VOLUNTEERS NEEDED FOR RESEARCH INTO GENETIC DELETIONS AND DUPLICATIONS

The genetic material of every individual can be different from other individuals, even if they are family members. Compared to others, some people may have extra sections of genetic material, or fewer sections of genetic material. These are called copy number variations (CNVs). A CNV can be either a duplication (where an individual has more of the genetic material than most other people) or a deletion (where they have less than most other people).

Children with copy number variations (CNVs) can experience learning difficulties. They may also experience behavioural problems at home and in school and may find it more difficult to make friends. Sometimes, their thinking and speech may be difficult to follow or may seem unusual to others. It is not clear at the moment to what extent children with different types of CNVs are at risk for these problems. Researchers also don't know to what extent CNVs play a role in the behavioural and thought problems some children can experience. It is possible that the learning difficulties these children can have contribute to their risk of behavioural and thought problems. However, if and to what extent they do is not understood.

Understanding more about the relations between learning, behavioural and thought problems in children with genetic deletions may lead to more effective methods of treatment and prevention, thus improving the lives of children and families affected by these disorders.

The aims of this study are to establish:

- How frequent learning disability, behavioural problems and thought problems are in children with CNVs.
- The relationships between learning disability, behavioural problems and thought problems in children with CNVs.

We are particularly interested in hearing from families who have children with these CNVs: **1q21.1 deletion and duplication, 2p16.3 exonic deletion, 3q29 deletion, 7q11.23 duplication, 9q34.3 deletion (Kleefstra Syndrome), 11q14.1 deletion, 15q11.2 deletion, 15q11-13 duplication, 15q13.3 deletion and duplication, 16p11.2 duplication, 16p11.2 distal deletion, 16p13.11 deletion & duplication, and 17q12 deletion & duplication, and 22q11.2 deletion & duplication.**

The study has ethical approval and is conducted by the Institute of Psychological Medicine & Clinical Neurosciences at Cardiff University. Dr. Marianne van den Bree is the principal investigator.

Your participation will involve:

- Assessment of the child's symptoms via clinical interview with the parent.
- Short interview with the child about how they experience themselves and others.
- Assessment of the child's cognitive ability. This involves completing a number of puzzles.
- Blood or saliva sample from the parents and the child for DNA extraction

Participation in this project is entirely voluntary. All families who take part will be reimbursed for their time. All information gained from the study will be confidential. You are free to withdraw from the study at any stage.

If participating families like to, we can provide their clinician with some of the information gathered in the study which may be helpful for treatment. We hope that the project will provide greater insights into the development of CNVs and this may in turn contribute to the development of new treatment strategies in the future.

Your family may be eligible for the study if:

- You have a child who is 6-17 years who has been diagnosed with a deletion or duplication syndrome listed above
- If your child has a sibling who does not have a CNV we would also ask them to take part. However, families of children with a CNV who do not have a sibling can still take part.

Please note if you live outside the UK we could conduct the interview with you via skype, but would not be able to carry out the cognitive assessments with your child.

If you would like to take part or require more information please contact:
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