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ORIGINAL PAPER

The Prevalence and Phenomenology of Repetitive Behavior in Genetic Syndromes

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Abstract We investigated the prevalence and phenomenology of repetitive behavior in genetic syndromes to detail profiles of behavior. The Repetitive Behaviour Questionnaire (RBQ) provides fine-grained identification of repetitive behaviors. The RBQ was employed to examine repetitive behavior in Angelman ($N = 104$), Cornelia de Lange ($N = 101$), Cri-du-Chat ($N = 58$), Fragile X ($N = 191$), Prader-Willi ($N = 189$), Lowe ($N = 56$) and Smith-Magenis ($N = 42$) syndromes and individuals with intellectual disability of heterogeneous aetiology ($N = 56$). Repetitive behavior was variable across syndromes. Fragile X syndrome scored highly on all subscales. Angelman syndrome demonstrated a significantly lowered probability for most behaviors. Prader-Willi, Cri-du-Chat and Smith-Magenis syndrome evidenced unique profiles of repetitive behavior. There is extreme heterogeneity of repetitive behavior across genetic syndromes, highlighting syndrome specific profiles.

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Repetitive behavior is an umbrella term used to describe behaviors characterised by frequency of repetition, inappropriateness and invariance (Turner 1997). The term 'repetitive behavior' is employed across different populations including those with neurological, psychological and developmental disorders. This universal application of terminology has implications for the way in which the aetiology, development and maintenance of these behaviors are conceptualised. It is important to identify the nature of repetitive behavior within and between populations in order to establish whether the use of universal terms is justified (Baron-Cohen 1989) and appropriate. This is particularly important within the intellectual disability population where differences in aetiology might underlie differences in behavior.

There is increased research interest in studying behavioral phenotypes as a means of understanding behavior disorder in individuals within the broader intellectual disability population. Examples within the syndrome literature are apparent in which the specificity of cognitive and behavioral associations within a genetic syndrome have enabled inferences to be made regarding potential aetiological pathways of repetitive behaviour at both the cognitive and neurobiological level. In Fragile X syndrome, a deficit in executive functioning has been identified (Wilding et al. 2002). Although there is no evidence for a causal link, a deficit of this kind has been suggested to account for the heightened prevalence of repetitive behavior in other populations including autism

spectrum disorders (Turner 1997) and Obsessive Compulsive Disorder (Greisberg and McKay 2003). Similarly, studies of repetitive behavior in Cornelia de Lange syndrome have identified an association between repetitive behaviors and the presence of self- injury, self-restraint behaviors and hyperactivity (Hyman et al. 2002; in review citation anonymised for blind review), suggesting that a deficit in behavior regulation might be a common underlying factor (Petty and Oliver 2005). Recent research within Prader-Willi syndrome has demonstrated the presence of a short-term memory deficit in affected individuals (Dykens et al. 2000) and compromised capacity for attention switching (in review citation anonymised for blind review) and these specific cognitive deficits might account for the repetitive questioning and preference for routine, which is commonly reported in the syndrome (Dykens et al. 1996). At the neurobiological level, study of mutant mouse models of a range of neurodevelopmental disorders has also revealed potential aetiological pathways for repetitive behavior. For example, mutant mouse models of Rett Syndrome (RS), with mutations on the MECP2 gene demonstrate repetitive forelimb movements similar to those characteristically observed in girls with RS. Similarly, GABRB3 knockout mice show intense stereotyped behaviors. The GABRB3 gene lies within the q11-13 region of chromosome 15. Mutations in this specific region are associated with Prader-Willi and Angelman syndromes both of which are reported to show increased levels of repetitive behaviour. Other mutant mouse models with links to Down Syndrome and obsessive compulsive disorder have also been reported to show increased rates of repetitive and stereotyped behaviour (Lewis et al. 2007). To date, these associations between aetiology and behavior are largely speculative. However, these examples demonstrate how the study of behavioral phenotypes provides insight into potential aetiology of behavior. In order to develop this line of research, detailed description of the phenomenology of repetitive behavior across syndromes is warranted.

This study will focus on repetitive behavior within Angelman (AS), Cri du Chat (CdC), Cornelia de Lange (CdLS) Fragile X (FXS), Prader-Willi (PWS), Lowe (LS) and Smith-Magenis (SMS) syndromes, each of which demonstrates an association with repetitive behavior. Table 1 provides a summary of the genetic markers, prevalence, degree of disability and reported repetitive behavior in these syndromes.

The table highlights a number of methodological and conceptual issues. The study of repetitive behavior within these syndromes has largely focused on investigating stereotyped behavior. Relatively little attention has been paid to 'compulsive' behavior and 'obsessions'. This is likely to reflect the fact that current definitions of 'compulsive' behaviors and 'obsessions' are difficult to apply to all individuals with intellectual disability. Furthermore, and probably downstream from these definitional issues, few assessments of 'compulsive' behavior and 'obsessions' suitable for use with individuals with intellectual disability have been developed. Subjective experiences are often central to the way in which 'compulsive' behavior and 'obsessions' are defined and identified (see the following definitions: APA 1987, 1994; Lewis and Bodfish 1998; Rachman and Hodgson 1980). This requires a level of insight and self report that is not always possible to ascertain within this population (Baron-Cohen 1989). The table highlights some of the difficulties that arise when this terminology is employed within the intellectual disability population. For example, the term 'Obsessive Compulsive Disorder' has been employed to describe repetitive behavior in Prader-Willi syndrome (Dykens et al. 1996). However, the specific topographies of behavior described within the syndrome include ordering, rituals and hoarding. These are very different to those reported within individuals with Obsessive Compulsive Disorder (Dykens et al. 1996), suggesting that the application of this term may not be entirely appropriate. Developing suitable assessments of repetitive behaviour, which are based on definitions of behavior that are appropriate for the intellectual disability population, is essential in order to ensure accurate description of phenomenology.

It is also notable that the reported prevalence rates of each class of repetitive behavior (stereotyped behavior, 'compulsive' behavior, 'obsessions') described within the table, are extremely variable within and between syndrome groups. The variability *within* syndrome groups is likely to reflect differences regarding terminology, definition and assessment of repetitive behavior across different studies. The variability *between* syndrome groups is more informative, highlighting the varied profile of repetitive behavior across genetic syndromes. Studies of repetitive behavior in Prader-Willi syndrome indicate a lower prevalence of stereotyped behavior (Clarke and Boer 1998) and a heightened prevalence of 'compulsive' behavior (Dykens et al. 1996; Dykens and Kasari 1997). In Fragile X syndrome there is a heightened prevalence of both of these subtypes of repetitive behavior (Backes et al. 2000; Hagerman and Lampe 1999; Mazzocco et al. 1998), suggesting a generalised heightened probability of repetitive behavior. Describing the nature of these different profiles in detail using a consistent measure of repetitive behavior across groups will enable further insight into the causal factors that underlie these differences.

Table 1 Summary of genetic markers, prevalence, degree of disability and prevalence of repetitive behavior in Angelman, Cornelia de Lange, Cri du Chat, Fragile X, Lowe, Prader-Willi and Smith-Magenis syndromes

Syndrome	Genetic marker and prevalence	Degree of ID	Stereotyped behavior		Compulsive behavior		Obsessions		Other
			Prevalence	Topography	Prevalence	Topography	Prevalence	Topography	
Cornelia de Lange	Chr 5, p13[1] ChrX, p11.2[2] Chr 10[2a] 1 in 10–50,000[3]	Mild–profound[3]	57%[4]	Body rocking Body postures Spinning objects[5]	87.5%[4]	Unknown	Unknown	Unknown	Upset by changes in routine[5]
Cri du Chat	Chr 5, p15[6] 1 in 50, 000[6]	Moderate–severe[7]	47.2–100%[8] CdC = ID[8]	Rocking[9]	Unknown	Unknown	66.6%[10]	Attachment to objects[10]	Echolalia[10]
Angelman	Chr 15, q11-13[11] 1 in 12–25,000[12]	Severe–profound[13]	9–84%[14] AS>DS, PWS[15]	Hand flapping[14]	Unknown	Unknown	Unknown	Unknown	Unknown
Prader-Willi	Chr 15, q11–13[16] 1 in 10–40,000[17]	Mild–moderate[18]	PWS<CdC, SMS[19]	Unknown	68%[20] OCD[21] PWS>ID, DS, typical, AS[22]	Ordering Rituals Hoarding[21]	88%[20] PWS>ID, DS[20]	Unknown	Need to tell/ask[21] Repetitive speech[23] Insistence on sameness[24]
Fragile X	Chr X, q27.3[25] 1 in 4,000 (m) 1 in 8,000 (f)[26]	Mild–moderate[27]	69.2–74.2%[28] FXS>ID, DS, AUT[29]	Hand flapping[30]	2% OCD[27]	Unknown	71%[31]	Restricted interests[31]	Echolalia upset by changes in environment[30]
Lowe	Chr X, q25–26 1 in 200,000[32]	Normal–profound[32]	85% LS>ID[33]	77% repetitive hand movements, mannerisms, habits[33]	Unknown	Unknown	Unknown	Unknown	Unknown
Smith-Magenis	Chr 17, p11.2[34] 1 in 25,000[35]	Moderate–profound[35]	100%[36] SMS>PWS, CdC[37]	Mouthing objects/ hand teeth grinding Body rocking Spinning/twirling[36] Lick and flip[38]	Unknown	Unknown	Unknown	Restricted interests Obsessional thinking[39]	Self hug[40] preference for adults Repetitive speech Preference for routine[39]

[1] Krantz et al. (2004); Tonkin et al. (2004); [2] Musio et al. (2006); [2a] Deardorff et al. (2007); [3]Opitz (1985); [4] Hyman et al. (2002); [5] Sarimski (1997); [6] Neibuhr (1978); [7] Cornish et al. (1999); [8] Cornish and Pigram (1996); Ross Collins and Cornish (2002); [9] Ross Collins and Cornish (2002); [10] Cornish and Pigram (1996); [11] Magenis et al. (1987); [12] Buckley et al. (1998); [13] Cassidy et al. (2000); [14] Summers et al. (1995); [15] Walz and Benson (2002); [16] Ledbetter et al. (1982); [17] Burd et al. (1990); [18] Dykens et al. (2000); [19] Clarke and Boer (1998); [20] Dykens and Kasari (1997); [21] Dykens et al. (1996); [22] Dykens and Kasari (1997); Walz and Benson (2002); [23] Clarke et al. (1996); [24] Steinhausen et al. (2002); Wigren and Hansen (2005); [25] Mazzocco (2000); [26] Dykens et al. (2000); [27] Backes et al. (2000); [28] Hagerman and Lampe (1999); Mazzocco et al. (1998); [29] Baumgardner et al. (1995); Hagerman et al. (1986); [30] Hagerman et al. (1986); Mazzocco et al. (1998); [31] Mazzocco et al. (1998); [32] Nussbaum and Suchy (2001); [33] Kenworthy et al. (1993); Kenworthy and Charnas (1995); [34] Smith et al. (1982); [35] Smith and Gropman (2001); [36] Dykens and Smith (1998); [37] Clarke and Boer (1998); [38] Dykens et al. (1997); Dykens and Smith (1998); [39] Dykens and Smith (1998); Smith and Gropman (2001); [40] Dykens and Smith (1998); Finucane et al. (1994)

In addition to the syndrome related profiles, the table demonstrates that highly specific and, in some instances unusual or apparently unique, repetitive behaviors have been identified within particular syndrome groups. For example, the lick and flip and self hug behaviors described in Smith Magenis syndrome (Dykens et al. 1997; Dykens and Smith 1998; Finucane et al. 1994; Smith and Gropman 2001) and the attachment to objects in Cri du Chat syndrome (Cornish and Pigram 1996). These highly specific behaviors are masked when a class level of description is employed, highlighting the need to describe behaviors at a fine-grained level.

To date, much of the research into repetitive behavior in genetic syndromes has been conducted using single syndrome cohort descriptions or limited comparisons. A systematic study of these behaviors using the same standardised assessment across groups has not yet been undertaken and this is the main aim of this study. Conducting a comparison across several syndrome groups using the same assessment would extend the existing descriptions of behavioral phenotypes and could prove important in identifying the underlying aetiological pathways of the behaviors (Hodapp and Dykens 2001). In order to generate useful data in this study a number of methodological and conceptual problems related to the definition and assessment of repetitive behavior will be addressed. Accurate identification of the nature of behaviors requires the use of a fine-grained approach and the use of appropriate terminology and definitional criteria applicable across a range of intellectual ability.

This study is part of a larger project comparing aspects of the behavioral phenotypes of the chosen syndromes. The comparison of the prevalence and phenomenology of self-injury and aggression are reported in (citation withheld for blind review) and for autism spectrum behaviors, affect and hyperactivity in (citation withheld for blind review). In this study, we seek to develop a fine-grained measure of repetitive behavior based on operational definitions with robust psychometric properties that is suitable for individuals with a wide range of intellectual disability and employ the measure to assess the nature of repetitive behavior in individuals with Angelman, Cornelia de Lange, Cri du Chat, Fragile X, Lowe, Prader-Willi and Smith-Magenis syndromes and individuals with intellectual disability of heterogeneous cause. Each of the selected syndrome groups have previously been demonstrated within the literature to show repetitive behaviours of varying frequency and phenomenology. These groups are not intended to be an exhaustive list of those syndrome groups which demonstrate repetitive behaviours but reflect a sample of syndrome groups with whom the researchers have conducted previous research.

Methods

Recruitment

A total of 2,446 participants with Angelman, Cornelia de Lange, Cri du Chat, Fragile X, Prader-Willi, Lowe and Smith Magenis syndromes and a group of individuals with intellectual disability of heterogeneous cause were invited to participate. Carers were contacted via the following syndrome support groups: Angelman Syndrome Support Education Research Trust, Cri du Chat Syndrome Support Group, Fragile X Society, Prader-Willi Syndrome Association, Lowe Syndrome Trust UK (and Lowe Syndrome Association USA) and Smith-Magenis Syndrome Foundation. In addition to this, 142 carers of individuals with Cornelia de Lange syndrome and 142 carers of individuals with intellectual disability of heterogeneous cause, who had been involved in previous research studies, were contacted directly. Remaining members of the Cornelia de Lange Syndrome Foundation (UK and Ireland) who had not taken part in previous studies were contacted via the Foundation. All carers received a covering letter, an information sheet, questionnaire pack and consent form. In order to avoid the effects of priming, the study was presented to carers as an investigation of behaviors associated with genetic syndromes.

Individuals with Angelman, Cri du Chat, Cornelia de Lange, Fragile X,[1] Prader-Willi, Lowe and Smith-Magenis syndromes were included in the study if they had a diagnosis of the given syndrome from a professional such as a paediatrician, clinical geneticist or physician. In the total sample, 43.7% of participants were diagnosed by a paediatrician and 46.6% were diagnosed by a clinical geneticist, 1.1% were diagnosed by their GP and 8.3% were diagnosed by another professional such as a neurologist (largely AS participants), ophthalmologist (largely LS participants) and an endocrinologist (largely PWS participants). Individuals who had additional chromosomal abnormalities were excluded from analyses. Any individual in the heterogeneous intellectual disability group diagnosed with any of the seven genetic syndromes included in the current study were excluded from analyses. Individuals with other genetic syndromes not participating in the current study were included in the heterogenous intellectual disability group including: Down ($N = 5$), Aicardi ($N = 1$), Hypomelanosis Ito ($N = 1$), Landau Kleffner ($N = 1$), Lennox Gastrout ($N = 1$), Miller Deiker ($N = 1$), Pierre Robin ($N = 1$), Rett ($N = 1$) and Soto ($N = 1$) syndromes,

[1] Only males aged five and upwards were contacted through the Fragile X Society due to an error during administrative procedures. Females with Fragile X syndrome were not included in the study due to reported differences in phenotypic characteristics (Dykens et al. 2000).

Cerebral Palsy ($N = 5$) and Trisomy 9 ($N = 1$). Individuals under the age of 4 years were excluded from analyses because one of the measures employed was only appropriate for those aged 4 years and upwards. Individuals who had not provided information regarding age or date of birth were excluded from the analysis. Individuals who were missing information on over 75% of items in the total questionnaire pack (which included seven different questionnaire measures) were not included in the study.

Participants

A total of 862 (35.24%) carers returned the questionnaires. 65 (7.5%) individuals were excluded based on the criteria described above. Five participants (.58%) were excluded due to missing information, 37 participants (3.13%) were excluded due to age and 28 participants (3.25%) were excluded due to diagnosis. Table 2 describes the characteristics of the remaining participants ($N = 797$). All participants were aged between 4 and 51 years (mean = 16.46; SD = 9.88) and 519 (65.1%) participants were male, 573 (71.9%) participants were able or partly able (score above six on the self help subscale of the Wessex Scale; Kushlick et al. 1973). 468 (58.7%) participants were fully mobile, 545 (68.4%) participants were verbal (more than 30 words/signs in their vocabulary), 575 (72.1%) participants had normal vision and 691 (86.7%) participants had normal hearing. No significant differences between the participant groups were revealed for age or gender.[2] Significant differences between the participant groups were identified on the following variables: level of ability, mobility, verbal ability, vision, hearing and presence of autistic phenomenology ($p < .001$).

Measures

The distributed questionnaire pack included a demographic questionnaire, the Autism Screening Questionnaire (ASQ; Berument et al. 1999), the Wessex Scale (Kushlick et al. 1973), and the Repetitive Behaviour Questionnaire (Moss and Oliver 2008). The present study was conducted as part of a larger postal survey. Three additional questionnaires were distributed to parents and carers, the Activity Questionnaire (Burbidge and Oliver 2008), the Challenging Behaviour Questionnaire (Hyman et al. 2002) and the Mood Interest and Pleasure Questionnaire—Short form (Ross et al. 2008), the results of which are not reported in the present study (see: citation withheld for blind review).

[2] FXS and LS groups excluded from analysis due to the X linked nature of the syndromes and exclusion of females in the FXS group.

Demographic Questionnaire

The demographic questionnaire provided information regarding date of birth, gender, mobility (able to walk unaided), verbal ability (more than 30 signs/words) and diagnostic status (whether or not a diagnosis had been made, the precise diagnosis made, when and by whom).

Autism Screening Questionnaire (ASQ, Berument et al. 1999)

The Autism Screening Questionnaire is used to screen for the presence of autism spectrum disorders in individuals of all age groups. The measure consists of forty items that comprise three subscales: communication, social interaction and repetitive and stereotyped patterns of behaviors. The authors suggest a cut-off point for autism spectrum disorder of fifteen. This score was found to differentiate individuals with Pervasive Developmental Disorders from other diagnoses (excluding those with intellectual disability) with a specificity of .80 and a sensitivity of .96 and differentiated individuals with autism from individuals with intellectual disability with a specificity of .67 and a sensitivity of .96. A higher cut-off point of 22 or more is required to differentiate individuals with autism from other Pervasive Developmental Disorders with a sensitivity of .75 and a specificity of .60. Internal consistency is good ($\alpha = .90$ for the total scale; Berument et al. 1999). A prorated communication subscale score was employed in the present study using the completed nonverbal items for all participants, in order to ensure that scores were comparable across syndrome groups and that groups with a high proportion of nonverbal individuals were not disadvantaged on this subscale.

Wessex Scale (Kushlick et al. 1973)

The Wessex Scale is an informant questionnaire designed to assess the social and physical characteristics of children and adults with intellectual disability. The Wessex Scale comprises five subscales including: continence, mobility, self help skills, speech and literacy. The Wessex Scale also provides information on vision and hearing. Inter-rater reliability at subscale and item level is good (Kushlick et al. 1973; Palmer and Jenkins 1982).

Repetitive Behaviour Questionnaire (RBQ; Moss and Oliver 2008)

The Repetitive Behaviour Questionnaire is an informant questionnaire for use with children and adults with a range

Table 2 Demographic characteristics, mean scores (and standard deviation) on the Autism Screening Questionnaire, statistical analyses and post hoc analyses for all participant groups: Angelman (AS), Cornelia de Lange (CdLS), Fragile X (FXS), Prader Willi (PWS), Lowe (LS), Smith Magenis (SMS) syndromes and individuals with intellectual disability of heterogenous cause (HID)

		HID	AS	CdC	CdLS	FXS	PWS	LS	SMS	F/χ^2	df	*p* value	Post hoc analyses
N*		56	104	58	101	191	189	56	42				
Age**	Mean	18.25	13.40	17.20	17.49	16.57	17.04	16.20	15.45	2.08	7	ns	N/A
	(SD)	(10.03)	(7.97)	(12.16)	(9.87)	(8.81)	(10.86)	(10.32)	(8.86)				
	Range	6–38	4–45	4–44	4–40	6–47	4–51	4–51	4–38				
Gender	% Male	64.3	55.8	36.2	40.6	100	52.7	100	40.5	208.72	7	ns[e]	N/A
Self Help[a]	% Partly able/able[b]	64.3	33.0	62.1	53.5	90.1	96.6	64.3	78.6	190.19	7	<.001	HID, CdC, FXS, PWS, LS, SMS>AS FXS>HID, CdC, CdLS LS, PWS>HID, CdC, CdLS, LS, SMS
Mobility[a]	% Mobile[c]	36.4	46.1	53.7	59.2	70.4	73.0	46.4	73.2	49.98	7	<.001	PWS>HID, AS, LS FXS>HID, AS SMS>HID
Vision[a]	% Normal	67.3	87.5	84.5	67.3	88.9	71.9	12.7	65.9	146.92	7	<.001	FXS>HID, CdLS, PWS, SMS HID, AS, CdC, CdLS, FXS, PWS, SMS>LS
Hearing[a]	% Normal	81.8	100	17.2	66.0	97.9	94.9	92.9	59.0	128.53	7	<.001	AS>HID, CdC, CdLS, SMS FXS>HID, CdC, SMS, CdLS PWS>CdLS, SMS LS>CdLS, SMS
Speech[d]	% Verbal	60.0	1.9	67.2	45.5	88.9	96.3	74.5	81.0	349.57	7	<.001	HID, CdC, CdLS, FXS, PWS, LS, SMS>AS SMS, LS, PWS, FXS>CdLS PWS>HID, CdC, LS, FXS>HID, CdC,
ASQ	Mean score	20.52	20.15	13.64	21.00	19.82	13.19	17.48	17.57	19.98	7	<.001	FXS, CdLS, AS>PWS, CdC HID>PWS
	(SD)	(7.80)	(5.83)	(6.92)	(7.84)	(6.79)	(6.97)	(7.10)	(6.55)				

* N may vary across analysis due to missing data

** In years

[a] Data derived from the Wessex Scale (Kushlick et al. 1973)

[b] Those scoring six or above on the total score of the self help subscale (items g–i). Categories collapsed due to small N in some samples

[c] Those scoring six on the total score of the mobility subscale (items e & f). Categories collapsed due to small N in some samples

[d] Data derived from item 3 of the demographic questionnaire

[e] FXS and LS groups excluded from analysis since all participants are male due to the X linked nature of both of the syndromes and exclusion of females in the FXS group

of intellectual abilities. It is suitable for use with verbal and non-verbal individuals and for individuals who fall within the autistic spectrum. The Repetitive Behaviour Questionnaire consists of nineteen items that comprise five subscales: stereotyped behavior, compulsive behavior, insistence on sameness, restricted preferences and repetitive speech. All items are based on operationally defined features of behaviors. Informants rate the frequency of behavior over the preceding month. The response format consists of a five-point Likert-type rating scale ranging from 'never' to 'more than once a day'.

Development of the RBQ

Behaviors included in the repetitive behavior questionnaire were identified by reviewing items/behaviors assessed in other measures of repetitive behavior that have been employed in previous research studies. Measures were selected for review if they were suitable for use with children, had robust psychometric properties and were informant-based assessments. Larger measures containing subscales designed to assess repetitive behavior were excluded. It was considered that items on these large scale measures would not have sufficient detail for the purpose of the current study. The measures that were used to identify behaviors for the Repetitive Behavior Questionnaire included: Childhood-Yale Brown Obsessive Compulsive Scale (CY-BOCS; Goodman et al. 1990), Stereotyped Behavior Scale (Rojhan et al. 1997), Compulsive Behavior Checklist (CBC; Gedye 1992), Childhood Routines Inventory (CRI); Evans et al. 1997); Repetitive Behaviour Scale-Revised (RBS-R; Bodfish et al. 1998).

Behaviours were selected from each measure based on the following criteria: (a) they could be defined in terms of discrete observable behaviors (b) they did not describe self-injurious behavior or involuntary movements such as tics or dyskinetic movements (c) they did not describe specific sensory behaviors associated with autism spectrum disorders such as sniffing, licking or touching. Selected behaviors were categorised into subscales referring to their 'class' of repetitive behavior: stereotyped behavior, compulsive behavior, restricted preferences. Repetitive speech and insistence on sameness. Clear descriptions of observable behavior were developed for each behavior selected for inclusion in the measure and several examples of the behavior were also provided in order to help informants identify behaviors. Table 3 describes each item of the RBQ and indicates which subscale the item falls into.

Scoring: Informants rate the frequency of each behavior over the preceeding month. The following five point likert scale is used to rate each behavior:

Never	Once a month	Once a week	Once a day	More than once a day
0	1	2	3	4

Four items of the Repetitive Behaviour Questionnaire require the individual to be verbal (more than 30 words or signs in their vocabulary) including repetitive questions, echolalia, restricted conversation and attachment to people. As a result, two different scoring methods can be employed. The total score for verbal individuals ranges from 0 to 76. The total score for non-verbal individuals ranges from 0 to 60. A total score excluding non-verbal items was used for all individuals, regardless of verbal ability in the current study in order to enable comparisons across groups. The restricted preferences and the repetitive speech subscales are not scored for individuals who are non-verbal since items on 65% of the subscale require the individual to be verbal.

Those behaviors which occur 'once a day' or 'more than once a day' were deemed to be of clinical importance. Consequently, item level clinical cut-off is attained if an individual endorses a score of three or more on an item. The clinical cut-off at subscale level is attained if and individual endorses a score of three of more on at least one item within the subscale. Missing items on the Repetitive Behaviour Questionnaire are prorated at subscale level. Items are prorated if the informant completes 65% of the relevant subscale.

Reliability and Validity of the Repetitive Behaviour Questionnaire

Reliability data were collected on a sample of 103 individuals with heterogeneous cause of intellectual disability who were recruited through four residential schools and colleges for people with intellectual disability. Participants were aged between 10 and 28 years (mean = 17.6; SD = 3.7). Seventy-three participants (70.9%) were male. Of the 103 participants, 47 (45.6%) were verbal and 87 (84.5%) were mobile. Information regarding gender, mobility and speech were missing for 7 (6.8%) participants. Spearman coefficients for inter-rater reliability ($N = 103$ individuals with intellectual disability of heterogeneous cause) range from .46 to .80 at item level with 73% of items above .60. For clinical cut-off scores, Kappa ranges from .23 to 1.0 at item level with 94% of items above .40. Spearman coefficients for test retest reliability ($N = 103$) ranges from .61 to .93 at item level with 52.6% of items above .80. Kappa scores ranged from .56 to .82 at item level for clinical cut-off scores.

Validity of the Repetitive Behaviour Questionnaire was assessed using the current study participant sample.

Table 3 Repetitive Behaviour Questionnaire items and subscales

	Repetitive Behaviour Questionnaire item:	Subscale
1.	**Object stereotypy:** repetitive, seemingly purposeless movement of objects in an unusual way *E.g. twirling or twiddling objects, twisting or shaking objects, banging or slapping objects.*	Stereotyped behaviour
2.	**Body stereotypy:** repetitive, seemingly purposeless movement of whole body or part of body (other than hands) in an unusual way. *E.g. body rocking, or swaying, or spinning, bouncing, head shaking, body posturing.* Does not include self-injurious behaviour.	
3.	**Hand stereotypy:** repetitive, seemingly purposeless movement of hands in an unusual way. *E.g. finger twiddling, hand flapping, wigging or flicking fingers, hand posturing.* Does not include self-injurious behaviour.	
4.	**Cleaning:** Excessive cleaning, washing or polishing of objects or parts of the body *E.g. polishes windows and surfaces excessively, washes hands and face excessively,*	Compulsive behaviour
5.	**Tidying up:** Tidying away any objects that have been left out. This may occur in situations when it is inappropriate to put the objects away. Objects may be put away into inappropriate places. *E.g. putting cutlery left out for dinner in the bin, removes all objects from surfaces.*	
6.	**Hoarding:** Collecting, storing or hiding objects to excess, including rubbish, bits of paper, and pieces of string or any other unusual items.	
7.	**Organising objects:** Organising objects into categories according to various characteristics such as colour, size, or function. *E.g. ordering magazines according to size, ordering toy cars according to colour, ordering books according to topic.*	
12.	**Rituals:** carrying out a sequence of unusual or bizarre actions before, during or after a task. The sequence will always be carried out when performing this task and will always occur in the same way. *E.g. turning round three times before sitting down, turning lights on and off twice before leaving a room, tapping door frame twice when passing through it.*	
16.	**Lining up or arranging objects***:* Arrangement of objects into lines or patterns *E.g. placing toy cars in a symmetrical pattern, precisely lining up story books,*	
18.	**Completing behaviour:** Insists on having objects or activities 'complete' or 'whole' *E.g. Must have doors open or closed not in between, story must be read from beginning to end, not left halfway through.*	
19.	**Spotless behaviour:** Removing small, almost unnoticeable pieces of lint, fluff, crumbs or dirt from surfaces, clothes and objects. *E.g. Picking fluff off a jumper, removing crumbs from the kitchen table.*	
8.	**Attachment to particular people:** Continually asking to see, speak or contact a particular 'favourite' person. *E.g. continually asks to see or speak to particular friend, carer, babysitter or schoolteacher.*	Restricted preferences
10.	**Attachment to objects:** Strong preference for a particular object to be present at all times. *E.g. Carrying a particular piece of string everywhere, taking a particular red toy car everywhere, attachment to soft toy or particular blanket.*	
13.	**Restricted conversation:** Repeatedly talks about specific, unusual topics in great detail. *E.g. conversation restricted to: trains, buses, dinosaurs, particular film, country, or sport.*	
9.	**Repetitive questions**: Asking specific questions over and over. *E.g. always asking people what their favourite colour is, asking who is taking them to school the next day over and over*	Repetitive speech
11.	**Repetitive phrases/signing:** Repeating particular sounds, phrases or signs that are unrelated to the situation over and over. *E.g. repeatedly signing the word 'telephone'.*	
14.	**Echolalia:** Repetition of speech that has either just been heard or has been heard more than a minute earlier. *E.g.: Mum:' Jack don't do that' Jack: 'Jack don't do that'.*	
15.	**Preference for routine:** Insist on having the same household, school or work schedule everyday. *E.g. likes to have the same activities on the same day at the same time each week, prefers to eat lunch at exactly the same time every day, wearing the same jumper everyday.*	Insistence on sameness
17.	**Just right behaviour:** Strong insistence that objects, furniture and toys always remain in the same place. *E.g. all chairs, pictures and toys have a very specific place that cannot be changed.*	

Concurrent validity and content validity ($N = 797$) between the Repetitive Behaviour Questionnaire and the repetitive behavior subscale of the Autism Screening Questionnaire (Berument et al. 1999) was good (.6; $p < .001$). Internal consistency was good at full-scale level ($\alpha > .80$) and for the stereotyped behavior and compulsive behavior subscales ($\alpha > .70$). Alpha levels for the restricted preferences, repetitive speech and insistence on sameness subscales were lower ($\alpha = .50$, .54 and .65, respectively). The low alpha levels for these subscales are not surprising.

These behaviors are considered to be related in function rather than form, consequently it might be expected that scoring highly on one item within a subscale would not necessarily be related to high scores on all other items within the same subscale.

Data Analysis

All data were tested for normality using Kolmogorov–Smirnov tests. Where data were not normally distributed (<.05), non-parametric techniques were employed. Scores on the Repetitive Behaviour Questionnaire were compared across groups at full-scale, subscale and item levels using Kruskall-Wallis non-parametric analyses of variances. Where significant differences were revealed, post hoc contrasts using pairwise Mann–Whitney *U* tests were conducted. Further item level-analysis was conducted using the clinical cut-off scores. The percentage of participants in each group scoring above the clinical cut-off were compared using a series of Chi Square tests. Where significant differences were revealed, post hoc contrasts using pairwise Chi square tests were conducted in order to identify the source of difference. A conservative alpha level ($p < .001$) was employed throughout analyses.

In accordance with Dykens's comparative approach to behavioral phenotypes (Dykens 1995), high specificity was considered to be present when a given participant group scored significantly *higher* than two or more other groups on a particular item or subscale of the *Repetitive Behaviour Questionnaire*. Low specificity was considered to be present when a given participant group scored significantly *lower* than two or more groups on a particular item or subscale of the Repetitive Behaviour Questionnaire.

The presence of repetitive behavior is one of three core diagnostic characteristics of autism spectrum disorder (APA 1994). In order to examine the association between autism and repetitive behavior, Pearson partial correlations were conducted between the Repetitive Behaviour Questionnaire and the Autism Screening Questionnaire (Berument et al. 1999) using a within group approach at full-scale and subscale levels and on item scores on which high specificity had been identified within the group. Self help score (determined by the Wessex) was partialled out of the correlation.

Due to the non-parametric nature of the Repetitive Behaviour Questionnaire data, differences regarding the demographic characteristics of the group could not be taken into account statistically during the analyses. However, these differences should be borne in mind when considering the results. All significant differences from the post hoc analyses are reported within the data tables. However for conciseness, only significant differences that are greater than or less than two or more other participant groups are reported within the text.

Results

Comparison of Scores on the Repetitive Behaviour Questionnaire

Full-Scale and Subscale Level Analysis

Full-scale and subscale level scores were compared across participant groups using Kruskall-Wallis non-parametric analyses of variances and pairwise Mann–Whitney *U* tests. Mean full-scale, subscale scores and post hoc analyses are reported in Table 4. Significant differences were identified on all subscale and full-scale scores. Post hoc analyses revealed that the Fragile X syndrome group demonstrated significantly higher scores than at least two other groups on three out of five subscales (compulsive behavior, insistence on sameness and repetitive speech subscales) and on the total score. The Angelman and Cri du Chat syndrome groups demonstrated significantly lower scores than at least two other groups on two subscales (compulsive behavior and insistence on sameness subscales) and on the total score. The Prader-Willi syndrome group scored significantly higher than at least two other groups on two subscales (compulsive behavior and insistence on sameness subscales) and significantly lower scores than at least two other groups on one subscale (stereotyped behavior). No significant differences were identified for the Cornelia de Lange, Lowe and Smith Magenis syndrome groups and the heterogeneous intellectual disability group. No significant differences were identified on the restricted preferences subscale.

Item Level Analysis

Item-level scores were compared across participant groups using Kruskall-Wallis non-parametric analyses of variances and pairwise Mann–Whitney *U* tests. Significant differences were revealed on all items with the exception of cleaning and spotless behaviors. Figure 1 demonstrates the repetitive behavior profile in each group, describing the mean scores at item level. The shaded areas represent the subscales of the Repetitive Behaviour Questionnaire. A '+' indicates a significantly higher score than one other group. A '−' indicates a significantly lower score than one other group.

Figure 1 demonstrates that the profile of item level scores across the eight participant groups is highly heterogeneous. The Fragile X syndrome group scored significantly higher than two or more other groups on eight items (hand stereotypy, tidying up, lining up, restricted conversation, preference for routine, just right behavior, repetitive phrases and echolalia). This group demonstrated the highest frequency and greatest number of topographies

Table 4 Mean scores, standard deviations, statistical analyses and post hoc analyses on full-scale and subscale level scores of the Repetitive Behaviour Questionnaire for all participant groups Angelman (AS), Cornelia de Lange (CdLS), Fragile X (FXS), Prader Willi (PWS), Lowe (LS), Smith Magenis (SMS) syndromes and individuals with intellectual disability of heterogenous cause (HID)

	Group								df	χ^2	*p* value	Post hoc analyses
	A HID ($n = 56$) Mean (SD)	B AS ($n = 104$)	C CdC ($n = 58$)	D CdLS ($n = 101$)	E FXS ($n = 191$)	F PWS ($n = 189$)	G LS ($n = 56$)	H SMS ($n = 42$)				
Stereotyped behaviour	5.16 (4.01)	6.16 (3.89)	5.64 (4.22)	6.81 (4.07)	6.73 (5.04)	*3.20 (3.84)*	6.90 (4.32)	6.90 (4.20)	7	94.55	<.001	ABCDEGH>F
Compulsive behaviour	4.66 (5.92)	*1.29 (2.92)*	*2.69 (3.59)*	6.74 (7.65)	**7.21 (7.06)**	**5.71 (6.09)**	6.10 (7.01)	4.43 (4.84)	7	100.91	<.001	ADEFGH>B EF>C
Restricted preferences**	4.36 (2.58)	***	4.13 (2.91)	4.48 (3.43)	5.61 (3.71)	3.97 (3.42)	6.11 (3.84)	5.53 (2.97)	6	27.47	<.001	E>F
Insistence on sameness	2.66 (2.49)	*0.90 (1.75)*	*1.81 (2.50)*	2.67 (2.84)	**4.38 (2.76)**	**3.52 (2.61)**	3.23 (2.82)	3.20 (2.75)	7	131.30	<.001	ADEFGH>B E>ACD F>C
Repetitive speech**	6.33 (3.40)	***	4.93 (4.05)	4.93 (4.05)	**7.31 (3.59)**	4.28 (3.41)	5.52 (3.30)	5.88 (3.85)	6	71.75	<.001	E>CDF
Verbal total score**	23.67 (12.06)	***	*16.39 (9.55)*	26.15 (15.74)	**30.92 (15.66)**	20.78 (15.62)	28.33 (15.09)	26.91 (13.19)	6	56.40	<.001	E>CF G>C
Nonverbal total score	14.77 (10.72)	*9.89 (6.74)*	14.07 (8.45)	19.30 (12.71)	**22.15 (12.90)**	14.45 (12.04)	19.22 (11.98)	17.85 (10.11)	7	92.54	<.001	E>ACF DEGH>B

** Analysis only includes participants who are verbal

*** AS group not included in analysis only 2 participants with AS are verbal

Mean scores reported. Median scores are uninformative with too many zeros

Scores in bold = high specificity (+2 or more other groups) scores in italics = low specificity (−2 or more other groups)

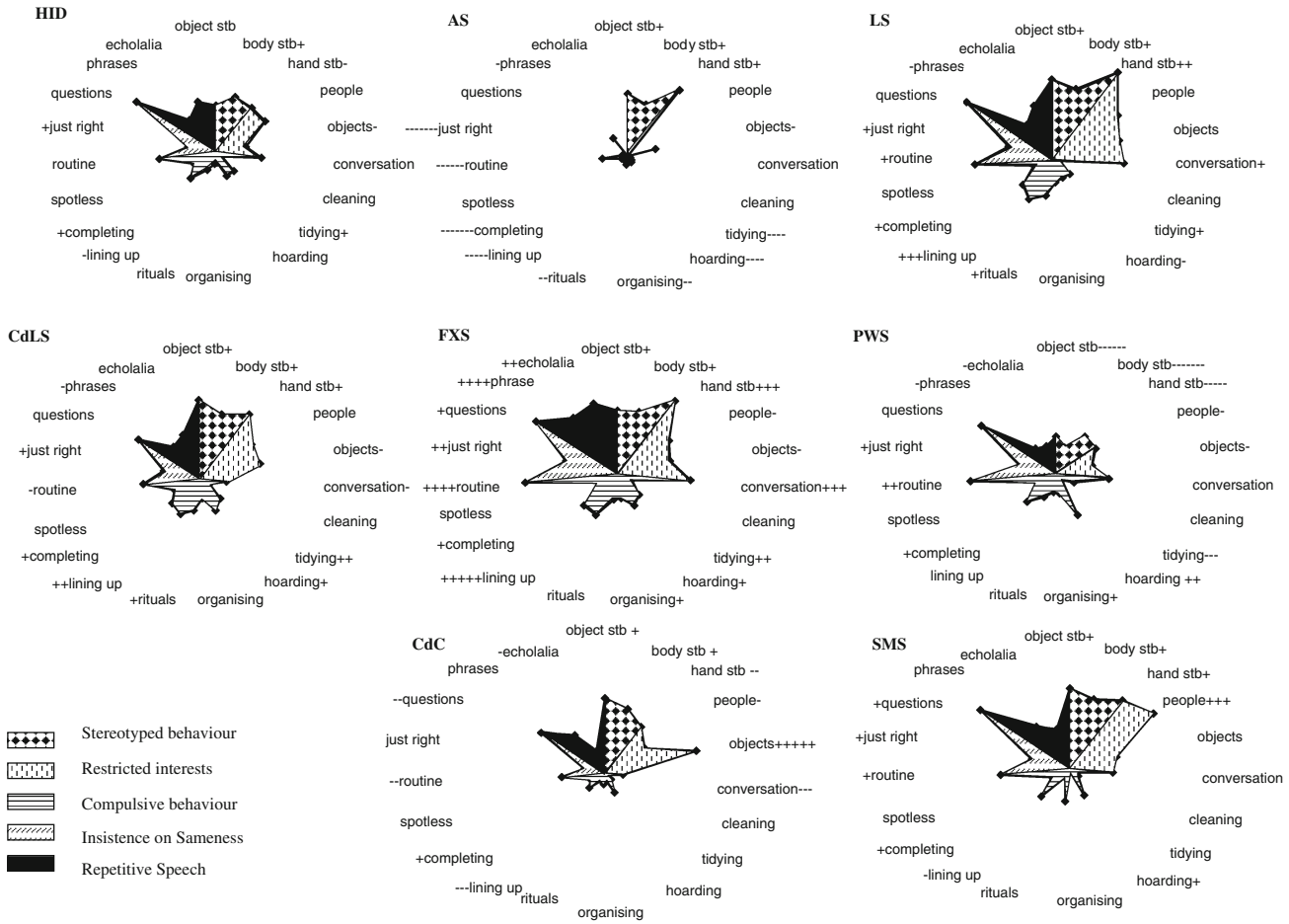

Fig.1 Mean item level scores on the Repetitive Behaviour Questionnaire

of repetitive behavior which is represented by a comparatively larger shaded area in Fig. 1. In direct contrast, the Angelman syndrome group scored significantly lower than two or more other groups on eight items (tidying up, hoarding, organising objects, rituals, lining up objects, preference for routine, just right behavior). The very compact profile on Fig. 1 highlights the low level of specificity of repetitive behavior within this group. These items on the repetitive behaviour questionnaire could all be considered to be 'higher level' repetitive behaviours which require a certain degree of intellectual ability and therefore low scores on these items may reflect the associated severe and profound degree of disability in this group rather than a lack of association with the syndrome.

The Prader-Willi syndrome group demonstrated a notably more mixed profile of repetitive behavior, scoring significantly higher than two or more groups on two items (hoarding behavior and preference for routine) and significantly lower than two or more groups on four items (stereotyped and tidying up behaviors). This is indicated by a very uneven profile in Fig. 1. Two groups demonstrated highly specific profiles. The Smith-Magenis and Cri du Chat syndrome groups both show high specificity on one item only (attachment to people and attachment to objects, respectively). In both cases, these are behaviors for which no other groups have demonstrable high specificity.

The Cornelia de Lange and Lowe syndrome groups demonstrated an interesting profile. In both groups, only two forms of repetitive behavior demonstrate specificity at the level defined for this study. In Cornelia de Lange these included tidying up and lining up behaviors. In Lowe these included hand stereotypies and lining up behaviors. Inspection of Fig. 1 however, indicates that although not reaching statistical significance, the profile of repetitive behavior in these groups might be more generalised than this. The shaded areas within the Lowe and Cornelia de Lange syndrome graphs appear very similar in shape to that of the Fragile X syndrome group.

The heterogeneous intellectual disability group did not score significantly higher or lower than two or more other groups on any of the items. Unlike the other participant groups, the profile of repetitive behavior in this group is not distinctive. Although this group appears to score relatively highly on repetitive questions this is not identified at a

statistical level and the resultant profile is comparatively even.

Clinical Cut-Off Analysis

The percentage of participants scoring above the clinical cut-off in each group was compared at item level using Chi-squared tests and paired Chi-squared post hoc comparisons. Table 5 demonstrates the percentage of individuals scoring above the clinical cut-off in each group and post hoc analyses. The percentage of participants scoring above the clinical cut-off ranges from 1.8 to 71.1% across the participant groups. On those items where high specificity was identified, the percentage of participants scoring above the clinical cut-off ranged from 20.8% to 71.1%. No significant differences were reported for cleaning, hoarding, lining up, repetitive questions and spotless behaviors. Post hoc analyses revealed significant differences in the same direction as those reported for item level scores when using conventional scoring of the questionnaire.

Association with Autism Spectrum Phenomenology

In order to assess the association between autism spectrum disorder and repetitive behaviors Pearson partial correlations (controlling for scores on the self help subscale of the Wessex Scale; Kushlick et al. 1973) were conducted between the Repetitive Behaviour Questionnaire and the Autism Screening Questionnaire (ASQ) at subscale and total score levels and on those items that had been identified as demonstrating high specificity within the syndrome groups. No significant association was identified between repetitive behavior and autism spectrum phenomenology in the Cornelia de Lange, Prader-Willi, Lowe and Smith-Magenis syndromes and the heterogeneous intellectual disability group. In the Angelman syndrome group, scores on the compulsive behavior subscale were significantly, negatively correlated with communication and total scores on the ASQ ($r = -.45$ and $-.41$, respectively). In the Cri du Chat syndrome group, scores on the stereotyped behavior subscale were significantly, positively correlated with the communication, social interaction subscales and total score of the ASQ ($r = .53$, .50 and .60, respectively). In the Fragile X syndrome group, scores on the compulsive behavior, insistence on sameness subscales and the total scores were significantly, positively correlated with scores on the social interaction subscale ($r = .31$, .34, .38, .36) and total score ($r = .29$, .28; total scores only) of the Autism Screening Questionnaire. At item level, just right behavior was significantly, positively correlated with the social interaction subscale of the ASQ ($r = .33$).

Discussion

This study is the first to examine and compare the prevalence and phenomenology of repetitive behavior in individuals with Angelman, Cornelia de Lange, Cri du Chat, Fragile X, Lowe, Prader-Willi and Smith Magenis syndromes and in a group of individuals with intellectual disability of heterogeneous cause. In order to identify repetitive behaviors at a fine-grained level, this study employed a detailed measure of repetitive behavior that was based on operational definitions of behavior, suitable for use in individuals with intellectual disability and demonstrated robust psychometric properties.

A heterogeneous profile of repetitive behavior was evidenced across the participant groups. Individuals with Angelman syndrome demonstrated a lower level of specificity on most forms of repetitive behavior. Previous studies of individuals with Angelman syndrome have noted high rates of hand flapping (Summers et al. 1995). However, the presence of hand stereotypies was not found to be distinctive within this group. It is possible that these behaviors in the Angelman syndrome group were masked by the high prevalence of hand stereotypies identified in other syndrome groups including Lowe and Fragile X syndromes. The Fragile X syndrome group demonstrated a generalised heightened specificity for repetitive behavior, scoring significantly higher than at least two other groups on a number of different items. In this group, hand stereotypies, lining up objects, restricted conversation, preference for routine and echolalia were the most prevalent forms of repetitive behavior. These findings are consistent with previous studies of repetitive behavior in Fragile X syndrome (Backes et al. 2000; Hagerman and Lampe 1999; Hagerman et al. 1986; Mazzocco et al. 1998), which provides some information about the validity of the Repetitive Behaviour Questionnaire. Only one of these behaviors (just right) was found to correlate with scores on the Autism Screening Questionnaire, suggesting that repetitive behaviors in Fragile X syndrome are not entirely related to autism spectrum phenomenology. It is also interesting to note that whilst over 40% of individuals with Angelman syndrome are reported to meet criteria for autism spectrum disorder (reviewed in Abrahams and Geschwind 2008), the scores of the Angelman syndrome group were significantly lower than that of the Fragile X syndrome which is reported to have a 25% prevalence rate of autism spectrum disorder. The low level of reported repetitive behaviour in Angelman syndrome and the poor correlation of repetitive behaviour scores the Fragile X syndrome group to scores on the Autism Screening Questionnaire has implications regarding the association between repetitive behaviour and other aspects of the triad of impairments but also raises some queries regarding the

Table 5 Percentage of individuals scoring above the clinical cut-off score on the Repetitive Behaviour Questionnaire: Item level scores and post hoc contrasts for all participant groups Angelman (AS), Cornelia de Lange (CdLS), Fragile X (FXS), Prader Willi (PWS), Lowe (LS), Smith Magenis (SMS) syndromes and individuals with intellectual disability of heterogenous cause (HID)

	Group								χ^2	*p* value	Post hoc analyses
	A HID ($n = 56$)	B AS ($n = 104$)	C CdC ($n = 58$)	D CdLS ($n = 101$)	E FXS ($n = 190$)	F PWS ($n = 188$)	G LS ($n = 56$)	H SMS ($n = 42$)			
Stereotyped behaviour											
Q1 Object stereotypy	33.9	43.6	51.7	54.5	43.9	*23.4*	50.0	52.4	40.57	<.001	BCDEGH>F
Q2 Body stereotypy	46.4	40.6	41.4	44.6	48.4	*18.6*	48.2	52.4	47.71	<.001	ABCDEGH>F
Q3 Hand stereotypy	44.6	64.7	*37.9*	59.4	**69.1**	*32.4*	**75.0**	59.5	77.12	<.001	BDEG>F EG>C
Compulsive behaviour											
Q4 Cleaning	1.8	1.0	1.7	7.9	10.0	12.8	7.1	4.8	21.21	ns	
Q5 Tidying	16.1	*2.9*	3.4	**20.8**	**19.4**	*2.7*	8.9	0	56.67	<.001	DE>BF
Q6 Hoarding	12.5	5.9	12.1	21.8	22.1	26.6	14.3	19.0	24.39	ns	
Q7 Organising objects	5.4	3.9	5.2	13.9	16.8	9.6	16.1	0	24.23	ns	
Q12 Rituals	5.4	*4.9*	8.6	22.8	18.8	11.2	25.5	26.2	32.49	<.001	DG>B
Q16 Lining up objects	12.5	*4.8*	*3.4*	**27.7**	**29.3**	16.5	**28.6**	7.1	51.17	<.001	DEG>BC
Q18 Completing behaviour	28.6	*1.9*	13.8	23.8	33.2	24.5	21.4	26.2	41.30	<.001	ADEFGH>B
Q19 Spotless behaviour	16.1	2.9	8.6	18.8	15.3	12.2	10.7	11.9	15.33	ns	
Restricted preferences											
Q8 Attachment to people**	33.3	***	23.1	45.7	41.8	25.0	51.2	**67.6**	34.93	<.001	H>CF
Q10 Attachment to objects	21.4	21.6	**67.2**	46.5	40.2	*24.5*	46.4	31.0	59.52	<.001	C>ABEFH D>F
Q13 Restricted conversation**	36.4	***	*10.3*	19.6	**52.4**	32.0	47.5	32.4	38.95	<.001	E>CDF G>C
Insistence on sameness											
Q15 Preference for routine	46.4	*17.8*	*32.8*	41.0	**71.1**	**60.6**	48.2	45.0	94.87	<.001	ADEFG>B E>CD F>C
Q17 Just right behaviour	21.4	*1.9*	12.1	22.8	34.7	21.8	25.0	26.2	46.28	<.001	ADEFGH>B
Repetitive speech											
Q9 Repetitive questions**	75.8	***	38.5	47.8	71.3	61.3	70.0	70.6	24.96	ns	
Q11 Repetitive phrases/signing	32.1	18.3	34.5	28.7	**52.6**	20.2	25.0	35.7	60.54	<.001	E>BDF
Q14 Echolalia**	42.4	50.0	10.3	28.3	**50.3**	*15.6*	36.6	26.5	60.18	<.001	E>CF A>F

** Analysis only includes participants who are verbal

*** not included only 2 participants with AS are verbal

Scores in bold = high specificity (+2 or more other groups) scores in italics = low specificity (−2 or more other groups)

strength of association between Angelman syndrome and autism spectrum disorder. This warrants further investigation.

The Prader-Willi syndrome group demonstrated a more mixed profile of repetitive behavior in which a heightened probability for some forms of repetitive behavior (hoarding

and a preference for routine) and a lowered probability for other behaviors (stereotyped and tidying up behavior) was identified. These findings are largely consistent with previous studies of repetitive behavior in Prader-Willi syndrome (Dykens and Kasari 1997; Dykens et al. 1996; Steinhausen et al. 2002; Wigren and Hansen 2003). However, whilst 61.3% of individuals within the Prader-Willi syndrome group scored above the clinical cut-off on the item referring to repetitive questioning, previously reported to be characteristic of the syndrome (Clarke et al. 1996) this behavior did not demonstrate high specificity within this group. The high frequency of repetitive language identified within the Fragile X syndrome group is likely to have masked the presence of these behaviors in the Prader-Willi syndrome group.

Within the Cri-du-Chat and Smith-Magenis syndrome groups, a more specific profile of repetitive behavior was identified. In these groups, highly specific and apparently unique forms of repetitive behavior were identified. Attachment to objects was highly prevalent within the Cri du Chat syndrome group (67.2% score above clinical cut-off) and attachment to people was highly prevalent within the Smith-Magenis syndrome (67.6% score above clinical cut-off). In both cases, no other groups scored highly on these items and no other highly prevalent behaviors were identified within these groups. These behaviors have previously been described within the literature (Cornish and Pigram 1996; Dykens and Smith 1998; Smith and Gropman 2001) although systematic study of these behaviors using standardised assessments has not been conducted previously. The fine-grained approach to repetitive behavior employed in this study enabled identification of these behaviors.

The Cornelia de Lange and Lowe syndrome groups demonstrate an interesting profile. In both groups, only two forms of repetitive behavior demonstrated specificity at the level outlined in the current study. Individuals with Lowe syndrome scored significantly higher than at least two other groups on hand stereotypies and lining up behaviors. These findings are consistent with previous literature within the syndrome (Kenworthy et al. 1993; Kenworthy and Charnas 1995). Individuals with Cornelia de Lange syndrome scored significantly higher than two other groups on tidying up and lining up behaviors. Whilst previous studies have identified a heightened prevalence of compulsive behavior within the syndrome (Hyman et al. 2002; in review citation withheld for blind review), this is the first study to indicate the precise nature of these behaviors. Although the Cornelia de Lange syndrome group scored highly on the Autism Screening Questionnaire, no correlations were identified between repetitive behavior and autism spectrum phenomenology within this group. Both the Lowe and Cornelia de Lange syndrome groups demonstrated notable similarities with the Fragile X syndrome group with regard to profile of repetitive behaviors, which were not identified at a statistical level.

The profile of repetitive behavior within the heterogeneous intellectual disability group is also noteworthy, although not for the purpose of highlighting a specific pattern of associated behaviors. The profile of repetitive behavior within this group is indistinctive both at subscale and item levels. In the context of the other participant groups, the heterogeneous intellectual disability group could be considered to be the central point of a spectrum of repetitive behaviors on which some participant groups such as the Angelman syndrome group score below this and others such as the Fragile X syndrome group score above. The other participant groups can be placed at various points on items and subscales within this spectrum.

In addition to demonstrating the varied profile of repetitive behavior across these participant groups, the results also highlight some important conceptual and methodological considerations. Analysis at item-level was more informative of the nature of repetitive behavior in all of the participant groups than analyses conducted at subscale and full scale level indicating that a fine-grained approach is essential for enabling accurate identification of behaviors within specific syndrome groups. Additionally, examples are apparent in which a given syndrome group scored highly on one item within a subscale but not on other items within the same subscale. For example, individuals with Prader-Willi syndrome demonstrated a heightened probability of hoarding behavior and a lowered probability of tidying up behaviors. The fact that the prevalence of these behaviors is discrepant within a single syndrome group challenges the class level approach to repetitive behaviors and suggests that considering these behaviors at the level of phenomenology might be beneficial.

There are several limitations of the study that should be noted. Due to the number of different genetic syndromes employed in the study, participants were not comparable on a number of risk markers known to impact on the development of repetitive behavior including: level of ability, mobility, verbal ability, vision and hearing (Ando and Yoshimura 1979; Fazzi et al. 1999; Guess 1966; McClintock et al. 2003; Vitiello et al. 1989). Due to the non-parametric nature of the Repetitive Behaviour Questionnaire data, these differences could not be taken into account at a statistical level. The small sample sizes within some of the syndrome groups restricted the way in which this could be assessed at an individual group level. The results from the current study are relative to the behavior of the other participant groups that were employed. Findings regarding a given syndrome group are highly dependent on who the comparison groups include.

Different results might be obtained with different comparison groups. The use of different sample sizes is also problematic since comparisons between larger groups have a greater level of power than those between smaller groups. Finally, although care was taken in the development of the measure to ensure that the measure encompassed a broad spectrum of repetitive behaviors, there are some forms of repetitive behavior such as the self-hug and the lick and flip behavior reported within Smith Magenis syndrome (Dykens and Smith 1998; Fincuane et al. 1994) that are unaccounted for within this measure.

To summarise, this study describes the prevalence and phenomenology of repetitive behavior associated with genetic syndromes and individuals in the wider intellectual disability population. The study has not only enabled further delineation of the behavioral phenotypes of the syndromes evaluated, but has highlighted important methodological and conceptual issues regarding the study of repetitive behavior within these populations. The findings demonstrate the extreme heterogeneity of repetitive behavior across genetic syndromes, highlighting the importance of adopting a fine-grained approach to repetitive behavior and challenging traditional conceptual approaches to these behaviors. It is the heterogeneity identified across these groups that will be important for identifying the underlying mechanisms and aetiological pathways of repetitive behavior in the future.

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