Summary of research findings: Executive functioning in Cornelia de Lange syndrome: domain asynchrony and age-related performance.

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Background: In this study we aimed to look at executive function skills in adolescents and adults with CdLS. Executive function skills are abilities that help us to process and manage information in the world around us. These abilities include memory, regulating our responses and our ability to act flexibly in novel or complex situations.

Methods: We evaluated executive function skills in 24 people with CdLS aged 13 to 42 years and in 20 people with Down syndrome aged 15 to 33 years. Comparing skills across different syndrome groups is really helpful because it allows us to better understand the results. We also explored the relationship between these abilities and chronological age in these participants.

Results: We found that people with CdLS in our study had more difficulties than people with Down syndrome on tasks which assessed the ability to think flexibly and in working memory skills. We also found that older individuals with CdLS in our study performed more poorly on one particular assessment of working memory than younger individuals with CdLS in our sample.

Interpretation of findings: Our research shows that individuals with CdLS might experience specific difficulties in working memory and flexible thinking to a greater extent than individuals with Down syndrome. The impact of these difficulties in day to day life might be that people with CdLS find novel situations or changes in routine difficult to navigate. They may also find it hard to remember what events are coming up. These difficulties might help us to understand some of the anxiety that has been described in individuals with CdLS.

We also found that difficulties in one particular aspect of working memory was more prominent in the older individuals in our CdLS sample. One interpretation of this finding is that abilities in this specific area of working memory might change with age in some individuals with CdLS. At this stage, we do not know the nature, the extent or the cause of this possible change. Other changes in specific aspects of behaviour with age have been described in other research studies conducted by our group and by others. Some researchers have hypothesised that this might be related to biological differences that occur as a consequence of genetic mutations in the cohesin pathway. Other explanations include environmental factors (increased demands with age), methodological factors (cohort effects whereby older people with CdLS are more likely to be more severely affected than younger individuals due to recent advances in diagnostic techniques) or individual differences. Studies that follow the same group of people with CdLS over time is needed to help us to document and understand these potential areas of change in greater detail.

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