

## Understanding behavioural characteristics in Sotos Syndrome

The Cross Syndrome Research Project is a large scale project that is being carried out by a specialised team of researchers at the Cerebra Centre for Neurodevelopmental Disorders based at the University of Birmingham (UK); supervised by Professor Chris Oliver and Dr. Jo Moss. Initially set up in 2003, the Cross Syndrome Project has since strived to address the questions raised by individuals with rare genetic syndromes and their families. The information provided from those individuals and families who take part in this project can help us to address these questions and learn more about rare genetic syndromes. We strongly believe that a thorough understanding of the behavioural characteristics of Sotos syndrome is essential to support individuals with Sotos syndrome and their families in the best way possible. We would like to give a huge thank you to everyone who has taken part in this research project, we are very grateful for your continued support for our work.

In 2010, we invited individuals with Sotos syndrome and their families to take part in the Cross Syndrome Project. This research was conducted in collaboration with Dr. Trevor Cole at the Clinical Genetics Department at Birmingham Women's Hospital and with the Clinical Genetics Department at Liverpool Alder Hey hospital. We wanted to examine the presence and prevalence of behavioural characteristics and social-communication skills in individuals with Sotos syndrome and then see how these characteristics compare to that of other syndromes, such as Autism Spectrum Disorder (ASD), Down's syndrome and Prader-Willi syndrome. This cross syndrome approach is important in helping us to understand the behavioural profile of Sotos syndrome and allows us to position Sotos syndrome relative to other groups in which the associated behavioural characteristics are well described.

In this study, we aimed to explore: 1) the presence of challenging behaviours in Sotos syndrome, 2) the proportion of people with Sotos syndrome showing ASD-like behaviours, and 3) the presence of repetitive behaviours in individuals with Sotos syndrome.

### **Who took part in the study?**

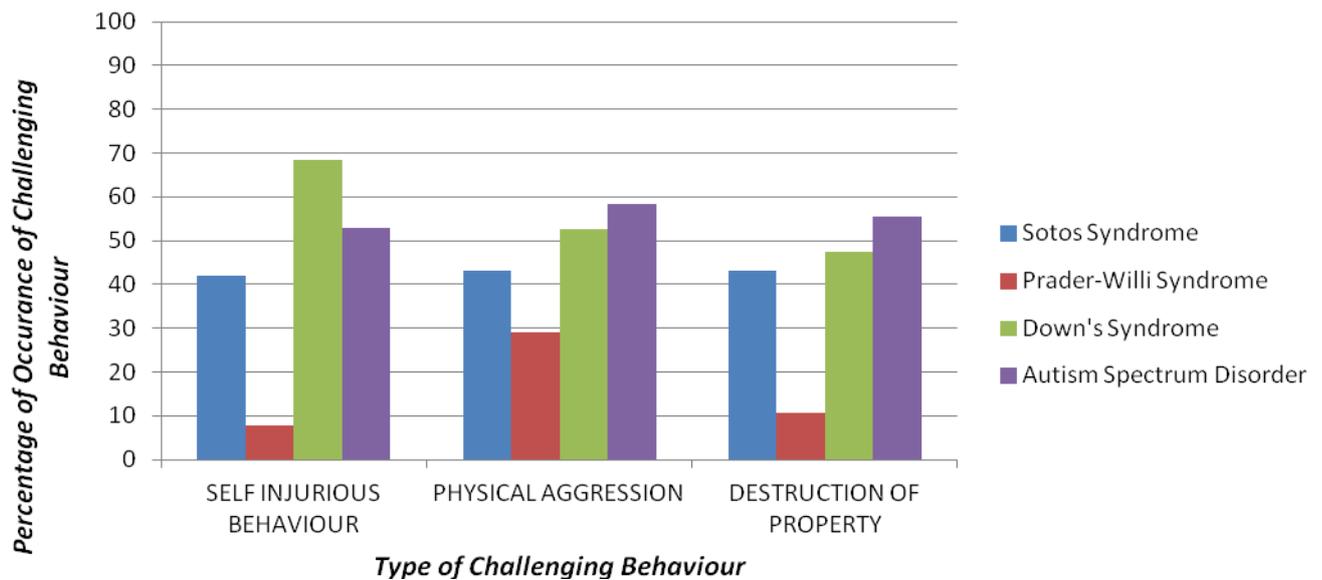
We invited individuals with Sotos syndrome and their families who were registered with the Child Growth Foundation (CGF) and those known to Clinical Geneticists working in Birmingham and Liverpool to take part in the study. In total, 38 individuals with a confirmed diagnosis of Sotos syndrome decided to take part. Individuals with Sotos syndrome were aged between 6 years and 43 years and included 25 males and 13 females.

### **Challenging behaviour in Sotos syndrome:**

Our first aim was to examine the presence of challenging behaviours including self-injurious behaviour, aggressive behaviour and destruction of property in children and adults with Sotos syndrome. Please note that when we use these terms to describe different types of challenging behaviours we do not infer any intention to harm.

We found that self-injurious behaviour, aggressive behaviour and destruction of property occurred in 42 – 43% of those with Sotos syndrome. These rates were higher than that observed in individuals with Down’s syndrome but lower than that observed in individuals with Prader-Willi syndrome and ASD. The findings suggest that while challenging behaviour is common in Sotos syndrome, many individuals with the syndrome do not to show these behaviours.

Figure 1 below shows the percentage of individuals with Sotos syndrome, Prader-Willi syndrome, Down’s syndrome and ASD reported to show challenging behaviours such as self-injury, physical aggression and destruction of property.



*Figure 1 –The occurrence of challenging behaviour in individuals with Sotos syndrome, ASD, Down’s syndrome and Prader Willi syndrome*

### **Characteristics of Autism Spectrum Disorder:**

Our second aim was to examine the proportion of individuals with Sotos syndrome who showed ASD-like behaviours. We found that a relatively high proportion (almost 70%) of individuals with Sotos syndrome showed ASD-like characteristics. This suggests that many individuals with Sotos syndrome might experience some social and communication difficulties that are similar to those observed in individuals with ASD who do not have Sotos syndrome. The findings do not necessarily suggest that individuals with Sotos syndrome should receive a diagnosis of ASD (although this may be appropriate in some cases) but indicate that there may be some shared characteristics with individuals with ASD. Interestingly, when compared on levels of mood, interest and pleasure, those with Sotos syndrome experienced greater levels of interest and pleasure in activities and a more “positive” mood than individuals with ASD.

### **Repetitive Behaviour:**

Our final aim was to evaluate repetitive behaviours in individuals with Sotos syndrome. An interesting finding was that compulsive behaviours (such as cleaning, tidying, rituals and lining up objects) and repetitive speech (such as repetitive questions, phrases or signing) were seen to be

significantly more frequent in individuals with Sotos syndrome compared to those with Down's syndrome. Individuals with Sotos syndrome showed very similar levels of these types of repetitive behaviours compared to those with Prader-Willi syndrome and ASD, suggesting that these behaviours may be a source of difficulty for some individuals with Sotos syndrome. However, they showed lower levels of stereotyped behaviour (such as repetitive movements or repetitive use of objects) and insistence on sameness behaviour (such as preference for routine or having objects arranged in particular way) than these groups, indicating that these behaviours are not frequently observed in individuals with Sotos syndrome.

Figure 2 below shows how those with Sotos syndrome compared to individuals of other syndromes on the different forms of repetitive behaviours.

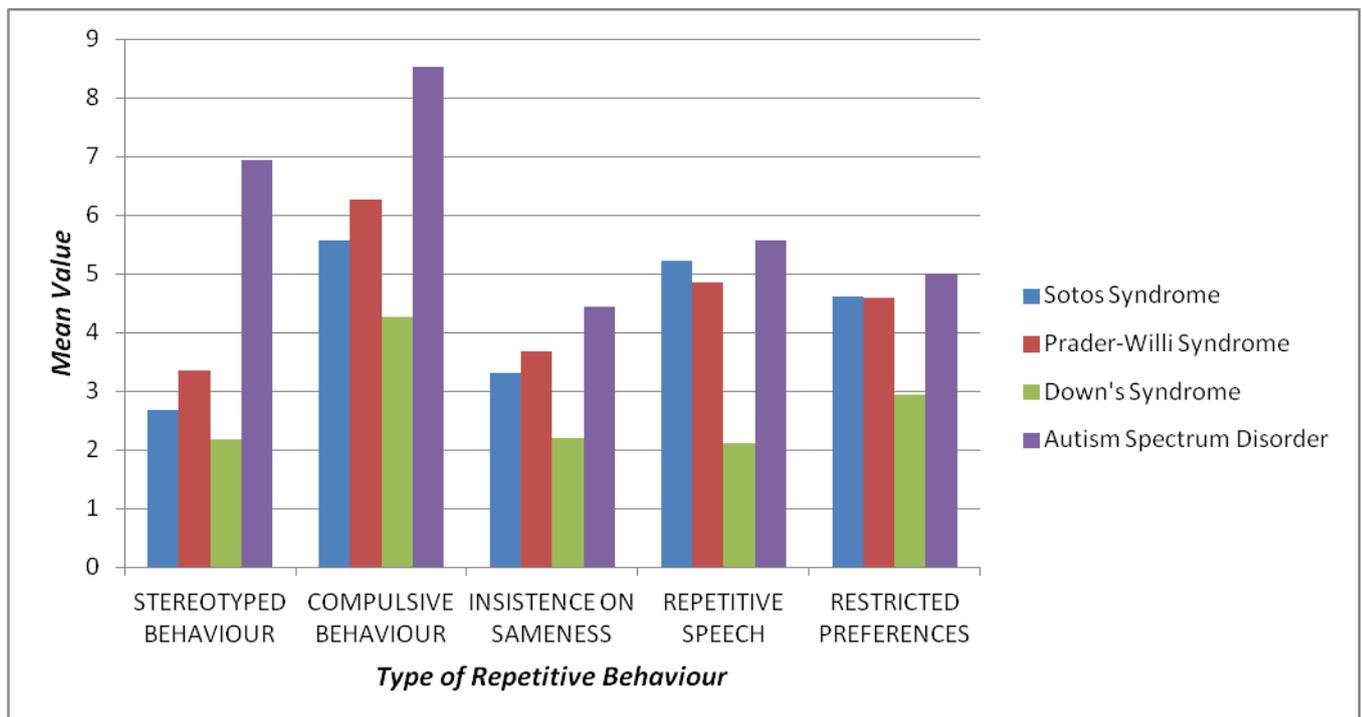


Figure 2: The average score on five types of repetitive behaviour in individuals with Sotos syndrome, ASD, Down's syndrome and Prader Willi syndrome.

### Summary of research findings:

Our research has shown that:

1. Challenging behaviours (self-injurious behaviour, aggressive behaviour and destruction of property) occur in approximately 42% of individuals with Sotos syndrome. This is higher than that observed in individuals with Down's syndrome but lower than that observed in individuals with ASD and Prader-Willi syndrome.
2. ASD-like behaviours are common in individuals with Sotos syndrome. This means that they may share some (but not all) characteristics with individuals with ASD and may find social situations difficult to understand and navigate.
3. Compulsive-like behaviours and repetitive speech are particularly common in individuals with Sotos syndrome. These behaviours may be important in understanding some of the challenging behaviours that were reported to occur. However, further research is needed to better understand this.

## **The next steps?**

The team at the University of Birmingham are now planning the next phase of this research project. We would like to understand more about how people with Sotos syndrome develop and progress as they get older. To help us to do this, we will be contacting those individuals and families that took part in this study again to find out how they are getting on, so look out for some information in the post soon!

If you have any questions or would like to hear more about this research, please contact Prof. Chris Oliver at [cndd-enquiries@contacts.bham.ac.uk](mailto:cndd-enquiries@contacts.bham.ac.uk). You can also now find us on our facebook page for continuous updates on the different projects we are currently work on and interesting information, articles and events surrounding the syndrome groups we work with:

 <https://www.facebook.com/pages/The-Cerebra-Centre-for-Neurodevelopmental-Disorders/230197213724784>

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