEMBER 2019

## The project so far

On behalf of the Autism Research group at Trinity College Dublin we would like to thank you very much for taking part in our research on NRXN1 deletions. To date over 50 people have participated, which has allowed us to gather lots of useful information. Volunteers have provided DNA samples, clinical information, cognitive data as well as images of their brains. From these measures we are beginning to get a better understanding of how NRXN1 deletions may affect brain development, cognition and mental health.

NRXN1 deletions were associated with differences in attention, social cognition (mental processes underlying social interactions) and executive function (e.g. planning, self-control). The data also showed connectivity differences in brain regions associated with these skills. Dr. Jackie Fitzgerald has presented some of the brain imaging and cognitive results at conferences in Europe and the U.S.A.

While these results are preliminary, they have improved our understanding of how NRXN1 deletions may impact day-to-day functioning. These results also highlight the importance of examining how rare genetic variation affects brain development. This work has led to exciting opportunities to develop our knowledge of NRXN1 deletions on a much larger scale.

### Who are we?

We are the Autism Research group at Trinity College Dublin. You might have met some of the researchers working on the NRXN1 deletion study when you last came to visit. We have new members of the team who are looking forward to continuing the research.



Prof. Louise Gallagher

Dr. Ciara Molloy

Dr. Jackie Fitzgerald

Dr. Lorna Lopez

Dr. Nadia Bolshakova

Thomas Dinneen

Matthew O'Sullivan

Richard O'Conaill



Trinity College Dublin  
Coláiste na Tríonóide, Baile Átha Cliath  
The University of Dublin

# Going forward

Our research team recently joined a large European consortium, called AIMS-2-TRIALS. The consortium consists of research groups from over 40 institutions across Europe, who are working together to explore different aspects of autism research. The goal of the consortium is to improve our understanding of how autism develops, how it varies among individuals, and how to predict who has or may develop certain autistic characteristics. Some groups are also focusing on the development of new diagnostic and therapeutic strategies for autism and related neurodevelopmental conditions.

As part of AIMS-2-TRIALS, we are expanding our research on NRXN1 deletions to collect some exciting new measurements which we hope will help us learn more about characteristics of NRXN1 deletions. We are working with our colleagues in King's College London to study different rare genetic conditions associated with autism. Together we are conducting the Synaptic Gene Study.



Dr. Jackie Fitzgerald is also conducting research aimed at understanding parents' experience of their child and possibly other family members having a genetic variation such as a NRXN1 deletion. This involves a short interview (either over the phone or in person). You can contact Jackie at: [fitzgeje@tcd.ie](mailto:fitzgeje@tcd.ie) or 089-4651563.

**We will be getting in touch with all of the families who have participated already very soon. We are hoping you will come back to see us for some more assessments and interviews!**

We are very excited to invite our participants to come and see our recently renovated research facilities.



We have a new electroencephalography (EEG) system, which measures brain activity by placing electrodes the surface of the head. We also have brand new eye-tracker, which is a special camera that lets researchers know what people like to look at while they are watching videos and looking at pictures.

## Our research goals

By expanding our research, we hope to learn more about how NRXN1 deletions affect individuals and their families. Through working with our collaborators and with the families who participate in the studies, we wish to get a better understanding of the biology of rare genetic variants and how this relates to autism and neurodevelopmental disorders. We are also interested in exploring how genetic diagnoses impact families and how we might provide better support to families in the future.

### More Information

If you have any questions in the meantime, please don't hesitate to get in touch with us: [autism@tcd.ie](mailto:autism@tcd.ie)

TCD Autism Research Group's website: <https://www.tcd.ie/medicine/psychiatry/research/neuropsychiatric-genetics/autism/>

AIMS-2-TRIALS website: <https://www.aims-2-trials.eu/>