



Behaviour and the Brain in Rubinstein-Taybi Syndrome

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Why study repetitive behaviour in RTS?

Over the last four years Chris Oliver's team at the Centre for Neurodevelopmental Disorders have been investigating the behavioural phenotype of Rubinstein-Taybi syndrome. The term 'behavioural phenotype' refers to behaviours that are more likely to be observed in a specific genetic syndrome relative to other genetic syndromes.

In 2008 members of the Cerebra team came to RTS weekend in Weymouth and asked parents whether they had any concerns that they would like the team to research. Many parents noted that their children engaged in repetitive-like behaviours. These included behaviours such as asking repetitive questions and adhering to strong routines.

Jane Waite (pka Appleby) was recruited to conduct research into these repetitive behaviours and to explore their possible causes.

Repetitive Behaviour and Executive Function

An initial search of the literature revealed that people with genetic syndromes and associated intellectual disability often engage in repetitive behaviour and that repetitive behaviour is not specific to RTS. However, the literature indicated the type of repetitive behaviours children engage in will often depend on the type of genetic syndrome they have. Despite this, there was nothing in the literature about how repetitive behaviour in people with RTS compared to people with other genetic syndromes.

The literature search also revealed that researchers have been speculating about the cause of repetitive behaviour in people with genetic syndromes. One of the most promising ideas is that repetitive behaviour might be linked to problems with a set of brain based processes; collectively these are known as the 'executive functions'.

Executive functions include:

Inhibition – the ability to suppress a learnt or impulsive response



Jenny completing the shape Stroop inhibition task.

Working memory – the ability to hold information in mind for a task at hand e.g. remembering a telephone number

Attention shifting – the ability to move attention away from one object, task or activity to another object, task or activity.

It is argued in the literature that repetitive behaviour might occur because a person cannot stop (inhibit) a behaviour once it has started, remember information and so repetitively ask for that information, or because a person becomes 'stuck' on one way of acting or behaving so cannot shift to another way of behaving.

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Special points of interest:

- Some behaviours in children with RTS were noted by parents to occur repetitively e.g. repetitive questions.
- Different syndrome groups are known to have different degrees of repetitive behaviour.
- Little is known about repetitive behaviour in RTS.
- There is some emerging evidence that repetitive behaviour may be linked to brain processes known as executive functions.

"He'll ask the same question over and over again. Sometimes I can distract him and move him on to something else, but five minutes later he'll start asking the same questions again."

Parent talking about her son.



But there is more to the executive functions....

Executive functions are of interest because of the potential links to repetitive behaviour, however, they are also of interest in their own right. People are not born with adult-like executive functions – the executive functions develop throughout childhood and adolescence. The development of executive functions have been linked to lots of abilities including achievement in school, the ability to think

about what other people are thinking, the ability to regulate emotions and control behaviour. Therefore, understanding how executive functions develop in people RTS may help to reveal lots about people with RTS more generally. One way to learn about the development of executive functions in people with RTS is to compare their performance on executive function tests to typically developing children.

In October 2009 the final research project was designed and aimed to:

- Describe repetitive behaviour in RTS and compare the behaviour to other groups to find out whether some behaviours occurred more often in RTS than in other syndrome groups.
- To map the development of inhibition, working memory and attention shifting (executive functions) in RTS and compare the development to typically developing children.
- To start to answer whether there are links between repetitive behaviour in RTS and the development of the executive functions.

Families who took part

In order to measure executive functions in people with RTS the Cerebra team adapted a number of tests of inhibition, working memory and task shifting that are usually used with young children. They then asked families and individuals with RTS whether they would be happy for the team to visit them at their homes to run the study. Many families came forward and offered their support of the research study and over 30 families were visited in total during 2009-2010.



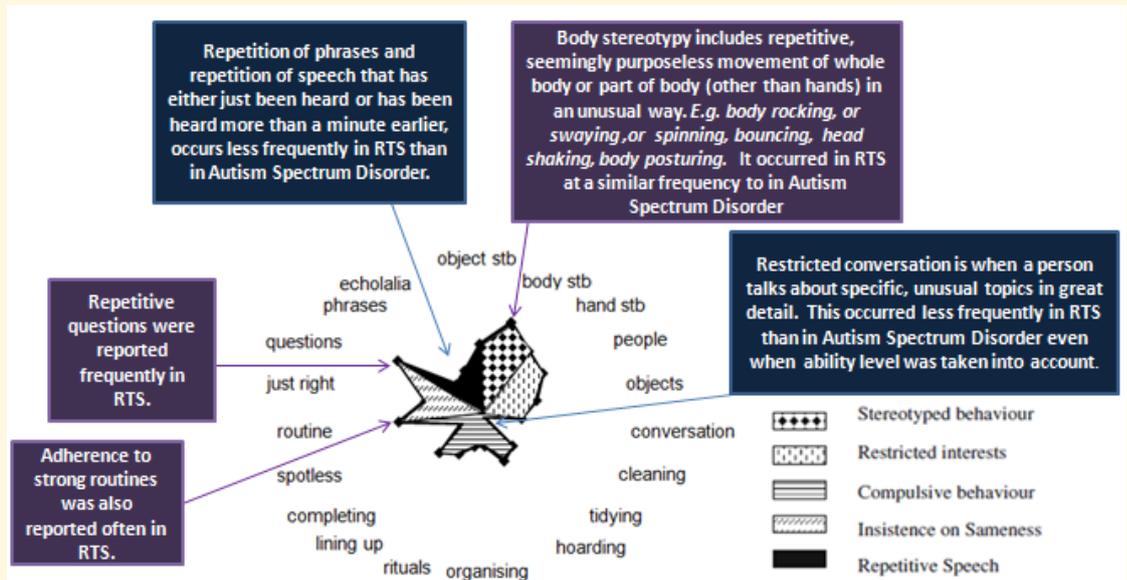
The Cerebra team would like to extend a huge thank you to all the families who took part in the project!

Left: Ben taking part in the buckets inhibition task.

Results: Repetitive Behaviour in RTS

The diagram on the right depicts the repetitive behaviour profile of RTS. This was compared to profile to Autism Spectrum Disorder (ASD) because repetitive behaviour is frequently reported in ASD.

The study confirmed that some behaviours occur frequently in RTS. These behaviours were repetitive questions, body stereotypy and adherence to strong routines. Other repetitive behaviours occurred less often.

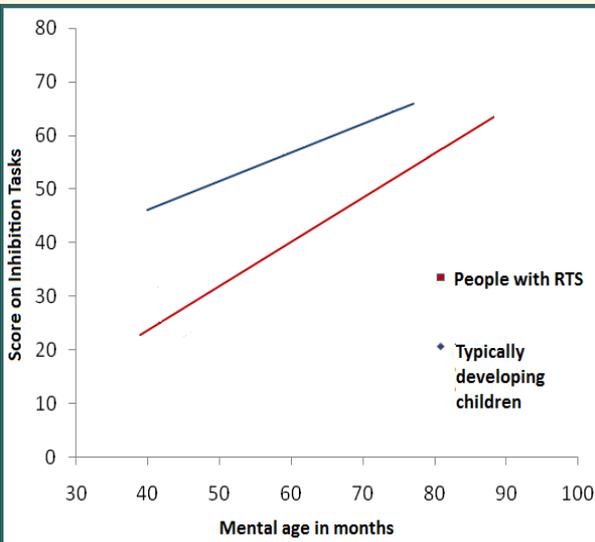


There are lots of types of repetitive behaviour ranging from attachment to people to compulsive-like behaviours

The Development of Inhibition



Left Nathan completing the Black-White Stroop task.



The participants with RTS completed an assessment to give the team an estimate of their mental age. This estimate allowed the team to compare the development of inhibition in people with RTS to the typically developing individuals.

It was found that the onset of the development of inhibition (the ability to suppress a response) occurred later in the people

with RTS relative to their mental age. This means that the development of inhibition was delayed not just in relation to the person's actual age but also in relation to their general level of skills and abilities. However, a positive finding was that those with higher mental ages performed better on inhibition tasks and that mental age appears to be related to actual age in RTS. Adolescents and adults with RTS were more likely to pass the inhibition tasks than the children. Thus, it may be that the onset of the development of inhibition is later in RTS rather than development not occurring at all.

The Development of Working Memory

The results also indicate that verbal working memory (e.g.. the ability to hold a list of words in mind for a short period of time) is similar to the inhibition. Verbal working memory is better in those with higher mental ages but the onset of development is later than in typically developing children.

On average individuals with RTS can hold between 2-3 words in working memory at one time. This may be

important to remember when presenting information to individuals with RTS. Giving information in small bite sized chunks and repeating information regularly is likely to



Left: Ben completing the Scrambled Boxes task.

help the person retain the information.

The results are also of interest because it was found that individuals with RTS found remembering sequences of visual information difficult particularly when the items to be remembered were similar. While aids such as visual timetables might be useful for people with RTS our results suggest that individuals with RTS would benefit from being given information verbally as well.

The Development of Attention Shifting

Attention shifting is the ability to shift from paying attention from one thing to another thing. If a person has problems with attention shifting they may keep doing the same thing over and over again because they cannot attend to new information that would help them to modify their behaviour.

A similar pattern of results was found for attention shifting as for inhibition.

However, the results suggest that more complex shifting (making lots of changes during a short period of time) might be particularly difficult for people with RTS than those without RTS with the same mental age. More research needs to be conducted to find out why lots of changes are difficult for people with RTS.



Above: The Dimensional Change Card Sort Task. Participants sort the cards by colour and later are asked to shift to sorting them by shape.

Links between Repetitive Behaviour and the Executive Functions

The final aim of the project was to begin to explore whether delays in the development of executive functions (inhibition, working memory, and shifting) were linked to the repetitive behaviours observed in RTS.

Several findings were of interest. Firstly, repetitive questions was strongly correlated with performance on a particular inhibition task (the Bear-Dragon task) and the verbal working memory task.

The Bear-Dragon task was also the most difficult inhibition task for individuals with RTS. We are beginning to explore what it is about this task that makes it so difficult and why it may be linked to repetitive questioning so strongly. Several families have taken part in the follow-up study that is addressing this.

No links were found between adherence to routines, body stereotypy and executive functions, however, a link

was found between completing behaviour (insisting that activities or objects are 'complete' or 'whole) and shifting. These links fit with previous reports of repetitive behaviour in the literature.



Above: The Bear and the Dragon from the Bear-Dragon task.

Future Research

This research has identified areas for further research. For example, there is now robust descriptions of repetitive behaviours in RTS and of how executive function development compares to typically developing individuals. Further research should be conducted to explore these findings in more detail as well as the factors that link particular repetitive behaviours to particular executive functions.

Strategies to manage and improve executive functions are being piloted

with individuals with genetic syndromes including training using computers.

This research project was also part of the broader Keystone Project that is addressing social behaviour in RTS. The study of social behaviour is still ongoing and parents will be updated as soon as the results are finalised.

Dr Jo Moss has also recently started an eye-tracking study with to learn about what attracts the attention of individuals with RTS.

Contact Details

If you would like any details of ongoing research at the Cerebra Centre for Neurodevelopmental Disorders or would like more details of anything reported in this article please contact:

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[Www.cndd.bham.ac.uk](http://www.cndd.bham.ac.uk)

Conclusion

This research project has highlighted that:

- Individuals with RTS often engage in repetitive questioning, adherence to routines and body stereotypy.
- Inhibition, verbal working memory and basic attentional shifting are linked to mental age in RTS but the onset of development may be delayed.
- There is some evidence that mental age is linked to actual age, which suggests that the executive functions may continue to develop over many years in individuals with RTS.
- Repetitive questions and completing behaviour are linked to executive functions.

Thank you!