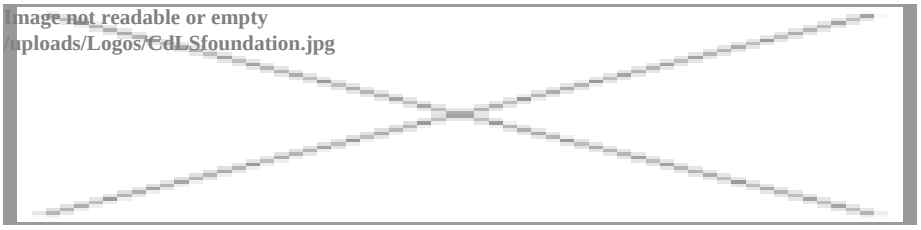
Get Involved - Research Projects

Decision making and executive function in Cornelia de Lange syndrome



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Background

The term ‘executive function’ refers to the skills we need to navigate the unpredictability of everyday life. These skills include memory, inhibition (being able to stop automatic behaviour) and switching attention. As these skills are very important for everyday functioning, and could impact on certain behaviours, it is important to investigate these in different genetic syndromes. People with particular syndromes have been found to have specific strengths and weaknesses in different executive functions, and this could impact on what interventions and strategies may be useful for different genetic syndromes.
One area that may be impacted by difficulties in executive function is decision making. Anecdotal reports from parents/carers of children with different genetic syndromes suggest that decision making skills may be significantly impaired in people with these syndromes. For example, parents report that scenarios in which an individual with Cornelia de Lange syndrome needs to make a decision can result in high levels of anxiety, aggression or self-injury. The aim of this project is to investigate executive functioning and decision making skills in children and adults with Cornelia de Lange (CdLS), Fragile X (FXS), and Rubinstein-Taybi syndromes (RTS); three rare genetic syndromes that are associated with specific behavioural profiles, which could indicate differing executive function strengths and weaknesses.

Aims

Across four studies, this project aimed to investigate the executive function profiles of people with Cornelia de Lange (CdLS), Fragile X (FXS), and Rubinstein-Taybi syndromes (RTS), using a variety of measures including questionnaires, table top tasks and parent/carer interviews.

Study 1

The aims of this study were to describe the executive function profile in CdLS, RTS and FXS and Autism Spectrum Disorder (ASD) using a questionnaire measure of executive function.

Study 2

The aims of this study were to further describe the executive function profile in CdLS, FXS and RTS using direct assessments of executive function, and to compare results between direct assessments and questionnaire measures of executive function.

Study 3

The aims of this study were to investigate the decision making skills and difficulties in CdLS ,RTS, FXS and typically developing mentally aged matched children) and how these relate to executive function and anxiety. This study used a novel decision making task developed specifically for people with intellectual disabilities.

Study 4

The aims of this study were to describe the everyday decision making difficulties and behaviours experienced by people with CdLS using semi-structured interviews with parents/carers. This will provide a richer understanding of what the decision making difficulties look like in people with CdLS.

Methods

Study 1

Parents/ carers of children with CdLS, RTS, FXS and ASD completed questionnaires to assess executive function and behaviours associated with ASD as part of a larger questionnaire pack.

Study 2

Participants with CdLS, RTS and FXS completed a battery of executive function assessments (developed by Waite, 2012) in the form of fun table-top tasks.

Study 3

Participants completed decision making task which involved them making decisions about eight everyday scenarios using picture cards. Video footage was taken of the decision making task and has been coded for behaviours indicating decision making difficulties and anxiety.

Study 4

Parents/carers of people with CdLS who took part in Study 3 were interviewed about their child's decision making difficulties, associated behaviour, and factors which make decision making easier or more difficult for their child.

Progress to date

Data collection for all four studies has now finished and results are currently being analysed. Preliminary results are reported below.

Study 1 and 2
Preliminary analysis indicated that people with CdLS, FXS and RTS have executive function difficulties in comparison to typically developing children who match them for verbal ability. Results also indicated that people with CdLS have particular difficulties with Verbal Memory and with Shifting Attention, whereas people with FXS and RTS have particular difficulties with Verbal Memory, Shifting Attention and also Inhibiting behaviour. These difficulties occur regardless of intellectual ability, which is important when considering educational strategies. Someone with CdLS, FXS or RTS who appears to be very able and articulate, may still struggle with certain tasks due to executive function difficulties. These results could be used to inform interventions in the future

Study 3

Preliminary analysis suggests that decision making may be difficult for a number of reasons; memory difficulties, switching attention difficulties and anxiety. People with CdLS showed particular anxiety when asked to do a task involving using memory, whereas people with FXS showed higher anxiety when asked to do a task involving being able to switch attention. It may be useful to have visual aids when asking someone with CdLS and FXS to make a decision, to aid the memory demand, and to reduce the number of options presented to two to reduce switching attention demand.

Study 4

Data analysis is still ongoing.

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