CARDIFF UNIVERSITY
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MRC Centre for Neuropsychiatric Genetics and Genomics

ECHO Study

The ECHO study aims to understand the life course of individuals with 22q11.2 Deletion Syndrome (22q11.2DS). We have been running this study since 2010 and close to 200 individuals with 22q11.2DS have taken part to date. We are also part of an international research group for 22q11.2DS, working with over 22 research sites worldwide to understand more about the syndrome.

We have put together this brief summary of our findings for parents and patient support charities to learn more about our work.

Psychopathology and cognition

This study investigated the mental health and intelligence of the first 80 children (average age 10) with 22q11.2DS who took part in the ECHO study and compared findings to 39 siblings who did not have the syndrome.

- Over 54% of children with 22q11.2DS met criteria for a mental health condition, compared to 10% of siblings.
- On average, children with 22q11.2DS had an IQ of 77, 30 points less than their siblings (average IQ = 109).
- Children with 22q11.2DS were more likely to perform worse on tests of attention, planning and reaction time.
- We did not find a relationship between the presence of a childhood mental health condition and a child's intelligence.

Niarchou, M.et al. 2014. Psychopathology and cognition in children with 22q11.2 deletion syndrome. British Journal of Psychiatry 204(1), pp. 46-54

How parents source information on the psychiatric manifestations of 22q11.2DS

This study investigated how parents of a child with 22q11.2DS first learned about different aspects of the syndrome.

- 78% were aware of the risk of mental health problems, but they were more likely to have found out about this from the internet (42%) than a clinician (27%).

This highlights the need for clinicians to provide more information and guidance to families regarding mental health in 22q11.2DS.

van den Bree, M.et al. 2013. The internet is parents' main source of information about psychiatric manifestations of 22q11.2 deletion syndrome (22q11.2DS). European Journal of Medical Genetics 56(8), pp. 439-441.

ADHD in children with 22q11.2DS

We wanted to find out whether ADHD is similar or different in children with 22q11.2DS to children without 22q11.2DS. We compared the symptoms of 44 children with 22q11.2DS and ADHD to 600 children without 22q11.2DS who had been diagnosed with ADHD.

- Children with 22q11.2DS were more inattentive, but less hyperactive.
- Children with 22q11.2DS were more anxious, but had less conduct issues.
- Only 2% of children with 22q11.2DS were receiving treatment for ADHD.
- Overall, the clinical presentation of ADHD in 22q11.2DS looks different to typical ADHD, potentially leading to an under-recognition of ADHD in this patient group by clinicians.

Niarchou, M.et al. 2015. The clinical presentation of attention deficit-hyperactivity disorder (ADHD) in children with 22q11.2 deletion syndrome. American Journal of Medical Genetics. Part B 168(8), pp. 730-738

International contributions

We have contributed to a range of international studies, which have found:

- Maternal age is not related to the incidence of 22q11.2DS, meaning older mothers are not more likely to have a child with the syndrome. (*Delio, M. et al, 2013*)
- ADHD is the most frequent mental health disorder/condition in children with 22q11.2DS (37.1%), and more likely to occur in males. (*Schneider, M. et al, 2014*)
- Anxiety disorders are more prevalent than mood disorders at all ages, particularly in children and adolescents, and more likely to occur in females. (*Schneider, M. et al, 2014*)
- Psychotic disorders (symptoms can include hearing voices, seeing visions and delusional thinking) were present in 41% of adults over age 25 with 22q11.2DS.[2] (*Schneider, M. et al, 2014*)
- Bipolar disorder and alcohol and drug addictions are rare in 22q11.2DS. (*Schneider, M. et al, 2014*)
- Subthreshold psychosis, mild psychotic-like symptoms which are not severe enough for a psychiatric disorder, was present in almost a third of study participants with 22q11.2DS. (*Weisman, Omri, et al. 2017*)

For full paper references please contact us using the details below.

Our funders, partners and friends

We would like to thank everyone who has participated in our research, as well as the charities, funders and NHS medical genetic clinics that have supported the study.

Get in touch

If you would like to learn more about our study, including how to participate, please contact: **echo@cardiff.ac.uk** or phone **029 2068 8039.**