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Prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes.

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Please use this reference when citing this work:

Arron, K., Oliver, C., Berg, K., Moss, J. and Burbidge, C. (2011). Prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes. *Journal of Intellectual Disability Research*, **55**, 109-120. DOI: 10.1111/j.1365-2788.2010.01337.x

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Abstract

Background. Self-injurious and aggressive behaviours are reported as components of some behavioural phenotypes but there are few studies comparing across syndrome groups. In this study we examined the prevalence of these behaviours and the associated person characteristics in seven genetic syndromes.

Methods. Questionnaire data on self-injury and aggression, mood, hyperactivity, autism spectrum disorder and repetitive behaviour were collected on Angelman (AS, n=104), Cornelia de Lange (CdLS, 101), Cri du Chat (CdCS, 58), Fragile X (FXS, 191), Lowe (LS, 56), Prader-Willi (PWS, 189) and Smith-Magenis (SMS, 42) syndromes.

Results. A significantly higher prevalence of self-injury was evident in CdCS, CdLS, FXS, PWS, LS and SMS. The prevalence of aggression was significantly heightened in AS and SMS. Self-injury was associated with repetitive and impulsive behaviour in CdLS, FXS, PWS and LS. Impulsivity and overactivity were significantly higher in those showing aggression across all syndrome groups.

Conclusions. These data quantify the risk for self-injury and aggression in the syndromes studied with implications for early intervention. The associations between these behaviours and person characteristics both within and between syndromes warrants further research.

Keywords: behavioural phenotype, ASD, repetitive behaviour, hyperactivity, impulsivity, affect, self-injurious behaviour, aggressive behaviour, genetic syndromes.

The prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes.

Prevalence estimates for self-injury in people with intellectual disability vary from 4% to 10% (Oliver, Murphy & Corbett, 1987, Cooper, Smiley, Allen et al., 2009a) and 7% to 25% for aggression (Emerson *et al.*, 2001; Cooper, Smiley, Allen et al., 2009b). These behaviours impinge significantly on the quality of life of people with intellectual disabilities and their families and can lead to exclusion and the need for costly services (Hassiotis, Parkes, Jones, Fitzgerald & Romeo; 2008; Hastings, 2002). There is broad consensus that the effectiveness of interventions for these behaviours is closely aligned with determining their cause given the multitude of potential contributory factors.

There is evidence from cohort studies that self-injurious behaviour (SIB) in people with intellectual disabilities is associated with a number of individual characteristics such as greater degree of intellectual disability, the diagnosis of autism spectrum disorder and the presence of stereotyped, compulsive and impulsive behaviours (Bodfish, Crawford, Powell, Parker, Golden & Lewis, 1995; McClintock, Oliver & Hall, 2003; Powell, Bodfish, Parker, Crawford, & Lewis, 1996; Rojahn, Matson, Naglieri, & Mayville, 2004, Cooper et al., 2009). Similarly, aggression is associated with ADHD, being male, compromised communication, and Autism Spectrum Disorder (McClintock et al., 2003; Cooper, Smiley Jackson et al., 2009). These associations allude to the importance of person characteristics in the aetiology of these behaviours. Similarly a robust literature on aetiology provides extensive evidence that the behaviours can be learned via social reinforcement (Hanley, Iwata & McCord, 2003; Oliver, 1995). This literature indicates the importance of environmental influence. These parallel literatures are, arguably, in conflict as each places different emphasis on the role played by potential biological and environmental determinants. One strategy for exploring this difference in perspective is to examine SIB and aggression in genetic syndromes in which the prevalence of the behaviour is suggested to be elevated.

Operant learning theory alone would predict no differences in prevalence or phenomenology across syndromes because environmental influences would, presumably, be randomly distributed across groups. Any difference in the prevalence of self-injury and aggression across syndrome groups would undermine the argument that these behaviours can be accounted for by operant learning theory alone. In studies with varying degrees of control, self-injury is more commonly reported in Lowe, Smith-Magenis and Prader-Willi syndromes (Clarke & Boer, 1998; Clarke, Boer Chung, Sturmey & Webb, 1996; Dykens & Smith, 1998; Einfeld, Smith, Durvasula, Florio & Tongue, 1999; Holland, Whittington, Webb, Boer & Clarke, 2003; Kenworthy & Charnas, 1995) with equivocal results for Angelman, Cornelia de Lange, Cri du Chat and Fragile X syndromes (Oliver, Sloneem. Hall and Arron, 2009; Collins & Cornish, 2002; Lensiak-Karpiak, Mazzocco & Ross, 2003). However, evaluation of the validity of these differences is currently problematic as studies employ different methods of sample ascertainment and assessment.

It is also important to examine person characteristics of those showing self-injurious and aggressive behaviours within and between syndromes. This might identify the same characteristics that are associated with SIB in all people with intellectual disability but that are more prevalent in syndromes in which the prevalence of SIB is also high (e.g. compulsive behaviours in Cornelia de Lange and Prader-Willi syndromes, Clarke, Boer, Whittington, Holland, Butler & Webb, 2002; Hyman, Oliver & Hall, 2002). There is emerging evidence that overactivity, impulsivity and the presence of repetitive behaviours, for example, might be important person characteristics that are associated with self-injury (Bodfish et al., 1995; Petty and Oliver, 2005; Oliver et al., 2009, Cooper et al., 2009).

Currently there is a paucity of directly comparable data on the prevalence and phenomenology of both self-injury and aggression across different genetic syndromes and the person characteristics that are associated with these behaviours within and across syndrome groups. These data would be informative for developing models of the causes of these behaviours within and between syndrome groups with implications for intervention. In this study we seek to generate these data for Angelman, Cornelia de Lange, Cri du Chat, Fragile X, Prader-Willi, Lowe and Smith-Magenis syndromes. This study is part of a larger project comparing aspects of the behavioural phenotypes

of the chosen syndromes. (Oliver et al., in review). The current study has two aims; firstly to examine the prevalence and phenomenology of self-injury and physical aggression in the whole sample and across syndrome groups and secondly to evaluate differences in the characteristics of those showing self-injury and physical aggression compared to those who do not in the whole sample and each syndrome group.

Method

Recruitment

Participants with Angelman (AS), Cornelia de Lange (CdLS), Cri du Chat (CdCS), Fragile X (FXS), Lowe (LS), Prader Willi (PWS) and Smith Magenis (SMS) syndromes were recruited. A comparison group (Comp.) of participants with intellectual disability of heterogeneous aetiology was also recruited. Participants were recruited via the: Angelman Syndrome Support Education and Research Trust (membership of approximately 320), Cri du Chat Syndrome Support Group (180), Fragile X Society (male membership of over five years of 432), Prader-Willi Syndrome Association (571), Lowe Syndrome Trust UK (25), Lowe Syndrome Association USA (150) and Smith-Magenis Syndrome Foundation (95). 142 individuals with Cornelia de Lange syndrome and 151 individuals with intellectual disability of heterogeneous aetiology who had previously taken part in a study of the behavioural phenotype of Cornelia de Lange syndrome were contacted directly (Hyman, Oliver and Hall, 2002; Oliver, Sloneem, Hall and Arron, 2009). This group was recruited from schools, residential and day services for people with intellectual disability throughout the UK within which a participant with CdLS was receiving services. A further 234 individuals with Cornelia de Lange syndrome were contacted via the Cornelia de Lange Syndrome Foundation (UK and Ireland). Thus, the total number of carers of individuals with Cornelia de Lange syndrome contacted was 376. Overall, approximately 2,300 individuals were contacted for participation in the study.

862 (35.24%) carers returned the questionnaires. Individuals under the age of four were excluded as some measures were not appropriate. Information regarding the diagnosis of genetic syndromes was obtained in order to establish the validity of diagnosis. Data on participants were excluded from the study if they did not have a diagnosis from a General Practitioner, Clinical Geneticist, Paediatrician,

Neurologist and Psychiatrist or if a large proportion (more than 25% of items on individual questionnaires) of information was missing. After excluding participants, 797 individuals were included in the study. The overall return rate was 35% (range 27% (CdLS) to 44% (SMS and FXS).

Procedure

A covering letter, information sheet, questionnaire pack, consent form and prepaid return envelope were sent to carer's of prospective participants who were asked to complete and return questionnaires and the consent form.

Participants

Table 1 shows the number of participants, mean age and range, the percentage of males, verbal and mobile individuals in each group and estimates of ability. Participants ranged in age from 4 to 52 years (mean 16.46, SD 9.88) and 65.1% were male. The Wessex Scale, (Kushlick et al., 1973) was used to describe levels of ability (self help skills), mobility (ability to walk unaided), visual impairment and hearing impairment. Overall, 573 (71.9%) of participants were able or partly able, 468 (58.7%) were fully mobile, 575 (72.1%) had normal vision and 691 (86.7%) had normal hearing. 545 (68.4%) of participants were verbal (used more than 30 words or signs).

+++++++++ (place Table 1 about here) +++++++++++++++

Measures

The questionnaires sent to carers were: a demographic questionnaire, the Wessex Scale (Kushlick, Blunden & Cox, 1973), the Autism Screening Questionnaire (ASQ; Berument et al., 1999), The Activity Questionnaire (TAQ; Burbidge & Oliver, 2008; Burbidge et al., in review) and an adapted version of the Mood, Interest and Pleasure Questionnaire (Ross & Oliver, 2003; Ross et al., 2008).

Demographic Questionnaire. The demographic questionnaire detailed age, gender, mobility, verbal ability, diagnostic status.

Wessex Scale (Kushlick et al., 1973). The Wessex Scale is an informant questionnaire designed to assess social and physical abilities in children and adults with intellectual disabilities. Subscales include continence, mobility, self help skills, speech and literacy and information on vision and hearing is also included. The Wessex Scale has good inter-rater reliability at subscale level for both children and adults (Kushlick *et al*., 1973).

Autism Screening Questionnaire (Berument et al., 1999). The Autism Screening Questionnaire was developed as a tool for screening for autism spectrum disorders in children and adults and is based on the Autism Diagnostic Interview. The measure consists of 40 items which are grouped into three subscales: communication; social interaction and repetitive and stereotyped patterns of behaviours. The ASQ shows good concurrent validity with the Autism Diagnostic Interview and with the Autism Diagnostic Observation Schedule (Berument et al., 1999). Internal consistency is also good (α = .90 for the total scale; Berument et al., 1999). No inter-rater or test-retest reliability data have been reported by the authors.

Activity Questionnaire (Burbidge & Oliver, 2008). The Activity Questionnaire is an information-based questionnaire designed to evaluate hyperactivity and impulsivity and is appropriate for use with people with intellectual disability including those with severe or profound intellectual disability. The questionnaire comprises eighteen items grouped into three subscales: overactivity (score range 0=36), impulsivity (0-24) and impulsive speech (0-12). Factor analysis and internal consistency of subscales confirm the integrity of the subscales (Burbidge et al., in review). Items are scored on a five-point Likert scale with responses ranging from 0 (never/almost never) to 5 (always/almost all of the time). Item level inter-rater reliability ranges from 0.31 to 0.75 (mean 0.56) and test-retest reliability ranges from 0.60 to 0.90 (mean 0.75). Inter-rater and test-retest reliability indices for subscales and total score exceed 0.70.

Mood, Interest and Pleasure Questionnaire (MIPQ; Ross & Oliver, 2003; Ross, et al., 2008). The Mood, Interest and Pleasure Questionnaire is an informant based questionnaire used to assess two constructs related to depression, mood and, interest and pleasure. It is designed for use with people with intellectual disability including those with severe or profound intellectual disabilities. Informants rate twenty five items describing operationally defined observable behaviours to give a

total score, a Mood subscale score and an Interest and Pleasure subscale score. A shorter version of this measure was developed (MIPQ-S) in which twelve items from the original measure were selected (six from each subscale) on the basis of their item total correlation and ensuring that all the original constructs of mood, interest and pleasure were included. This version shows good internal consistency (Cronbach's alpha coefficients: total = 0.88, Mood = 0.79, Interest and Pleasure = 0.87), test-retest (0.97) and inter-rater reliability (0.85). Each item is rated using a five point Likert scale to give a total score of between 0 and 48 where 48 is the maximum score indicating positive affect and elevated interest and pleasure

Assessment of self-injurious behaviour and aggression. The Challenging Behaviour Questionnaire (CBQ; Hyman, Oliver and Hall, 2002) is a brief informant based questionnaire evaluating the presence or absence of self-injury, physical aggression, verbal aggression, destruction of property and inappropriate vocalisations over the last month. The measure also examines eight topographies of self-injurious behaviour, adapted from Bodfish et al. (1995). Items regarding self-injury and physical aggression were used for the current study. Previous examination of the psychometric properties of the questionnaire has demonstrated good inter-rater reliability with reliability coefficients ranging from 0.61 to 0.89 (Hyman, Oliver and Hall, 2002).

Procedure

The study was subject to ethical review. Questionnaires were distributed to families and carers of children and adults diagnosed with syndromes via the relevant syndrome support groups. Full details are provided in (Oliver et al., in review).

Data Analysis

Examination of skewness, kurtosis and results of Kolmogorov-Smirnov tests indicated that parametric tests could be used to examine age. Scores on the subscales of the measures employed (Mood, Interest and Pleasure, Communication Deficit, Social Interaction, Repetitive Behaviour, Overactivity, Impulsivity, Compulsive Behaviour and Stereotyped Behaviour did not evidence a normal distribution for comparisons between those showing behaviours and those who did not

within groups. As there was also large variation in the size of each syndrome group, non-parametric tests were used to analyse these measures. The percentages of individuals showing self-injurious behaviour and physical aggression in the total sample and each of the syndrome groups were derived from the CBQ. In order to examine physical aggression, self-injury and topographies of self-injury across the syndrome groups, odds ratio statistics were calculated comparing the likelihood of individuals in each syndrome group showing self-injury and physical aggression with the comparison group of individuals with mixed aetiology intellectual disabilities. Due to the large number of odds ratios being conducted 99% confidence intervals were used to indicate significant results.

Potential differences in characteristics of those showing of self-injurious and aggressive behaviour were examined separately for each group. Participants showing self-injury or physical aggression were compared to those who did not show the behaviour. Chi-square statistics were applied to categorical data including age group (categorised into 4-10 years, 11-20 years, 21 years and above), gender, speech, ability, mobility, vision and hearing. The Mann-Whitney test was used to examine the subscale scores of the MIPQ-S, ASQ, RBQ and AQ. In the Smith-Magenis syndrome group, due to the low number of individuals not showing self-injury, statistical comparisons including self-injury could not be conducted. This process of analysis was then repeated for physical aggression. Due to the high number of statistical tests being conducted, an alpha value of less than .01 was used to indicate significance where appropriate.

Results

Demographic Characteristics

The mean age of the 797 participants was 16.46 years (standard deviation, 9.88 years). 35% of the sample was female, with 69% verbal, 61% mobile and 32% able. 73% of the participants had normal vision and 88% had normal hearing. The mean age, standard deviation, and information regarding gender, level of ability, mobility, speech and sensory impairments within the eight participant groups are presented in table 1. Results of statistical comparisons are detailed in (anonymised for blind review). In summary: individuals in the Angelman syndrome group were significantly younger than individuals in the comparison group, Cornelia de Lange, Cri du Chat,

Fragile X, and Prader-Willi syndrome groups. As only males with Fragile X and Lowe syndrome were recruited for the study, expected significant differences were found for gender in these groups. Individuals with Angelman syndrome showed significantly less speech than all the other groups and were of lower ability than individuals in the comparison group and with Cri du Chat, Fragile X, Lowe, Prader-Willi and Smith Magenis syndrome. The Cornelia de Lange syndrome group showed less speech than the other syndrome groups, excluding Angelman and Cri du Chat syndrome groups Individuals with Cornelia de Lange and Smith Magenis syndrome showed poorer hearing than other groups. The Lowe syndrome group had significantly poorer vision than all other groups, while individuals with Fragile X syndrome showed greater vision than other groups. Individuals in the Fragile X and Prader-Willi syndrome groups generally showed increased levels of ability and speech than other groups. These group differences are consistent with published literature.

Prevalence of Self-Injury and Physical Aggression

55.8% of the total participants showed self-injury and 52.8% showed physical aggression. Table 2 shows the percentage of individuals who had displayed self-injury and physical aggression in the last month in each of the groups. Odds ratios and 99% confidence intervals calculating the likelihood of individuals in each syndrome group showing self-injury and physical aggression compared to the comparison group are shown in Table 2. Individuals in all syndrome groups except Angelman syndrome were significantly more likely to show self-injury than the comparison group. It is notable that individuals in the Smith-Magenis syndrome group were at least 6.32 times more likely to show self-injury. Physical aggression was significantly more likely to be shown by individuals with Angelman and Smith-Magenis syndromes than the comparison group. Odds ratios, used to compare the topographies of self-injury shown by individuals in each syndrome group with the comparison group, are presented in Table 3.

+++++++ (Place Tables 2 and 3 about here) ++++++++++

No specific topographies of self-injury were more likely to be shown by individuals with Angelman syndrome than the comparison group. Individuals with Prader-Willi syndrome were more likely to

show rubbing or scratching, Lowe syndrome participants were more likely to display inserting, and individuals with Fragile X syndrome were more likely to demonstrate biting. Individuals with Cri du Chat syndrome were more likely to show pulling and rubbing or scratching and people with Cornelia de Lange syndrome were more likely to show hitting and pulling. A range of topographies of self-injury were more likely to be demonstrated by individuals with Smith-Magenis syndrome.

In summary, self-injury was significantly more likely to be shown by individuals with Cornelia de Lange, Cri du Chat, Fragile X, Lowe, Prader-Willi and Smith Magenis syndromes than the comparison group. Individuals with Angelman and Smith Magenis syndromes were more likely to show physical aggression. Specific topographies of self-injury were identified in some syndrome groups including rubbing or scratching in Prader-Willi syndrome, inserting in Lowe syndrome and biting in Fragile X syndrome.

Predictors of Self-Injury and Physical Aggression within Groups

The second aim of the study was to examine differences in specific variables between individuals showing self-injury compared to those not showing self-injury and individuals showing physical aggression compared to individuals not showing physical aggression within each of the groups. Table 4 shows the significant differences in demographic characteristics of participants in each group with and without self-injury and with and without physical aggression. Chi-square statistics indicated that age, gender, speech, mobility, vision and hearing were not associated with self-injury in any of the groups. Having a lower level of ability was more likely in those individuals with Cornelia de Lange syndrome who showed self-injury. The presence of physical aggression was more likely to occur in younger individuals (aged 4 to10 and 10 to 20 vs. 21+) with Cri du Chat (χ (2) = 14.23, p=.001), Fragile X (χ (2) = 14.95, p=.001) and Prader-Willi syndromes (χ (2) = 12.75, p=.002). Individuals with physical aggression in the Cornelia de Lange (χ (2) = 14.48, p=.001) and Prader-Willi syndromes (χ (2) = 11.86, p=.003) groups were more likely to be of lower ability. Being male was associated with physical aggression in Prader-Willi syndrome (χ (1) = 7.43, p=.006).

++++++++ (Place Table 4 about here) ++++++++++++

In summary, individuals with self-injury had lower levels of ability than those without self-injury in the Cornelia de Lange syndrome group. Compared to participants without physical aggression, participants with physical aggression in the Cri du Chat, Fragile X and Prader-Willi syndrome groups were younger, individuals with Cornelia de Lange and Prader-Willi syndrome were also of lower ability and individuals with Prader-Willi syndrome were more likely to be male.

To explore the difference in mood, autism spectrum behaviours, hyperactivity and repetitive behaviours between participants with and without self-injury and physical aggression in each syndrome group Mann-Whitney tests were conducted. Table 5 summarises these analyses.

++++++++++ (Place Table 5 about here) ++++++++++++++

Table 5 shows that there were significant differences in a cluster of behaviours between people showing and not showing self-injury in the Cornelia de Lange, Fragile X, Prader-Willi and Lowe syndrome groups. Individuals in these groups with self-injury displayed higher scores on measures of autistic like Repetitive Behaviour, Overactivity and Impulsivity. However, these behaviours did not vary with the presence of self-injury in the Comparison, Angelman and Cri du Chat syndrome groups. Individuals with self-injury also had significantly lower scores for Interest and Pleasure in Cornelia de Lange, Fragile X and Prader-Willi syndrome and higher scores on autistic like Social Interaction in people with Cornelia de Lange and Fragile X syndrome. In the comparison and Cri du Chat syndrome groups, no variables were associated with the presence of self-injury. Individuals with self-injury in the Angelman syndrome group showed significantly lower scores on Mood.

In contrast to self-injury, the data in Table 5 demonstrate that individuals with physical aggression showed significantly higher scores on Impulsivity across all the groups, except Smith Magenis

syndrome, and higher scores on Overactivity in Cri du Chat, Cornelia de Lange, Fragile X, Prader-Willi and Smith Magenis syndrome. At least one other variable in each of the groups was significantly different in relation to the presence of physical aggression. In the Angelman syndrome group, individuals with physical aggression showed lower scores on autistic like Social Interaction. People showing physical aggression also showed lower Mood scores in Cri du Chat and Smith-Magenis syndrome, higher levels of autistic like Repetitive Behaviour and Compulsive behaviour in Cornelia de Lange syndrome and Lowe syndromes.

In summary, significantly higher scores were demonstrated in a cluster of behaviours in people with Cornelia de Lange, Fragile X, Prader-Willi and Lowe syndrome showing self-injury compared to individuals not showing self-injury. These included autistic like repetitive behaviour, overactivity and impulsivity. These differences were not observed between those showing and not showing self-injury in the comparison group, Angelman and Cri du Chat groups. In contrast, individuals showing physical aggression demonstrated higher scores on measure of overactivity or impulsivity across all the syndrome groups.

Discussion

The data show that individuals with Cri du Chat, Cornelia de Lange, Fragile X, Lowe, Prader-Willi, and Smith Magenis syndromes are more likely to show self-injury than a comparison group. For most syndrome groups the odds ratio ranged from three and nine. Notably, participants with Smith Magenis syndrome were at least six times and approximately 35 times more likely to show self-injury. Consistent with previous research there is no association between Angelman syndrome and self-injury (Summers and Feldman, 1999). The prevalence of physical aggression was significantly higher in the Angelman and Smith Magenis syndrome than the comparison group. Thus, the raised probability of self-injury in groups does not extend uniformly to physical aggression.

The results are consistent with previous research findings indicating that Fragile X, Prader-Willi and Smith Magenis syndromes are associated with self-injury (Clarke et al., 1996; Dykens and Smith, 1998; Finucane, Dirrigl and Simon, 2001; Symons et al., 2003) and support suggestions that

Cri du Chat, Cornelia de Lange and Lowe syndromes are associated with self-injury (Berney, Ireland and Burn, 1999; Collins and Cornish, 2002; Hyman, Oliver and Hall, 2002). However, previous studies have indicated that clinically significant self-injury in Cornelia de Lange syndrome is not more prevalent than in individuals matched for age, gender, ability and mobility (Oliver et al., 2009). Use of a single matched comparison group was not possible or consistent with the aims of this study and this difference suggests that the demographics of the comparison group are important to consider when interpreting results.

The prevalence data and broader results should be considered in relation to the methodology adopted in this study. A survey methodology using measures of known psychometric properties informants has the advantages of being able to sample a wide array of environments known to the informant and across time but may be compromised by the absence of direct observational data. The majority of participants with genetic syndromes were recruited via support groups and it has been hypothesised that families and carers are more likely to join support groups if they care for a person showing challenging behaviour (Hyman, Oliver and Hall, 2002). However, if evident, this bias is comparable across groups and therefore comparisons of self-injury and the correlates of self-injury within the syndrome groups remain valid. Additionally, the consistency between the data generated in this study and past research for this sample on SIB and aggression and other behaviours (Oliver et al., in review) suggests the samples were representative. Although the study incorporated a large sample of participants, some of the genetic syndromes examined are rare consequently the numbers of participants in specific syndrome groups, such as Lowe and Smith-Magenis syndromes, were small. Finally, the resultant odds ratios are influenced by the prevalence of the behaviours of interest (and their correlates) in the comparison group. The proportion of people showing self-injury and aggression in the comparison group is higher than many prevalence estimates and this may indicate a bias in this sample. These high proportions indicate that the odds ratios are conservative estimates for increased risk in the genetic groups.

Comparisons of topographies of self-injury indicated that single topographies of self-injury are associated with specific syndromes. Consistent with previous research, Fragile X syndrome was associated with self biting (Symons et al., 2003) and Prader-Willi syndrome with scratching (Clarke

et al, 1996; Dykens and Smith, 1998). Individuals with Smith Magenis syndrome were more likely to show a range of topographies of self-injury, those with Cri du Chat syndrome were more likely to show self pulling and rubbing or scratching, and self hitting and pulling was more probable in Cornelia de Lange syndrome. Previous studies examining self-injury in Smith Magenis, Cri du Chat and Cornelia de Lange syndrome illustrate that various topographies of self-injury are common (Collins and Cornish, 2002; Dykens and Clarke, 1997; Dykens and Smith, 1998; Finucane, Dirrigl and Simon, 2001; Hyman, Oliver and Hall, 2002). This is a notable difference from Fragile X and Prader-Willi syndrome. Finally, the results of the study indicate a novel finding, demonstrating that individuals with Lowe syndrome were more likely than the comparison group to show inserting objects or body parts. However, the item on the self-injury questionnaire relating to inserting objects and body parts included eye poking. 88% of the participants with Lowe syndrome had a visual impairment; therefore this finding may reflect increased eye poking associated with visual impairment. Previous literature has suggested that inserting objects and body parts is more likely in individuals with Smith Magenis syndrome (Dykens and Smith, 1998). This association in the current study may have been masked by the inclusion of eye poking in the definition of inserting objects and body parts.

The examination of the demographic correlates of self-injury within syndrome groups revealed that lower level of ability was associated with self-injury in people with Cornelia de Lange syndrome only. In line with the whole sample, lower levels of ability were more common in individuals with self-injury. The presence of physical aggression showed differences across a number of demographic variables in people with Cri du Chat, Cornelia de Lange, Fragile X and Prader-Willi syndrome. This suggests that the demographic variables relating to self-injury do not generalise to physical aggression in the syndrome groups.

The study demonstrates a specific clustering of behaviours associated with self-injury in Cornelia de Lange, Fragile X, Lowe and Prader-Willi syndrome only. In these groups with self-injury was associated with repetitive behaviour, overactivity and impulsivity. Overactivity and impulsivity may also be important in relation to self-injury in people with Smith Magenis syndrome. The Smith Magenis syndrome group showed the highest prevalence of self-injury and also displayed very high

scores on the measure of impulsivity (Oliver et al., in review). However, it was not possible to examine this relationship in people with Smith Magenis syndrome in the current study. There were no differences in these behaviours between those with and without self-injury in people with mixed aetiology intellectual disability, Angelman and Cri du Chat syndrome. A very different profile was seen in relation to physical aggression. Overactivity or impulsivity, but not repetitive behaviour, was associated with the presence of physical aggression in all participants, regardless of group. The findings in relation to self-injury provide evidence to support previous suggestions that self-injury is associated with repetitive behaviours in people with Prader-Willi syndrome (Clarke et al., 2002; Dykens, Cassidy and King, 1999) and forms of repetitive behaviour in Cornelia de Lange and Fragile X syndrome (Moss et al., 2009; Hyman, Oliver and Hall, 2002; Symons, Clark, Hatton, Skinner and Bailey, 2003), However, the study also establishes impulsivity and overactivity as important correlates of self-injury in specific syndromes and, more generally, aggression.

The results have clear implications for theories of self-injury and aggression in people with intellectual disability. An exclusively operant account cannot explain differences in prevalence or phenomenology across syndromes or within syndrome differences in person characteristics between those who show self-injury and aggression and those who do not. However, there is evidence that operant theory is applicable to these behaviours seen in people with these syndromes (Hall, Oliver and Murphy, 2001; Arron et al., 2006; Taylor and Oliver, 2008, Strachan et al., 2009). It is clear therefore that more complete causal models of self-injury and aggression must encompass both person characteristics (including those of genetic origin) and environmental factors (Oliver, 1993; 1995).

References

Barkley, R. A. (1997). Behavioral inhibition, sustained attention, and executive functions: Constructing a unifying theory of ADHD. *Psychological Bulletin, 121*, 65-94.

1 Barkley, R. A. (1999). Response inhibition in Attention Deficit Hyperactivity Disorder. *Mental Retardation and Developmental Research Reviews, 5,* 177-184.

2

Berney, T. P., Ireland, M., & Burn, J. (1999). Behavioural phenotype of Cornelia de Lange syndrome. *Archives of Disease in Childhood, 81*, 333-336.

Berument, S. K., Rutter, M., Lord, C., Pickles, A., & Bailey, A. (1999). Autism Screening Questionnaire: Diagnostic validity. *British Journal of Psychiatry, 175,* 444-451.

Bodfish, J. W., & Lewis, M. H. (2002). Self-injury and comorbid behaviors in developmental, neurological, psychiatric, and genetic disorders. In S. R. Schroeder, M. L. Oster-Granite & T. Thompson (Eds.), *Self injurious behavior: Gene brain behavior relationships.* (Vol. 39, pp. 1-55798). Washington: American Psychological Association.

Bodfish, J. W., Crawford, T. W., Powell, S. B., Parker, D. E., Golden, R. N., & Lewis, M. H. (1995). Compulsions in Adults with Mental-Retardation - Prevalence, Phenomenology, and Comorbidity with Stereotypy and Self-Injury. *American Journal on Mental Retardation, 100,* 183-192.

Bodfish, J. W., Symons, F. J., Parker, D. E., & Lewis, M. H. (2000). Varieties of repetitive behavior in autism: Comparisons to mental retardation. *Journal of Autism and Developmental Disorders, 33*, 237-243.

Burbidge, C. & Oliver, C. (2008). Activity Questionnaire: Manual for administration and scorer interpretation. University of Birmingham.

Burbidge, C., Oliver, C., Moss, J., Arron, K., Berg, K., Hill, L., Trusler, K. and Furniss, F. (In review). The association between repetitive behaviours, impulsivity and hyperactivity in people with intellectual disability. *Journal of Intellectual Disability Research*

Clarke, D. J., & Boer, H. (1998). Problem behaviors associated with deletion Prader-Willi, Smith-Magenis, and cri du chat syndromes. *American Journal on Mental Retardation, 103*, 264-271.

Clarke, D. J., Boer, H., Chung, M. C., Sturmey, P., & Webb, T. (1996). Maladaptive behaviour in Prader-Willi syndrome in adult life. *Journal of Intellectual Disability Research, 40,* 159-165.

Clarke, D. J., Boer, H., Whittington, J., Holland, A., Butler, J., & Webb, T. (2002). Prader-Willi syndrome, compulsive and ritualistic behaviours: the first population-based survey. *British Journal of Psychiatry, 180,* 358-362.

Collacott, R. A., Cooper, S. A., Branford, D., & McGrother, C. (1998). Epidemiology of self-injurious behaviour in adults with learning disabilities. *British Journal of Psychiatry, 173,* 428-432.

Collins, M. S. R., & Cornish, K. (2002). A survey of the prevalence of stereotypy, self-injury and aggression in children and young adults with Cri du Chat syndrome. *Journal of Intellectual Disability Research, 46,* 133-140.

Cooper, S,A,, Smiley, E., Jackson, A., Finlayson, J., Allan, L., Mantry, D., & Morrison, J. (2009). Adults with intellectual disabilities: prevalence, incidence and remission of aggressive behaviour and related factors. *Journal of Intellectual Disability Research,* **53**, 217-232.

Cooper, S,A,, Smiley, E., Allan, L., Jackson, A., Finlayson, J., Mantry, D., & Morrison, J. (2009). Adults with intellectual disabilities: prevalence, incidence and remission of self-injurious behaviour and related factors. *Journal of Intellectual Disability Research,* **53**, 200-216.

Dykens, E. M., & Clarke, D. J. (1997). Correlates of maladaptive behavior in individuals with 5p- (cri du chat) syndrome. *Developmental Medicine and Child Neurology, 39*, 752-756.

Dykens, E. M., & Smith, A. C. M. (1998). Distinctiveness and correlates of maladaptive behaviour in children and adolescents with Smith-Magenis syndrome. *Journal of Intellectual Disability Research, 42*, 481-489.

Dykens, E. M., Cassidy, S. B., & King, B. H. (1999). Maladaptive behavior differences in Prader-Willi syndrome due to paternal deletion versus maternal uniparental disomy. *American Journal on Mental Retardation, 104*, 67-77.

Einfeld, S. L., Smith, A., Durvasula, S., Florio, T., & Tonge, B. J. (1999). Behavior and emotional disturbance in Prader-Willi syndrome. *American Journal of Medical Genetics, 82*, 123-127.

Emerson, E., Kiernan, C., Alborz, A., Reeves, D., Mason, H., Swarbrick, R., Mason, L., & Hatton, C. (2001). Predicting the persistence of severe self-injurious behavior. Research in Developmental Disabilities, 22(1), 67-75.

Finucane, B., Dirrigl, K. H., & Simon, E. W. (2001). Characterization of self-injurious behaviors in children and adults with Smith-Magenis syndrome. *American Journal on Mental Retardation, 106*, 52-58.

Hanley, G. P., Iwata, B. A. & McCord, B. E. (2003). Functional analysis of problem behavior a review. *Journal of Applied Behavior Analysis, 36*, 147-185.

Hassiotis, A., Parkes, C., Jones, L., Fitzgerald, B. & Romeo, R. (2008). Individual characteristics and service expenditure on challenging behaviour for adults with intellectual disabilities. *Journal of Applied Research in Intellectual Disabilities, 21, 438-445.*

Hastings, R. P. (2002). Parental stress and behaviour problems of children with developmental disability. *Journal of Intellectual and Developmental Disability, 27, 149-160.*

Holland, A. J., Whittington, J. E., Webb, B. T., Boer, H., & Clarke, D. (2003). Behavioural phenotypes associated with specific genetic disorders: evidence from a population-based study of

people with Prader-Willi syndrome. *Psychological Medicine, 33*, 141-153.

Hyman, P., Oliver, C., & Hall, S. (2002). Self-injurious behavior, self-restraint, and compulsive Behaviors in Cornelia de Lange syndrome. *American Journal on Mental Retardation, 107*, 146-154.
Kenworthy, L., & Charnas, L. (1995). Evidence for a Discrete Behavioral-Phenotype in the Oculocerebrorenal Syndrome of Lowe. *American Journal of Medical Genetics, 59*, 283-290.

Kuschlick, A., Blunden R., & Cox, G. (1973). A method of rating behaviour characteristics for use in large scale surveys of mental handicap. *Psychological Medicine, 3,* 466-478.

Lesniak-Karpiak, K., Mazzocco, M. M. M., & Ross, J. L. (2003). Behavioral assessment of social anxiety in females with Turner or fragile X syndrome. *Journal of Autism and Developmental Disorders, 33*, 55-67.

Marston, G. M., Perry, D. W., & Roy, A. (1997). Manifestations of depression in people with intellectual disability. *Journal of Intellectual Disability Research, 41*, 476-480.

McClintock, K., Hall, S., & Oliver, C. (2003). Risk markers associated with challenging behaviours in people with intellectual disabilities: a meta-analytic study. *Journal of Intellectual Disability Research, 47*, 405-416.

Moss, J. & Oliver, C. (2008). Repetitive Behaviour Questionnaire: Manual for administration and scorer interpretation. University of Birmingham.

Moss, J., Oliver C., Arron, K., Burbidge, C. & Berg, K. (2009).The prevalence and phenomenology of repetitive behavior in genetic syndromes. *Journal of Autism and Developmental Disorders. 39, 572-588.*

Oliver, C. (1995). Annotation: Self-injurious behaviour in children with learning disabilties: Recent advances in assessment and intervention. *Journal of Child Psychology and Psychiatry, 30*, 909-927.

Oliver, C., Sloneem, J., Hall, S., and Arron, K. (2009). Self-injurious behavior in Cornelia de Lange syndrome.: 1. Prevalence and phenomenology. *Journal of Intellectual Disability Research,* **53**, 575-589.

Oliver, C., Berg, K., Burbidge, C, Arron K. and. Moss, J. (In review). Delineation of behavioral phenotypes in genetic syndromes. Autism spectrum disorder, affect and hyperactivity. *Journal of Autism and Developmental Disorders.*

Petty, J. and Oliver, C. (2005). Self-injurious behaviour in people with intellectual disability. *Current Opinion in Psychiatry, 18*, 484-489.

Powell, S. B., Bodfish, J. W., Parker, D., Crawford, T. W., & Lewis, M. H. (1996). Self-restraint and self-injury: Occurrence and motivational significance. *American Journal on Mental Retardation, 101*, 41-48.

Rojahn, J., Matson, J. L., Naglieri, J. A., & Mayville, E. (2004). Relationships between psychiatric conditions and behavior problems among adults with mental retardation. *American Journal on Mental Retardation, 109*, 21-33.

Ross, E. & Oliver, C. (2003). Preliminary analysis of the psychometric properties of the Mood, Interest and Pleasure Questionnaire (MIPQ) for adults with severe and profound learning disabilities. *British Journal of Clinical Psychology*, **42**, 81-93.

Ross E., Oliver, C. & Arron, K. (2008). The Mood, Interest and Pleasure Questionnaire (MIPQ): Manual for administration and scorer interpretation. University of Birmingham.

Strachan, R., Shaw, R., Burrow, C., Horsler, K., Allen, D. and Oliver, C. (2009). Experimental functional analysis of aggression in children with Angelman Syndrome. *Research in Developmental Disabilities, 30, 1095-1106.*

Summers, J. A., & Feldman, M. A. (1999). Distinctive pattern of behavioral functioning in Angelman syndrome. *American Journal on Mental Retardation, 104*, 376-384.

Symons, F. J., Clark, R. D., Hatton, D. D., Skinner, M., & Bailey, D. B. (2003). Self-injurious behavior in young boys with fragile X syndrome. *American Journal of Medical Genetics Part A, 118A*, 115-121.

Turner, M. (1999). Annotation: Repetitive behaviour in autism: A review of psychological research. *Journal of Child Psychology and Psychiatry, 40*, 839-849.

Table 1: Percentage of females, mean age (standard deviation) and percentage of participants who were verbal, mobile and able in each syndrome group.

Syndrome Group	Mean age[1] (SD)	% female	% verbal	% mobile	% able or partly able[3]	% normal vision	% normal hearing
Comparison group	18.25 (10.03)	35.7	60.0	36.4	64.3	67.3	81.8
Angelman	13.40 (7.97)	44.2	1.9	46.1	33.0	87.5	100.0
Cri du Chat	17.20 (12.16)	63.8	67.2	53.7	62.1	84.5	82.8
Cornelia de Lange	17.49 (17.49)	59.4	45.5	59.2	53.5	67.3	66.0
Fragile X	16.57 (8.81)	0.0[2]	88.9	70.4	90.1	88.9	97.9
Prader-Willi	17.04 (10.86)	47.3	96.3	73.0	96.6	71.9	94.9
Lowe	16.20 (10.32)	0.0[†]	74.5	46.4	64.3	12.7	92.9
Smith Magenis	15.45 (8.86)	59.5	81.0	73.2	78.6	65.9	56.1

[1] In years

[2] Only male participants were included in the study.

[3] Based on ability to feed, dress and wash independently or with some help.

Table 2: Percentage of individuals showing self-injury and physical aggression in each group. Odds ratios and 99% confidence intervals are shown to demonstrate the likelihood of individuals in each syndrome group showing self-injury and physical aggression compared to the comparison group. Bold text indicates a significant difference ($p<.01$).

	Self-injurious behaviour		Physical aggression	
Group	Percentage	Odds ratio (99% CI's)	Percentage	Odds ratio (99% CI's)
Comparison group	26.8		46.3	
Angelman	45.1	2.24 (0.89-5.69	73.0	3.14 (1.26-7.8)
Cri du Chat	76.8	9.04 (2.93-27.88)	70.2	2.73 (0.98-7.60)
Cornelia de Lange	70.3	6.47 (2.48-16.86)	40.2	0.78 (0.32-1.88)
Fragile X	51.3	2.88 (1.22-6.82)	52.1	1.26 (0.57-2.80)
Prader-Willi	51.6	2.91 (1.23-6.91)	43.0	0.88 (0.39-1.95)
Lowe	64.3	4.92 (1.71-14.17)	64.8	2.1 (0.77-5.90)
Smith Magenis	92.9	35.53 (6.32-199.92)	73.8	3.27 (1.04-10.27)

Table 3: Odds ratios demonstrating the likelihood of individuals in each syndrome group showing topographies of self-injury compared to the comparison group (significant results i.e. where range of confidence interval is above 1, are highlighted in bold).

Syndrome Group	Hits self with body	Hits self against object	Hits self with object	Bites self	Pulls self	Rubs/ scratches self	Inserts
Angelman	0.83	1.60	2.94	0.92	0.96	0.91	1.26
Cri du Chat	3.05	2.98	5.75	2.89	10.90	4.51	2.44
Cornelia de Lange	4.03	2.69	4.71	2.55	4.99	3.04	3.83
Fragile X	1.85	0.87	0.88	3.39	1.87	1.09	0.79
Prader-Willi	0.45	0.44	0.29	0.31	1.64	6.04	0.73
Lowe	3.92	2.56	3.86	2.98	3.55	2.04	6.68
Smith Magenis	8.49	6.36	12.10	11.50	9.75	2.96	1.76

Table 4: Demographic variables showing significant difference between participants with and without self-injury and with and without physical aggression in each group. *P*-values of <.01 were used to indicate significance.

Syndrome	Demographic variable		Percentage of individuals with self-injury	Percentage of individuals without self-injury	χ^2	*P* value
Cornelia de Lange	Level of ability	Able	5.6	33.3	14.27	.001
		Partly Able	40.8	36.7		
		Not Able	53.5	30.0		

Syndrome	Demographic variable		Percentage of individuals with physical aggression	Percentage of individuals without physical aggression	χ^2	*P* value
Cri du Chat	Age	4-10 years	47.5	23.5	14.23	.001
		11-20 years	37.5	11.8		
		> 21 years	15.0	64.7		
Cornelia de Lange	Level of ability	Able	2.6	20.7	14.48	.001
		Partly Able	59.0	24.1		
		Not Able	38.5	55.2		
Fragile X	Age	4-10 years	36.7	14.4	14.95	.001
		11-20 years	48.0	53.3		
		> 21 years	15.3	32.2		
Prader-Willi	Gender	Female	35.4	55.7	7.43	.006
		Male	64.6	44.3		
	Age	4-10 years	39.2	27.4	12.75	.002

	11-20 years	43.0	30.2		
	> 21 years	17.7	42.5		
Level of	Able	45.5	69.4	11.86	.003
ability	Partly Able	51.9	26.5		
	Not Able	2.6	4.1		

Table 5: Differences in affect, autism spectrum behaviours, hyperactivity and repetitive behaviours shown by participants showing and not showing physical aggression/self-injurious behaviour within syndrome groups. + = significantly **higher score for SCQ, RBQ and TAQ subscales or significantly lower score for MIPQ subscales for indvidiuals within each syndrome group showing physical aggression/self-injurious behaviour at the p<.01 level, ++ = p<.001. / = incalculable due to group size (see text).**

Self-injurious behaviour

Measure	Subscale	Comp.	AS	CdC	CdLS	FXS	PWS	LS	SMS
MIPQ	Mood	-	++	-	-	-	-	-	/
	Interest and pleasure	-	-	-	+	+	+	-	/
SCQ	Communication	-	/	-	-	-	+	-	/
	Socialisation	-	/	-	+	+	-	-	/
	Rep. Behaviour	-	-	-	+	+	+	+	/
TAQ	Overactive	-	-	-	++	++	+	++	/
	Impulsive	-	-	-	++	++	+	+	/
RBQ	Compulsive Behaviour	-	-	-	-	-	+	-	/
	Stereotyped Behaviour	-	-	-	++	++	-	+	/

Aggression

Measure	Subscale	Comp.	AS	CdC	CdLS	FXS	PWS	LS	SMS
MIPQ	Mood	-	-	+	-	-	-	-	+
	Interest and pleasure	-	-	-	-	-	-	-	-
SCQ	Communication	-	/	-	-	-	-	-	-
	Socialisation	-	/	-	-	-	-	-	-
	Rep. Behaviour	-	-	-	+	-	-	+	-
TAQ	Overactive	-	-	+	+	++	++	-	+
	Impulsive	+	+	+	++	++	++	++	-
RBQ	Compulsive Behaviour	-	-	-	++	-	-	+	-

Stereotyped Behaviour	-	-	-	-	-	-	-	-