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Research Summary

Autism in genetic syndromes: Implications for assessment and intervention

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Autism in genetic syndromes: Implications for assessment and intervention

This research summary has been prepared to help parents and carers of children with genetic syndromes understand how and why autism or related characteristics might occur in children with genetic syndromes and what this might mean for assessment and intervention related to these characteristics in this population. This research summary provides a review of information that has been presented in peer reviewed scientific articles and reviews. [1,2,3] This research summary was updated in March 2019 following a review of relevant literature published since the research summary was originally printed in 2013.

1. What is Autism Spectrum Disorder?

Autism occurs in up to 1% of children and adults in the general population[4] and up to 40% of individuals within the learning disability population.[5] Autism is diagnosed on the basis of characteristic behaviours and difficulties in two core areas. These are:

- persistent differences in social communication and social interaction across multiple contexts and
- the presence of repetitive behaviours and restricted interests and activities.[6]

In autistic children, difficulties in these areas are typically evident before the age of three years. Autism is a spectrum condition, which means that while autistic people share certain characteristics, there is a great deal of variability in the way in which they manifest and in how significantly they impact on the individual's everyday life.

2. What is the cause of Autism Spectrum Disorder?

The specific causes of autism remain unknown but most studies indicate a biological or genetic cause.[7] A high level of heritability (passing within families) of autism characteristics in families and siblings of affected individuals has been reported.[8] However, this is not always the case and there are many autistic individuals for whom there are no familial links to the condition.[9] While several chromosomes and genetic locations have been linked to autism, there is very little evidence to suggest that any one of these is solely responsible for it's occurance. In fact, it has been suggested that each of the multiple genes associated with autism, with the exception of fragile X syndrome, accounts for less than 2% of autism cases.[10] This has led researchers to suggest that autism results from complex interactions between several genes (rather than a single genetic mutation) which lead to differences on a number of biological levels.[7, 10, 11, 12]

3. What medical conditions are associated with autism?

Research has shown that some medical conditions are more likely to occur in autistic individuals than in the general population including epilepsy, sleep disorders, gastrointestinal disorders and immune functioning problems. These medical conditions can impact on children's broader development, social functioning, and education outcomes.

Approximately 10% of autistic individuals have an accompanying medical problem that requires medical evaluation.[13] Sleep problems affect between 50% to 80% of autistic children. Poor sleep affects not only the daily functioning of the individual but also the quality of life of the whole family. Some of these problems may include difficulties falling asleep, not sleeping long enough, night wakefulness or low quality of sleep.[13]

4. Occurrence of autism in genetic syndromes associated with learning disability.

The term 'genetic syndrome' is used to refer to a condition in which there is a known genetic cause that results in a cluster of physical and behavioural symptoms. Approximately 50% of individuals within the learning disability population have a genetic syndrome and this figure is around 20% for individuals who have a mild learning disability.[14] A well-known and recognised example of a genetic syndrome associated with a learning disability is Down syndrome. Down syndrome is typically caused by the presence of an additional copy of chromosome 21 (trisomy 21). In a small proportion of individuals an unbalanced translocation involving chromosome 21 (meaning that material from one chromosome 21 gets stuck or translocated to another chromosome) has been identified. Individuals with Down syndrome have distinctive facial and physical features (physical phenotype), cognitive difficulties (cognitive phenotype) and behavioural characteristics (behavioural phenotype).[15]

Research has consistently shown that individuals with genetic syndromes associated with a learning disability are at increased risk for autism relative to the wider learning disability and general populations.[16] The strength of association or co-occurrence between a given genetic syndrome and autism is variable, with prevalence estimates ranging from 5% (less than one person out of ten people) in individuals with Down syndrome to 90% (nine out of ten people) in individuals with Tuberous Sclerosis Complex.

Just as the percentage of children with a genetic syndrome who have autism varies, so the nature and severity of autism characteristics also differ across syndrome groups. Some studies describe individuals with a given syndrome as showing "autism-like characteristics" or "autism traits", which suggest there may be some similarities with autistic individuals but that the challenges, skills and behaviours observed are not entirely the same as those identified in autistic individuals who do not have a genetic syndrome. Other studies describe a much stronger association between syndromes and autism, and this may lead to individuals receiving a dual diagnosis of the genetic syndrome *and* autism. In either case, recognising that an individual may share some or all of the challenges, skills and behaviours that are characteristic of autism may help carers and professionals to understand the children's needs. Unfortunately, these subtle differences lead to challenges in assessing and diagnosing autism in individuals with genetic syndromes.[17] Studies report significantly reduced or delayed diagnosis of autism in the genetic syndrome population[18] and consequently poorer outcomes relative to autistic individuals who do not have a genetic syndrome.[17]

5. Why do individuals with some genetic syndromes show an association with autism?

This is not well understood. Some researchers suggest that the genes underlying those syndromes in which autism characteristics are very common, lead to shared differences at the biological , cognitive and neurological level, which in turn give rise to autism characteristics.[7] Such researchers believe that understanding these pathways in genetic syndromes is helpful in determining the causes of autism more widely.[12] However, this is not a view held by all researchers. Others suggest that it is not the underlying genetic causes which give rise to autism in certain genetic syndromes but an effect of the degree of learning disability associated with the syndrome, which increases the risk that autism characteristics will co-occur.[19]

6. Genetic syndromes commonly associated with autism characteristics

Three genetic syndromes that have commonly been reported to be associated with autism include fragile X syndrome, Cornelia de Lange syndrome and Tuberous Sclerosis Complex.

6.1. Fragile X syndrome:

Fragile X syndrome (FXS) is the most common cause of inherited learning disability, occurring in 1 in 3,600 males and 1 in 8,000 females.[20] FXS is caused by the presence of an apparently unstable or 'fragile' site located on the X chromosome. The instability is caused by an excess of genetic code in this region.[21] Males with FXS typically show mild to severe learning disability while females with FXS usually have a mild learning disability.[22]

Recent studies of individuals with FXS show a fairly consistent pattern of association with autism. The percentage of individuals with FXS showing autism characteristics or meeting criteria for a clinical diagnosis ranges from 21% to 50%. In females, this figure is much lower; between 1 and 3%.[1, 2, 3] Severe autism is relatively rare in FXS and a milder presentation of autism characteristics is more commonly observed.[23,24] Further, premutation carriers of FXS also have increased susceptibility for developing characteristics of autism.[25]

On closer inspection, some of the autism related difficulties observed in individuals with FXS appear to be different to those identified in autistic individuals who do not have a genetic syndrome. Impairments in social interaction in FXS are characterised by social anxiety, extreme shyness and eye gaze avoidance.[26] These characteristics are observed in autistic individuals but are not considered to be core features. Furthermore, individuals with FXS are reported to have a strong willingness to engage socially with others.[27, 28, 29, 30] This is somewhat different to the social indifference often described in autistic individuals. Greater emotional sensitivity and less idiosyncratic speech are also reported in individuals with FXS relative to autistic individuals who do not have a genetic syndrome.[17] In further contrast to autistic individuals, the social impairments in FXS are thought to become more prominent with age.[29, 31]

Evidence suggests that individuals with FXS who meet criteria for autism show increased eye gaze avoidance and reduced gaze to the parent when compared with FXS-only individuals. This difference between autistic and non-autistic individuals with FXS shows that the presence of autism may predispose this subset of

children with FXS to respond differently to social information.[32] Individuals with FXS and autism are more severely impaired in communication and social interaction and demonstrate greater problems in cognitive and receptive language skills compared to individuals with FXS-only.[33] Similarly, social skills in FXS seem to be related to the severity of autism characteristics. Individuals with mild autism characteristics have better social skills which improve with age, in contrast, individuals with more autism characteristics have a very low level of social skill and there is no improvement with age.[34] As might be expected, individuals with FXS and autism have lower levels of adaptive functioning than those without autism characteristics. Specifically, they show lower abilities in using time and schedules, money, maths and writing skills, personal hygiene and abilities to perform common household tasks. Autism in FXS is also related to lower levels of independent living at adult age.[35]

Repetitive behaviours and restricted interests are part of the diagnostic criteria for autism but are also highly characteristic of FXS more broadly. Individuals with FXS and comorbid autism are reported to show even higher levels of these behaviours when compared to those with FXS who do not have autism.[21,36,33] Autism in FXS is also likely to be accompanied by other problems, such as ADHD and anxiety.[22] Persistence of sleep problems and behavioural problems are also reported in individuals with FXS and autism.[33] In general, autism increases the severity of FXS and individuals with both conditions demonstrate more serious problems.[36]

6.2. Cornelia de Lange syndrome

Cornelia de Lange syndrome (CdLS) is a multisystemic congenital syndrome that affects approximately one child in every 10,000 – 30,000 37 CdLS is primarily caused by a deletion (and in some cases mosaicism) on chromosome 5. Other deletions on chromosomes 8, 10 and X have also been identified.[38, 39, 40, 41, 42, 43] Other, as yet unidentified, chromosomal abnormalities may also contribute to the cause of CdLS. The degree of learning disability in CdLS ranges from mild to profound [37, 44]. It is a malformation disorder with a distinctive physical appearance, small stature, medical complications and variable developmental and behavioural presentation.[45, 46]

Studies suggest that autism characteristics occur in 27% to 82% of individuals with CdLS.[47] Scores on assessments of the severity of these characteristics are significantly higher in CdLS compared to other individuals with similar levels of learning disability who do not have CdLS.[48, 49, 50] The reported occurrence rates of autism characteristics in CdLS are similar to those identified in fragile X syndrome. 1 Higher levels of adaptive functioning are associated with less severe autistic characteristics. [45]

Similar to fragile X syndrome, some of the autism characteristics observed in CdLS appear to be different to those identified in autistic individuals who do not have a genetic syndrome. Social anxiety and extreme shyness are common and unusually, selective mutism (speaking in one environment but not another or speaking only to certain individuals), is particularly prominent.[51] Social avoidance and difficulties in socialisation are also common.[2, 45, 52] Other studies have demonstrated that the nature of repetitive behaviours appears to be different in individuals with CdLS compared to autistic individuals. In particular, individuals with CdLS show fewer sensory related behaviours such as licking, sniffing and finger flicking.[1] Some research shows that older individuals with CdLS have increased prevalence and severity of social difficulties related to autism than younger individuals with the syndrome.[46]

6.3. Tuberous Sclerosis Complex

Tuberous Sclerosis Complex (TSC) is a multisystem disorder, with an incidence rate of 1:6000 newborns; it is caused by a mutation on either the TSC1 or TSC2 gene on chromosome 9 or 16.[53,54] These genetic mutations results in the

development of benign tumours in the brain, skin and internal organs.[55] The effect of these tumours are extremely varied with some individuals having only superficial skin problems while others show severe physical effects and profound learning disabilities. Typically, TSC2 mutations appear to be associated with a greater severity of clinical characteristics than TSC1 mutations.[56]

Significant impairments in social interaction, stereotyped behaviour, absent or abnormal speech and social withdrawal were reported in early descriptions of TSC. Recently studies have reported that between 17% and 61% of individuals with TSC show autism like characteristics. [1,2,57]

One study has suggested that repetitive behaviours are not as frequent in individuals with TSC when compared to individuals with autism, while the severity of social interaction and communication impairments in these groups is very similar.[58]

These difficulties have been reported to occur both in those with mild and those with severe effects of TSC.[58]

Abnormal cognitive and behavioural functions in TSC are associated with the presence of a learning disability and the presence of autism characteristics. This indicates that these areas of behaviour and functioning may share common underlying causal pathways.[58] In fact, both individuals with idiopathic autism and TSC exhibit similar neural connectivity deficits.[59]

7. Other genetic syndromes associated with autism characteristics

Above we have described three genetic syndromes that are most commonly associated with autism characteristics. In this section, we describe a number of other genetic syndromes in which autism characteristics have been reported, including Angelman syndrome, Down syndrome, Rett syndrome, Prader-Willi syndrome, Williams syndrome and Phelan McDermid syndrome.

7.1. Angelman syndrome

Angelman syndrome (AS) occurs in approximately 1 in 12,000 to 15,000 individuals[60, 61] and is caused by abnormalities on chromosome 15.[16] Individuals with AS typically show severe to profound learning disability,[62] significant difficulties with mobility and communication and seizures.[15]

Studies report that 50% to 81% of individuals with AS meet criteria for autism. However, the characteristic features of AS are not consistent with these findings. Individuals with AS show strong motivation to engage socially with others, showing very high levels of laughing and smiling during situations in which social contact is available.[63, 64] While communication skills are impaired and language significantly delayed, social approaches are very frequent. When compared to autistic individuals, individuals with AS are less impaired in some of the core characteristics of autism such as social smile, shared enjoyment, facial expression, unusual interests or repetitive behaviour and response to name.[65] Some behaviours that are typically associated with autism, such as hand mannerisms, are present in the majority of individuals with AS. It is possible that these behaviours are characteristic of the syndrome per se rather than an indicator of autism in this group.[16] While it is likely that some individuals with AS do genuinely show high levels of autism characteristics, it is possible that the profound level of learning disability associated with the syndrome and the unusual social interaction skills in this group may result in overinflated scores on diagnostic measures of autism. Such factors would need to be carefully considered when assessing an individual with AS on such assessments.[16]

7.2. Down syndrome

Down syndrome (DS) is the most common chromosomal cause of learning disability and occurs in 11.8 in 10,000 live births.[66] As described in Section 4 on page 5, DS is typically caused by an extra copy of chromosome 21. Individuals with DS have a mild to severe level of learning disability.[67] DS accounts for approximately 5–6% of learning disability cases and exists as one of the most common genetic causes of learning disability.[18]

Typically, individuals with DS are skilled in social contact and are motivated to engage socially with others. Communication skills are delayed but many individuals develop good language skills. These characteristics have previously led researchers and clinicians to believe that autism characteristics are relatively rare in DS. However, recent studies suggest that around 5% to 39% of individuals with DS meet diagnostic criteria for autism.[1, 2, 18] These figures are lower than those reported in the other syndrome groups described in this research summary but remain higher than that of the general population (around 1%; see Section 1 on page 4). It is possible that genetic factors contribute to the risk of autism in DS and most likely the combined effect of several genes underlies that increased susceptibility.[68]

Recent studies have suggested that individuals with DS who meet diagnostic criteria for autism have lower levels of ability and higher rates of repetitive behaviour, hyperactivity and impaired

speech compared to those with DS who do not show these characteristics.[67 69, 70] Individuals with comorbid DS and autism also demonstrate increased levels of self-injurious behaviours, unusual sensory interests (such as a fascination with a whirling fan or flickering lights), lethargy, behavioural disturbances and self-absorbed behaviours when compared to individuals with DS only. Further, these behaviours are different from the behaviours seen in individuals with DS only. Individuals with DS and comorbid autism and autistic individuals show fewer positive vocalisations than those with DS only.[18]

While similarities between people with DS and autism and autistic individuals are evident, individuals with DS and autism are also thought to have relatively milder social difficulties than autistic individuals who do not have a genetic syndrome. For example, they have fewer problems in reciprocal social interaction, emotional and peer-related problems, imitative social play, eye gaze, facial expression, social smiling and quality of social interaction. They are more likely to comfort others and respond positively when approached by others.[70]

7.3. Rett syndrome

Rett syndrome (RS) is a neurological disorder that is caused by a mutation on the X chromosome. RS predominantly affects females and occurs in 1 in 15,000 to 22,800 live female births.[71] Typically, development appears to be normal in the first six to eighteen months but this is followed by a period of regression resulting in a loss of language and motor skills, leading to severe or profound learning and physical disabilities.[72] A small number of individuals with RS have been reported to retain and develop their language skills.[73, 74]

Autism characteristics were noted in the very first description of RS in 1966. Studies have since estimated that 25% to 40% of individuals with RS show autism characteristics.[1, 2] Autism is the most common misdiagnosis in children with RS, with 18% of individuals being diagnosed with autism prior to receiving a diagnosis of RS.[75] As with FXS, while autism characteristics are very common in the syndrome, there are some distinct differences in these features. For example, despite severe impairments in social interaction skills, eye contact is reported to be good.[72] Additionally, the repetitive behaviour that is most characteristic of individuals with RS is a stereotyped hand wringing movements. This is very different to the stereotyped behaviour typically observed in idiopathic autistic individuals who do not have a genetic syndrome. Other studies have confirmed that individuals with RS are less likely to show the core features of autism.[76]

7.4. Prader–Willi syndrome

Prader-Willi Syndrome (PWS) is a result of loss of function of genes on chromosome 15.[16] The prevalence of PWS ranges from 1/25,000 to 1/10,000 children.[77, 78] The intellectual functioning in PWS varies from average abilities to a moderate learning disability with the majority of individuals having a mild learning disability.[16, 77] PWS is considered a cause of early-onset childhood obesity, as individuals with this disorder experience increased appetite, excessive hunger and food seeking behaviours. It is also associated with characteristic facial features including almond eyes, narrow forehead, thin upper lip and small hands and feet.[77]

Autism characteristics are prevalent in PWS, with about 12%-27% of individuals receiving a diagnosis of autism.[79,80] Those with comorbid autism show lower verbal, socialisation and adaptive daily living skills along with elevated frequency of repetitive behaviour and restricted interests.[80]

7.5. Williams syndrome

Williams syndrome (WS) is a rare neurodevelopmental disorder caused by the deletion of several genes on chromosome 7 and occurring in 1 of every 7,500-20,000 live births. WS is characterised by mild to moderate learning disability, profound visuospatial impairments and motor difficulties.[81] Individuals with WS

have relative strengths in language and social interactions, showing heightened empathy and openness and an increased desire to communicate.[81] Additionally, people with WS have strengths in their responses to social stimuli, for example in imitating adults. Some researchers believe that the increased social approach in WS is caused by the perception of faces as highly rewarding and salient.[81]

Although referred to by some authors as the 'polar opposite to autism', autism is diagnosed in 7%- 12% of individuals with WS, which is notably higher than in the neurotypical population.[82] Like DS, it was previously considered that individuals with WS have lower susceptibility to autism due to their characteristically marked differences in sociability.[81] Even though the social behaviours in WS and autism are remarkably different it is possible that they share some similar difficulties in the way in which they process and interpret social information. For example, difficulties in following and understanding eye gaze, reduced rates of some social responses such as giving and showing objects to others and difficulties in forming and maintaining friendships are reported in both autistic individuals and individuals with WS.[81, 83]

7.6. Phelan McDermid syndrome

Phelan McDermid syndrome (PMS) is a rare genetic condition with an estimated almost 2,000 people diagnosed worldwide.[84] PMS is typically caused by a deletion on chromosome 22 which almost always includes the SHANK3 gene.[85] Individuals with PMS exhibit developmental delay, language difficulties, motor delay and increased tolerance to pain.[85] Additionally, autism has a high occurrence in PMS with prevalence rates of 39% - 90% [86, 87, 88]. Research suggests that the SHANK3 gene, located on chromosome 22, is related to the autism characteristics observed in PMS.[89] SHANK3 mutations are found in approximately 5% of individuals with autism.[86] However, there is no clear relationship between the deletion size and the presence of autism characteristics.[87]

Both individuals with PMS and autistic individuals are reported to share similar impairments in communication and social interaction. However, rates of stereotyped behaviour, compulsive behaviours and insistence on sameness have been found to be lower in autistic individuals with PMS relative to those with autism who do not have a genetic syndrome.[87,88] It is proposed that language and cognitive impairment contribute to the autism characteristics seen in PMS and that the neural networks leading to these problems are different to those observed in autistic individuals.[87]

Similarly, to other genetic syndromes, diagnosis of autism in PMS is challenging because many of the diagnostic tools are not adapted for individuals with severe learning disability and with conditions with comorbid sensory or motor impairments.[86]

8. Assessing autism in genetic syndromes

In the sections above, we have described the nature of association between a number of genetic syndromes and autism characteristics. In most cases, these descriptions are not entirely clear cut and while reported levels of association are often high, subtle differences in the characteristics have been reported. These differences can often make assessment of autism characteristics in genetic syndromes by professionals and clinicians more difficult. Many of the assessment tools that are available to evaluate autism characteristics are designed to assess and diagnose 'typical' autism in individuals without genetic syndromes and may not always be able to detect and describe the subtle differences reported across different syndrome groups. The assessment tools that are considered the best to use are the Autism Diagnostic Observation Schedule (ADOS90) in combination with the Autism Diagnostic Interview (ADI91). Sometimes a 'screening' questionnaire like the Social Communication Questionnaire (SCQ92)

is used. However, even with these tools, careful and detailed assessment is required alongside a good knowledge of the behavioural phenotype associated with the genetic syndrome in order to diagnose autism accurately in individuals with genetic syndromes.

Another factor which makes assessment of autism in genetic syndromes difficult is the severity of learning disability associated with the syndromes. This is particularly problematic when the level of learning disability is in the severe to profound range. Autism is identified by the absence, delay or impairment of particular skills in communication and social interaction. However, there may be other reasons why an individual shows impairments, delay or absence of skill in these areas. One reason might be the presence of autism but another might be the presence of a learning disability. In individuals with a severe to profound level of learning disability, it is very difficult to identify which of these two reasons explains the absence or impairment of these skills, since the individual may not have yet reached the developmental level required to achieve that particular skill and, of course, both reasons may be valid and present. Additionally, individuals with this level of disability are also very likely to show repetitive behaviours, specifically stereotyped behaviours, play and movements and these behavioural similarities cause further confusion.

While these factors make it difficult to evaluate autism characteristics in individuals with genetic syndromes, assessment may still be valuable and should be considered if appropriate. However, professionals and clinicians may be cautious in diagnosing autism in an individual with a genetic syndrome, particularly when there may be some differences in the nature of these difficulties. Even in cases where a diagnosis of autism is not appropriate, recognition that the individual shows some autism characteristics may be helpful in understanding the needs of the individual.

9. Practical implications

The idea that there may be an association between particular genetic syndromes and autism characteristics can be very confusing and may result in families feeling overwhelmed at the thought that not only does their child have a genetic syndrome but they also have another disability. However, it is important to be aware that the presence of autism characteristics in an individual with a genetic syndrome does not necessarily mean that the person has two different disorders. Rather, it is perhaps more appropriate and helpful to consider that these autism related difficulties are characteristic of the syndrome itself (i.e. part of the behavioural phenotype) and the individual. Recognition of these shared difficulties is important for ensuring that the person receives appropriate intervention and educational management and to enable parents, carers and professionals working with the individual to seek appropriate advice and resources.

Individual clinical case studies of children and adults with genetic syndromes suggest that recognising these characteristics is important for ensuring that the appropriate behavioural management and educational programmes can be put in place. However, there have been no research studies which have evaluated how useful interventions that have been developed to target autism characteristics and designed for autistic individuals who do not have a genetic syndrome, might be for individuals with genetic syndromes who show autism characteristics. What is also unclear is whether such interventions would be appropriate for use in individuals who show some but not all of the diagnostic characteristics of autism. In such cases, it is possible that a combination of autism specific interventions and targeted intervention for particular difficulties, such as selective mutism in CdLS or gaze avoidance in FXS, might be appropriate. However, further research is required in order to gain a better understanding of how effective these interventions might be.

Summary and key points

- Autism is a lifelong developmental condition that affects an individual's communication and social interaction skills. The nature of autism is very variable and characteristics may vary across individuals.
- Autism and autism characteristics are commonly observed in individuals with genetic syndromes associated with learning disability. Specific examples of genetic syndromes in which a strong association with autism has been described include fragile X syndrome, Cornelia de Lange syndrome and Tuberous Sclerosis Complex.
- In most cases, when autism characteristics have been described to be common in a syndrome, there are some subtle differences in the nature of these characteristics compared to individuals with autism who do not have the syndrome.
- Recognising any shared characteristics of autism and the areas where there may be differences to autistic individuals who do not have a genetic syndrome is important for understanding the individual's educational and day-to-day needs and for enabling parents, carers and professionals to access appropriate resources and advice.
- Assessing autism characteristics in individuals with genetic syndromes can be complex but is by no means impossible and a clinical evaluation should be conducted in situations where this might be appropriate
- The effectiveness of autism specific interventions for use in individuals with genetic syndromes who show autism characteristics is not known. It is likely that a combination of autism specific interventions and targeted interventions which focus on specific areas of difficulty will be appropriate. However, further research is required to evaluate such approaches.

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Authors

Dr Jo Moss

Dr Jo Moss is a Senior Lecturer in Psychology at the University of Surrey and Co-Director of the Cerebra Network for Neurodevelopmental Disorders (www.cerebranetwork.com). Through her research, Jo aims to better understand social, emotional and cognitive diversity in individuals with a range of neurodevelopmental conditions. Jo's research has a particular focus on individuals with genetic syndromes associated with intellectual disability and combines detailed behavioural description with in-depth profiling of social-cognitive abilities. Jo uses novel research methods, supporting an inclusive approach to research in populations that are typically underrepresented within the field.

Galina Ignatova

Galina is a final year Psychology student at Aston University who completed a placement year at Cerebra Centre, University of Birmingham in 2018-2019. While on placement she worked on several projects including the i-RISK project, aiming to develop a tool to help professionals and parents recognise possible future self-injuring behaviours in children with neurodevelopmental disabilities. She also provided recruitment and testing support on 3 Fragile X related projects and co-authored a systematic literature review on the behavioural phenotype of Phelan-McDermid syndrome. Additionally, she is a one-to-one tutor, providing students of various ages with support in their English and Mathematics studies.

In her spare time, Galina enjoys travelling to different places, hiking and kickboxing. One of the most exciting adventures she had so far is the climbing of peak Uhuru (5895m) in mount Kilimanjaro, Tanzania.

Professor Chris Oliver

Chris Oliver is Professor of Neurodevelopmental Disorders at the University of Birmingham and Director of the Cerebra Centre for Neurodevelopmental Disorders. He trained as a clinical psychologist at Edinburgh University before completing a PhD on self-injurious behaviour in people with intellectual disability at the Institute of Psychiatry, London. He is currently researching early intervention, behaviour disorders in people with severe intellectual disability and autism spectrum disorder, behavioural, cognitive and emotional phenotypes in genetic syndromes and neuropsychological and behavioural assessment for people with severe intellectual disability. He has published over 180 peer-reviewed articles in scientific journals, was previously Editor-in-Chief for the Journal of Intellectual Disability Research and serves on a number of scientific advisory committees for autism and syndrome support groups. Summaries of research are available at www.findresources.co.uk.

Cerebra Network

This programme will take forward previous research completed by the Cerebra Centre for Neurodevelopmental Disorders, led by Professor Chris Oliver at the University of Birmingham. In the next phase of this research programme, the Cerebra Network for Neurodevelopmental Disorders will be making an exciting transition by expanding to comprise a collaborative and dynamic network of researchers. The Cerebra Network for Neurodevelopmental Disorders will be led by four alumni of the Cerebra Centre; Dr Caroline Richards (University of Birmingham), Dr Jo Moss (University of Surrey), Dr Jane Waite (University of Aston) and Dr Hayley Crawford (University of Warwick). Network research hubs located at each university will focus on key themes that are central to improving the lives of individuals with severe and complex needs and their families including research into sleep, atypical autism and mental health, while continuing their work on self-injurious behaviour and pain in this population.

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Postal Address

Cerebra
The MacGregor Office Suite
Jolly Tar Lane
Carmarthen
SA31 3LW

Tel: 01267 244200
Freephone: 0800 328 1159

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