

WELCOME!

Welcome to the CHARGE syndrome research news letter for Spring 2022. We are very excited to share with you all the work we've been doing, how the research is progressing and information about the launch of our new sleep research!

THE RESEARCH

The project is designed to investigate sleep, pain and behaviour in children with CHARGE syndrome. The results from our project will provide new and valuable information about CHARGE syndrome, helping us to improve things for people with CHARGE syndrome in the future.



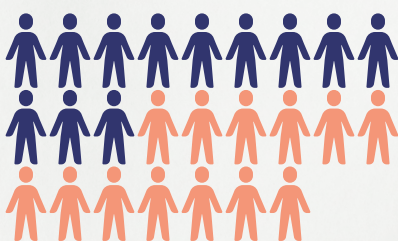
Q1: WHAT BEHAVIOURS HAVE BEEN REPORTED IN CHARGE SYNDROME AND HOW COMMON ARE THEY?

Study one: Meta-analysis

A meta-analysis is a statistical process used to bring together the best available evidence from the published literature. We used meta-analysis to find out what behaviours were reported in CHARGE syndrome and how commonly they occurred

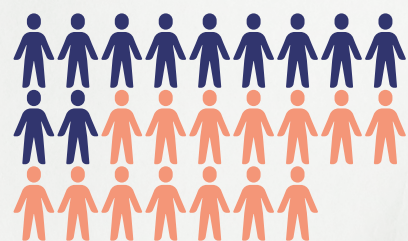
RESULTS

The most common behaviour difficulty was aggressive behaviour 48%



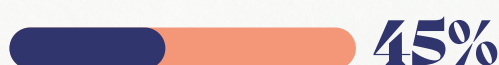
RESULTS

The second most common behaviour difficulty was self-injurious behaviour 44%



RESULTS

Around 45% of people with CHARGE Syndrome have difficulties with sleep



What do we need to know now?

Behaviours such as self-injury and aggression have a negative impact on quality of life and increase the risk of isolation for the individual and their family. Early intervention is crucial to improve outcomes, but for interventions to be successful we need to understand the function or purpose of the behaviours.

Q2: WHAT ARE THE POTENTIAL FUNCTIONS OF BEHAVIOURS THAT CHALLENGE AND HOW DO THEY RELATE TO EVERYDAY SKILLS AND CHALLENGES?

Study Two: Function of Behaviour Interview

Study two is an interview study that parents or caregivers who have a child with CHARGE syndrome (4–15 years) can complete over the phone or through video conferencing if outside the UK. As the interview focuses on behaviours that challenge, we ask that children have experienced at least one behaviour difficulty in the last month.

Given the critical role for early intervention the first phase of our research has focused on children aged 15 years and younger

Reasons/functions of behaviour

Behaviours happen for a reason. However, this does not mean that they are deliberate. Many of the behaviours people show, particularly in times of stress or frustration, are almost automatic. By identifying the times and situations that behaviours occur, as well as the consequences of the behaviour, we can begin to understand the underlying function.

The most common functions/purposes for behaviours that challenge include attention, escape from demands, self-stimulation, physical discomfort, access to tangibles, escape from social attention, reinstatement of routine/repetitive behaviour and sensory sensitivity. We explore these functions in our interview study and provide the results to parents in a feedback report.

Adaptive behaviour

The interview also asks questions about everyday skills and challenges, referred to as adaptive behaviours. Adaptive behaviours are the day-to-day activities necessary to take care of oneself and to get along with others. Adaptive behaviour represents the typical performance, rather than the potential ability of the individual – what a person actually does as opposed to what a person is capable of doing.

It is important for researchers to think about adaptive behaviour when looking at the functions of challenging behaviour. If a child requires support with day-to-day activities or has challenges communicating they may behave in ways that help them get their needs met. While the needs are reasonable, the person may need help learning different ways to communicate them

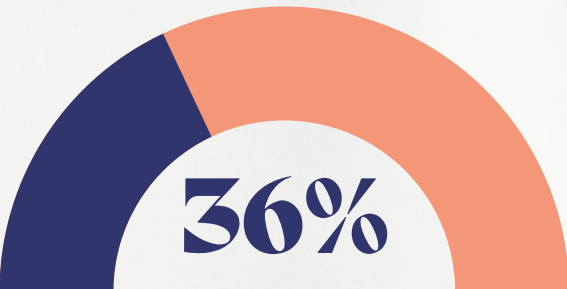
How to take part

You can register to take part in the interview study at <https://tinyurl.com/4wv5yhz7>



Progress to date

Since August 2021, 18 parents and caregivers from all around the world have taken part. Our target is 50!



Q3: DO BEHAVIOURS THAT INDICATE PAIN DURING THE DAY DISRUPT SLEEP QUALITY AT NIGHT? DOES SLEEP QUALITY IMPACT BEHAVIOUR THE NEXT DAY?

Study Three: 10 day sleep assessment

Study three is a 10-day sleep assessment. This is the first time we have used this type of assessment to learn about sleep in CHARGE syndrome and we are very excited!

We invite children with CHARGE syndrome aged 4–15 years and their parents/caregivers to take part in this study. As the research looks at behaviour, we ask that children have experienced at least one behaviour difficulty in the last month. We would love to hear from families whose children do and do not experience sleep difficulties or chronic pain.

Actiwatch

To take part in the study, we ask children to wear an actiwatch for 10 days. We also send out dummy watches for children to try to see if they would like to take part in the study



Research Diary

We provide parents with a research diary with brief sections to be completed twice a day for the 10 days. Full training is provided, and we are only a phone call away!

Further details

Everything needed to take part in the research is posted out to you. As such, we are initially only recruiting families in the UK.

How to take part

You can register your interest in taking part in the interview study by email to:

Andrea Thomas

ATT644@bham.ac.uk



Recruitment Aim

This is a brand new study! Recruitment is opening in March 2022.

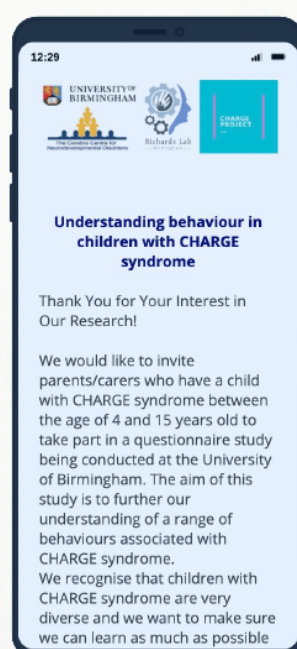
Our target is 50 families

Q4: WHAT ARE THE SKILLS AND CHALLENGES CHILDREN WITH CHARGE SYNDROME EXPERIENCE, AND HOW DO THEY COMPARE TO CHILDREN WITH DIFFERENT GENETIC/CHROMOSOMAL CONDITIONS?

Study Four: Online Questionnaire

Study three is an online questionnaire. We invite English speaking parents/caregivers from around the world who have a child with CHARGE syndrome (4–15 years) to complete this online questionnaire.

The questionnaire takes around 45 minutes to complete, but if you leave your browser window open you can always come back and finish it later!



Further details

If you live in the UK and you would prefer to complete a paper copy of the questionnaire, please email

Andrea Thomas

ATT644@bham.ac.uk

How to take part

To learn more, or to take part in this study please go to:
<https://tinyurl.com/ycbr8dad>



Progress to date

So far 28 parents and caregivers from all around the world have taken part. Our target is 70!

Interested in taking part in future research?

If you are interested in taking part in CHARGE Syndrome research, but you or the person you care for is over 15, or if you would just like to hear about future research opportunities, you can learn more about joining our regular participant database here:

<https://tinyurl.com/h4ez4rs3>



If you have any questions please contact Andrea Thomas at ATT644@bham.ac.uk

UK FAMILY SUPPORT GROUP WEBINAR NOVEMBER 2021

Professor Chris Oliver, Emeritus Professor of Neurodevelopmental Disorders at the University of Birmingham discusses how extending what we already know about behaviour difficulties in children with neurodevelopmental conditions can help us to understand factors that can influence the severity and persistence of these behaviours.

Self-injurious, aggressive and destructive behaviours are common in children who experience delayed development, particularly when

communication is affected. In this talk, Professor Oliver discusses the traditional approach used to understand behaviour difficulties. This approach is called the associative learning account. It is used to explain the presence of difficult behaviours with reference to events in the child's environment. Professor Oliver will then present research data from children with developmental delay and rare genetic conditions, which show the importance of extending these traditional approaches to include factors such as pain and painful health conditions that can influence the presence and severity of difficult behaviours, both directly and via sleep disturbance. Finally, Professor Oliver will discuss how this extended understanding of behaviour difficulties can be integrated with the associative learning account and the clinical implications of the observed relationships between pain and behaviour.



tinyurl.com/46ys756f

For further information and resources for children with CHARGE syndrome, please visit:

- The CHARGE Syndrome Foundation: <https://www.chargesyndrome.org/>
- CHARGE Family Support Group <http://chargesyndrome.org.uk/>
- For the latest information about this study and related studies at Richard's Lab <https://www.chargeproject.org.uk>

For further information and resources for challenging behaviour in children, please visit:

- Cerebra's research summary guide and quick look fact sheet <https://w3.cerebra.org.uk/tag/self-injury/>
- The FIND (Further Inform Neurogenetic Disorders) website's section about challenging behaviour, which contains a video of Dr Caroline Richards talking about challenging behaviour in children. <http://www.findresources.co.uk/common-issues>
- The Challenging Behaviour Foundation's website which provides accessible information for parents and carers of children with intellectual disabilities who display challenging behaviours <https://www.challengingbehaviour.org.uk/>