Repetitive Behavior in Rubinstein–Taybi Syndrome: Parallels with Autism Spectrum Phenomenology

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Repetitive behaviour in Rubinstein-Taybi syndrome: Parallels with autism spectrum phenomenology

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Abstract

Background. Syndrome specific repetitive behavior profiles have been described previously. A detailed profile is absent for Rubinstein-Taybi syndrome (RTS).

Method. The Repetitive Behaviour Questionnaire and Social Communication Questionnaire (SCQ) were completed for children and adults RTS (N = 87), Fragile-X (N = 196) and Down (N = 132) syndromes, and individuals reaching cut-off for Autism Spectrum Disorder (ASD) (N = 228).

Results. Total and matched group analyses were conducted. A phenotypic profile of repetitive behavior was found in RTS. The majority of behaviors in RTS were not associated with social-communication deficits or degree of disability.

Conclusions. Repetitive behavior should be studied at a fine-grained level. A dissociation of the triad of impairments might be evident in RTS.

Keywords: Rubinstein-Taybi syndrome, Autism Spectrum Disorder (ASD), Repetitive behavior, ritualistic behavior,

Repetitive Behavior in Rubinstein-Taybi Syndrome – Parallels with Autism Spectrum Phenomenology

Rubinstein-Taybi syndrome (RTS) occurs in approximately 100,000 - 125,000 live births and is caused by breakpoints, mutations and microdeletions on chromosome 16p13.3 or by a mutation in the E1A-binding protein (p300) or CREB-binding protein (CBP). Intellectual disability ranges from mild to profound with moderate intellectual disability occurring most commonly (Hennekam, 2006; Lacombe, Saura, Taine & Battin, 1992). It has been hypothesised that impairments in RTS may be underpinned by long-term memory deficits associated with mutations in the CREB binding protein (Alarcon et al., 2004; Wood et al., 2005*;* Weeber & Sweatt, 2002). Diagnosis is usually by clinical features such as the characteristic facial phenotype including broad nasal bridge, high arched eyebrows and downwards slanting palpebral fissues. Other characteristics include growth deficiency, microcephaly, broad thumbs and big toes (Hennekam, 2006; Udwin & Dennis, 1995).

Repetitive behavior has been noted as a phenotypic behavioral characteristic in RTS. 'Repetitive behavior' is an umbrella term encompassing a diverse range of behaviors including adherence to routines, insistence on sameness, stereotyped behaviors and restricted preferences (Turner, 1999). Repetitive behavior has traditionally been associated with Autism Spectrum Disorder (ASD), because it forms one aspect of the triad of impairments (World Health Organisation, 1993; American Psychiatric Association, 2013), and with a greater degree of intellectual disability (Bartak & Rutter, 1976; Gabriels, Cuccaro, Hill, Ivers & Goldson, 2005; Smith & VanHouren, 1996). In RTS, Stevens, Carey and Blackburn (1990) found that over 50% of a sample of children (N=50) engaged in stereotyped motor movements including hand stereotypy, spinning and rocking. Over 75% were reported to insist on sameness. Recently, Galéra et al. (2009) reported higher rates of repetitive motor movements in contrast to a heterogeneous intellectual disability group; however, because this study did not contain syndrome specific comparison groups, conclusions cannot be drawn about how these repetitive behaviors compare to behaviors observed in other syndromes. A systematic cross syndrome approach to studying repetitive behavior in RTS is absent from the literature.

A systematic cross syndrome study of repetitive behavior in RTS could be conducted at a broad or fine-grained level of description. Researchers who have studied repetitive behavior in syndrome groups and neurodevelopmental disorders have focused on the severity or frequency of repetitive behavior at a broad level, with behaviors grouped together and described using composite scores (e.g. Lopez, Lincoln, Ozonoff & Lai, 2005). In contrast, Turner (1997) examined repetitive behavior in ASD and noted that some subclasses of repetitive behavior, such as stereotyped behavior, can be further divided into more specific topographies, for example body, hand or object stereotypy. Turner proposed that, in order to understand the aetiology of repetitive behavior, behaviour needs to be described at fine-grained level as specific behaviours may have specific underpinnings.

Moss, Oliver, Arron, Burbidge and Berg (2009) used a fine-grained approach to study repetitive behavior in seven neurodevelopmental disorders, not including RTS, and found significant group differences, many of which were unrelated to degree of disability. These findings suggest that a fine grained approach to studying repetitive behavior in other syndromes might reveal phenotypic repetitive behavior profiles. For example, individuals with Fragile-X syndrome (FXS) were more likely to engage in a greater number of topographies of repetitive behavior in comparison to other syndrome groups and the majority of these behaviors occurred more frequently than at least two groups. In contrast, individuals with Prader-Willi syndrome showed a mixed profile and engaged in hoarding and adherence to routine more frequently than at least two other groups. In addition, Moss et al. found syndrome specific repetitive behaviors in Smith-Magenis (SMS) and Cri du Chat syndromes: a strong preference for a particular people and attachment to specific objects respectively. These findings concur with other studies of behavior in these syndromes (Cornish & Pigram, 1996; Haas-Givler, 1994; Wilde, Silva & Oliver, 2013).

Delineation of the repetitive behavior profile of RTS will have clinical utility and will inform research, as has been demonstrated in other syndrome groups. For example, the observation that a high proportion of individuals with Prader-Willi syndrome are likely to have strong preferences for routines alerts clinicians to aspects of this syndrome that might contribute to a person experiencing anxiety or negative emotions when routines are not followed (Woodcock, Oliver & Humphreys, 2009a). Hence, greater understanding of repetitive behavior in RTS

will aid clinical formulation by helping identify which repetitive behaviors might lead to difficulties. Furthermore, a detailed description of the repetitive behavior in RTS will aid the design of studies that focus on the aetiology of repetitive behavior in RTS. For example, an executive dysfunction account of repetitive behavior has been proposed in ASD and there is also growing evidence that executive dysfunction may underpin specific repetitive behaviors in a number of genetic syndromes, neurodevelopmental disorders and psychiatric conditions such as Obsessive Compulsive Disorder (Lawrence et al., 2006; Lopez et al., 2005; Lysaker, Whitney & Davis., 2009; Turner, 1997; Woodcock, Oliver & Humphreys, 2009a, 2009b, 2009c; Yerys et al., 2009). Woodcock et al. (2009b, 2009c) for example, linked preference for routine in Prader-Willi syndrome to difficulties with cognitive set-shifting.

This study describes the repetitive behavior profile of RTS at a fine-grained level and the profile is compared to three other neurodevelopmental disorders in which repetitive behavior has been described in detail: Fragile-X syndrome (FXS), Down syndrome (DS), and individuals meeting the cut-off for ASD on an ASD Screening Tool: the Social Communication Questionnaire (SCQ; Rutter, Bailey & Lord, 2003). This is the first time the repetitive behavior profile of RTS has been described, and to our knowledge the first time that the repetitive profiles of ASD and DS have been compared to other disorders at a fine-grained level.

FXS, DS and ASD will act as a bench mark against which RTS can be contrasted. Individuals meeting the cut-off for ASD on the SCQ are a suitable contrast group because individuals with ASD engage in a higher intensity and frequency of repetitive behavior than mental age matched comparison groups (Bodfish, Symons, Parker & Lewis, 2000; Hermelin & O'Conner, 1963; Lord, 1995; Lord & Pickles, 1996; Richler, Bishop, Kleinke & Lord, 2007; Watt, Welnerby, Barber & Morgan, 2008). FXS are a suitable group because, similarly to ASD, individuals engage in a wide range of repetitive behaviors and this syndrome is genetically defined in comparison to the ASD cut-off group (Hatton et al., 2006; Moss et al., 2009; Udwin & Dennis, 1995). Finally, DS are a suitable group as there is evidence that individuals with DS engage in fewer repetitive behaviors than those with a codiagnosis of ASD (Hepburn & MacLean, 2009; Moss, Richards, Nelson & Oliver, 2012). In particular,

Moss et al. (2012) found that individuals with DS and ASD engaged in more stereotyped behavior and repetitive speech than those without ASD.

Studying the repetitive behavior profile of RTS in contrast to individuals with ASD and FXS has a further implication. As noted previously, repetitive behavior has been associated with a greater degree of intellectual disability and autism spectrum phemenonology, which includes social and communication deficits. There is debate about whether the triad of impairments in ASD represent a unified deficit or whether the triad can be fractionated (Happe & Ronald, 2008). Individuals with RTS could present with a dissociation of ASD characteristics: repetitive behavior without, or with fewer, social-communication impairments. This is because people with RTS may have fewer social deficits than individuals with ASD as parents often describe RTS children as friendly, particularly around adults (Goots & Liemohn, 1977; Baxter & Beer, 1992; Stevens et al., 1990), and reports suggest a greater degree of sociability in RTS relative to controls (Galéra et al., 2009; Nelson, 2010). While, social behavior is not the main focus of this study, correlational analyses exploring the link between repetitive behavior, social-communication deficits, and degree of disability will be conducted to explore the factors associated with the repetitive behavior profile of RTS.

A direct comparison of these syndromes is difficult given the varying degrees of intellectual and physical disability across groups. For example, FXS is characterised by mild-moderate intellectual disability, RTS by moderate ID, and hearing difficulties are common in DS (Udwin & Dennis, 1995). A greater degree of intellectual disability, poorer vision and poorer hearing are known risk markers for repetitive behavior (Bachara & Phelan, 1980; McClintock, Hall & Oliver, 2003; Murdoch, 1996; Smith & VanHouren, 1996; Tröster, Brambring & Beelman, 1991). One way of managing demographic differences across groups is by matching participants on these characteristics. However, this reduces the likelihood of each syndrome sample representing their population. Therefore, both a total group (total sample) and matching approach (matched sample) were adopted in this cross-sectional questionnaire study. A matching approach was particularly useful for non-parametric data where differences between the groups could not be controlled for statistically. In summary, the aims of this study were to: 1) compare the profile of repetitive behaviors across RTS, ASD, DS and FXS, 2) explore the association between repetitive behavior and ASD phenomenology

(communication and social interaction) across groups using the total sample, 3) to explore associations between repetitive behavior and degree of disability across groups using the total sample, and 4) to repeat aim one with matched participants to control for age, degree of disability, verbal ability, and degree of mobility.

Methods

Recruitment

RTS. 202 primary caregivers of individuals with Rubinstein-Tabyi Syndrome were sent an invitation letter and a counter-balanced questionnaire pack through the Rubinstein-Taybi Syndrome UK Support Group. Questionnaires were sent to all families on the RTS UK Support Group database. These families, along with the families recruited for the comparison groups, were recruited as part of a large scale questionnaire study investigating cognitive and behavioral difference in rare genetic syndromes and neurodevelopmental disorders (Arron, Oliver, Berg, Moss & Burbidge, 2011; Berg, Arron, Burbidge, Moss & Oliver, 2007; Burbidge et al., 2010; Moss et al., 2009; Moss et al, 2008; Oliver, Berg, Burbidge, Arron & Moss, 2011). As the participants were recruited as part of an ongoing study investigating cognitive and behavioral difference across a wide range of syndrome groups, nine other syndromes and six other questionnaire measures were excluded from the analyses reported here. Moss et al. (2009) has previously published the data depicting the repetitive behavior profiles for six of these excluded groups and for the FXS group whose data was reanalysed as a comparison group in the current study.

Comparison Group Recruitment. 500 families of individuals with Down syndrome were invited through the Down Syndrome Association, 432 families of individuals with Fragile-X syndrome were invited through the Fragile-X Society, and 1467 families of individuals who were suspected as meeting diagnostic criteria for ASD were invited through eight branches of the National Autistic Society in the London and West Midlands area.

Participants

Overall, 735 participants (28.26%) returned a questionnaire pack. The questionnaire packs were then screened and exclusion criteria applied. Participants were excluded if they did not: confirm on a demographic/background questionnaire that their child had a diagnosis from a

professional of RTS, FXS, DS, or ASD (N = 41); had an additional chromosomal disorder (N = 1); complete over 75% of the questionnaire pack (N = 8); have a child over four years of age (the SCQ was inappropriate for children younger than four) or did not confirm their child's age (N = 19). In addition to the above criteria, SCQ scores on the SCQ were examined for participants recruited via the National Autistic Society to ensure individuals in the ASD group reached cut-off for an ASD diagnosis on this screening tool (SCQ; Rutter, Bailey & Lord, 2003). The latter inclusion criterion was applied to ensure the ASD cut-off group displayed the behavioral characteristics associated with ASD as it was not possible to confirm diagnosis through clinical assessment due to the large sample size and the use of questionnaire methodology. A further twenty-three individuals were excluded because they did not meet the cut-off.

The demographic characteristics of the remaining participants (N = 643) are displayed in table 1 (left hand side). Of these remaining participants 57.5% were diagnosed by a paediatrician, 25.43% by a clinical geneticist, 4.53% by a GP, 4.73% by a psychiatrist, 2.15% by a clinical psychologist, 1.45% by an educational psychologist and 4.2% by another professional.

Insert table 1 about here

Measures

Demographic Questionnaire. The demographic information of the person with a genetic syndrome was collected using the background questionnaire. This included information on age, gender, mobility, verbal ability (more than 30 words/signs), primary/secondary diagnosis of a genetic syndrome, and when and by whom the diagnosis was made.

Wessex Scale (Kushlick, Blunden & Cox, 1973). The Wessex Scale is a short informant rating scale of degree of disability. The items evaluate the social and physical attributes of the individual forming five subscales: self help, continence, mobility, speech and literacy. The measure has modest reliability but it has been as noted to be an effective tool for large scale questionnaire studies (Palmer & Jenkins, 1982).

Repetitive Behavior Questionnaire (RBQ; Moss et al., 2009). The RBQ is a 19 item informant questionnaire that measures discrete, observable repetitive behaviors. Each repetitive behavior is operationally defined and examples of each behavior are provided. Repetitive behaviors form a total score and five scaled subscales: stereotyped behavior, restricted preferences, insistence on sameness, compulsive behavior and repetitive speech. The frequency of the behaviors is scored on a five-point likert scale. Participants engaging in a repetitive behavior once or more than once a day (scoring 3 or 4 on an item) are deemed to be scoring above the clinical cut-off for that behavior. For brevity and
to avoid repetition the clinical-cut off scores and analyses of these scores are not included in this paper; however, they can be obtained at [insert link to electronic supplementary file 1]. Moss et al. (2009) found that the RBQ has good inter-rater reliability, good test-retest reliability at *item level*, and good concurrent and content validity with the repetitive behavior subscale of the Autism Screening Questionnaire (Berument, Rutter, Lord, Pickles & Bailey, 1999).

Social Communication Questionnaire: Lifetime Version (SCQ; Rutter et al., 2003; previously the Autism Screening Questionnaire [1](ASQ); Berument et al., 1999). The SCQ is a 40 item informant screening questionnaire for the presence of Autism Spectrum Disorder in people with intellectual disabilities. A total score and three subscales can be calculated: communication, repetitive and stereotyped patterns of behavior, and social interaction. Individuals who score 15 or above on the SCQ meet the screening cutoff for ASD. Berument et al. (1999) validated the SCQ with 200 children sampled from developmental disorder clinics (sensitivity = 0.85; specificity = 0.75). The total score on the SCQ relates strongly to total score on the Autism Diagnostic Interview - Revised even after age, gender, language ability and performance IQ were taken into account (Lord, Rutter & Couteur, 1994), which suggests the measure has high concurrent validity (Berument et al., 1999).

In the current study a proportional communication subscale was used for the SCQ (Moss, Oliver, Nelson, Richards & Hall, 2013) because the number of nonverbal participants varied

[1] The Autism Screening Questionnaire (ASQ) was used in the FXS group while the SCQ was used for the other groups. Item 20 (social chat) differed between the versions for nonverbal participants so to ensure consistency this item was treated as missing data and prorated for nonverbal participants by computing the mean score for other completed items within the communication subscale. The use of the prorated item did not alter the significance or direction of results.

across the groups. Nonverbal participants cannot score on all items in the standard communication subscale so the proportional subscale avoids group means being artificially lowered. This was calculated by applying a formula to the communication subscale for nonverbal participants (proportional formula = score on communication subscale / 8 x 13).

Data analysis

Total and Matched Groups Analyses. To maximise the sample size and the likelihood of each sample
representing their given population the majority of analyses was conducted with the total sample (N = 643). In addition, to control for the impact of particular demographic variables, a subset of the total sample formed a matched sample (N = 168). The sample was matched for verbal ability (speech score on the Wessex), self-help score, and age. Self-help score was employed as an indicator of degree of disability. Participants were also matched for mobility, although this was only partially successful due to the severity of mobility problems in FXS. The groups were not matched for vision and hearing because it was not possible to match a sufficient number of participants if these variables were taken into account. The demographic characteristics for the matched group are displayed in table 1 (right hand side).

Data analysis strategy. Data obtained from the Repetitive Behavior Questionnaire at subscale and item level violated the assumption of normality (Kolmogorov-Smirnov: $p < .05$). These data could not be transformed so non-parametric analyses of variance were used. The groups were compared at full scale, subscale and item level of the RBQ using Kruskall-Wallis non-parametric analyses of variance and pairwise Mann-Whitney U tests. The RBQ has item level reliability so analyses were carried out for all items for all groups, irrespective of group differences at subscale level. The above analyses were conducted for the total sample and also for the matched sample of participants (matched for age, self-help score and verbal ability across groups).

In addition, for the total sample, Pearson partial correlations were conducted to examine associations between scores on the subscales of the RBQ and the social interaction and communication subscales of the SCQ while controlling for a proxy measure of intellectual disability (self help score from the Wessex Scale). The repetitive and restricted behavior

subscale of the SCQ were not compared to the RBQ due to overlap of items. Pearson partial correlations were deemed appropriate for subscale analyses even though some subscales were non-Gaussian because of a moderate sample size and the absence of skewness greater than 2.0 (Motulsky, 1995; Kendall & Stuart, 1958). The partial correlations would have been less robust at item level. Finally, Spearman Rho correlations were conducted to explore the association between repetitive behavior and intellectual disability (self help score of the Wessex Questionnaire). This analysis was carried out at item level so that the possible association with intellectual disability could be discussed in relation to the item level analysis of the repetitive behavior profiles. Spearman Rho correlations were conducted as opposed to Pearson correlations because at item level several variables were skewed above 2.0, and several over 4.0. These analyses were exploratory so a conservative alpha level of .005 was used throughout in order to minimise the chances of type II error.

Results

Subscale Level Analyses. Analyses were conducted to compare the total and matched samples' scores on subscales and items of the Repetitive Behavior Questionnaire. The results of these analyses for the total and matched groups are displayed in table 2 (left and right side respectively). The total and matched analyses revealed a significant main effect of group for all the subscales and for the verbal and nonverbal full-scale scores (ps <.005).

Insert table 2 about here

In the total and matched group analyses RTS had significantly higher scores than DS on the stereotyped behavior, compulsive behavior, and verbal full scale subscales; however they did not differ from the ASD or FXS groups. In the total group, the RTS and ASD groups fell between the DS and FXS groups on the repetitive speech subscale with significantly higher scores than the DS group but lower scores than the FXS group. This result differed slightly in the matched group because RTS had a significantly lower score than FXS on the repetitive speech subscale, but did not differ from ASD or DS. The RTS group did not differ from any group on the restricted preferences and insistence on sameness subscales in either the matched or total analyses, although the ASD and FXS groups scored more highly on these subscales than the DS group.

Item Level Analysis. In the total group analyses, there were significant differences for 16 out of 19 items ($p < .005$). Significant differences were absent for tidying up, organising objects, and spotless behavior. The results of the post hoc analyses for the total sample are displayed in figure 1 in the format devised by Moss et al. (2009). In this figure a plus sign indicates that the group is scoring significantly higher than another group on an item, whereas a minus sign indicates that the group is scoring significantly lower than another group on that item.

Insert Figure 1 about here

Visual inspection of figure 1 reveals that people with RTS had the most varied profile of repetitive behavior in comparison to DS who had lower levels of repetitive behavior, and ASD and FXS who had heightened levels of repetitive behavior across a wide range of behaviors. RTS had heightened levels of stereotypy, hoarding, restricted conversation, preference to routine, repetitive questions and phrases in comparison to DS. However, RTS had lower levels of restricted conversation, repetitive phrase and echolalia than ASD and FXS, lower levels of adherence to routine and hand stereotypy than FXS, and lower levels of cleaning than ASD. When the analyses were repeated with the matched sample, significant group differences remained for body and hand stereotypy, restricted conversation, preference for routine, repetitive questions, repetitive phrase, and echolalia. These analyses are displayed in table 3. In agreement with the total group analyses RTS had heightened scores on body stereotypy relative to DS, which did not differ from scores for ASD or FXS. RTS had lower levels of restricted conversation and repetitive phrases relative to ASD and FXS. However, RTS were no longer significantly different from DS for repetitive questions but the group did not differ from FXS either. There was a significant difference between FXS and DS indicating that the scores for RTS were between these two groups.

Insert table 3 about here

Association with Autism Spectrum Phenomenology. The association between repetitive behavior and social-communication deficits for the total group are displayed in table 4.

Insert Table 4 here

In RTS, there were no significant associations between the repetitive behavior measured by the RBQ and social-communication deficits measured by the SCQ. There are associations between repetitive behavior and social-communication deficits in ASD, DS and FXS (*p*s < .005). In ASD greater social-communication deficits were associated with compulsive behavior, insistence on sameness and total nonverbal score. Social interaction deficits were associated with compulsive behavior and total score. In FXS social-communication deficits were associated with repetitive language, whereas social interaction deficits were associated with compulsive behavior, insistence on sameness and total nonverbal score. In DS social-communication deficits were associated with stereotypy, repetitive language and total nonverbal score, and social interaction deficits were associated with stereotyped behavior and total nonverbal score.

Ability Level and Repetitive Behavior. Spearman Rho correlations exploring the association between degree of disability (measured by the Wessex Self Help Score) and items from the RBQ are displayed in Table 5. The fewest significant correlations between repetitive behavior and self-help score were for the RTS group. Intellectual disability was associated with object
stereotypy and echolalia.

Insert Table 5about here

In people with ASD, ability level was associated with all topographies of stereotypy, rituals, lining up and attachment to objects, repetitive phrases and echolalia. In DS object stereotypy, body stereotypy, and attachment to objects, repetitive phrase and echolalia were related to ability level. A similar pattern was observed in FXS whereby all topographies of stereotypy, all repetitive speech items, and attachment to people were related to ability level.

Discussion

Previous research has found that specific repetitive behavior profiles can be described for a range of neurodevelopmental disorders, lending support for studying repetitive behavior at a fine-grained level of description (Turner, 1997; Moss et al., 2009). In addition, repetitive behavior has been linked to social-communication deficits, associated with ASD, and ability level (World Health Organisation, 1993; American Psychiatric Association, 1994; McClintock et al., 2003). In this paper we described the repetitive behavior profile of RTS in relation to ASD, DS and FXS. This was the first time the repetitive behavior profile of RTS has been described, and to our knowledge the first time that the repetitive profiles of ASD and DS have been compared to other disorders at a fine-grained level. In addition we examined how the repetitive behavior profile relates to ASD phenomenology and ability level. This was conducted at a total group level and at a matched group level.

A descriptive summary of the key findings is displayed in table 6.

Insert table 6 about here

Analyses were conducted initially with the total sample. The RTS total group had an uneven profile of repetitive behavior, engaging in a wide range of behaviors at a higher rate than DS and at a similar level observed in ASD, for example, heightened repetitive questions. The profile was consistent with that reported in previous literature on RTS that has highlighted body stereotypy and adherence to routines as phenotypic behaviors (Stevens et al., 1990). However, other behaviors such as restricted conversation, repetitive phrase and echolalia were less pronounced in RTS and did not differ significantly from DS. The pattern of results observed in the total group for RTS lends further support to studying repetitive behaviors at a fine grained level of description. In particular, if repetitive vocalisations had been grouped together it would have obscured this pattern in RTS.

The specificity of the RTS profile is of interest when considered in comparison to ASD, FXS and DS generally. The DS group had lower repetitive behavior scores across the majority of RBQ items, while ASD and FXS had heightened generalised scores across the majority of items. Heightened generalised repetitive behavior in ASD is consistent with previous reports of behavior in this disorder (Bodfish, Symons, Parker & Lewis, 2000; Hermelin & O'Conner, 1963; Lord, 1995; Lord & Pickles, 1996; Richler, Bishop, Kleinke & Lord, 2007; Watt,

Welnerby, Barber & Morgan, 2008). However, even within the ASD and FXS profiles there was evidence of specificity, for example, FXS engaged in more repetitive questioning than any other group in all analyses conducted.

Matched group analyses were conducted to examine which of these results would remain significant when age, intellectual disability and verbal ability were accounted for. A proportion of the significant results were lost when the matched sample analyses were conducted. Matching groups has the disadvantage of lowering sample size, thus, it is difficult to be conclude whether changes occurred because the associations in the total group analyses were driven primarily by the demographic variables, or whether they were lost because of less power to detect group differences. However, the key benefit of the matched analyses in this study is that it added validity to the positive results that remained significant.

These analyses revealed that the RTS group had comparable levels of body stereotypy to ASD (significantly more than in DS). The RTS group engaged in restricted conversation and repetitive phrase less frequently than both ASD and FXS, and although scores on the repetitive questioning item were no longer significantly heightened in comparison to DS the scores are not significantly lower than ASD and FXS either. Inspection of the mean score on the repetitive behavior item reveals that RTS have a similar mean on this item to ASD. Taken together these results continue to support the dissociation of repetitive behaviors within groups, along with varying levels of repetitive behavior across groups.

We explored how repetitive behaviors related to social-communication deficits in RTS relative to the comparison groups. Repetitive behavior in RTS was not associated with the social-communication deficits measured by the SCQ. The absence of associations occurred even though individuals with RTS scored highly on specific topographies of repetitive behaviors, in line with scores obtained by the ASD group (body stereotypy, routine and repetitive questions). This is an interesting result as repetitive behavior and socio-communication deficits form the triad of impairments in ASD (World Health Organisation, 1993; American Psychiatric Association, 1994) , and the absence of an association fits with a recent review of the literature that summarised evidence from twin studies and the ASD literature to argue that different sets of genes may underpin the separate components of the triad of impairments (Happe, 2008). It is important to note that an absence of an association between repetitive behavior and socio-communicative deficits in RTS does not negate the

possibility of a unified mechanism underpinning triad of impairments in ASD because these findings do not prove that ASD cannot emerge from one genetic origin that codes for the entire pathway. However, it does point towards the possibility that different genetic components may give rise to different aspects of the triad of impairments. Furthermore, because repetitive behaviors scores in ASD were found to mirror some scores in RTS and FXS, it is possible that the mechanisms that underpin these repetitive behaviors are not distinct to ASD.

It was surprising that the strongest relationships between social-communication deficits and repetitive behavior were found for FXS and DS and not for the ASD group. However, this pattern is likely to be a product of the sampling method used for the ASD group. The ASD group were a more homogenous group because they were selected based on their behavioral characteristics. Hence, in comparison to the ASD group, there may be more individuals obtaining low scores on the RBQ and low scores on SCQ in the contrast groups, and this may have given rise to stronger, more linear associations between these measures.

The finding that RTS has some highly specific heightened repetitive behaviors but that these repetitive behaviors are not related to social-communication deficits suggests that this syndrome might sit between disorders characterised by high repetitive behavior and social-communication deficits (e.g. ASD & FXS) and disorders with low repetitive behavior and fewer social-communication deficits (e.g. DS). The confirmation of anecdotal reports of repetitive behavior in RTS and the finding that these behaviors do not relate to social-communication deficits fits with the anecdotal reports that RTS might have fewer social deficits in comparison to other disorders (Goots & Liemohn, 1977; Baxter & Beer, 1992; Stevens et al., 1990). It is unclear why repetitive questioning may be heightened in RTS relative to other repetitive vocalizations such as echolalia and phrases. As socio-communicative deficits are not associated with repetitive behavior in RTS it may be that repetitive questions may have utility in RTS in the social environment. For example, in RTS repetitive questions may elicit caregiver attention or be related to memory difficulties that have been pointed towards in this syndrome (Alarcon et al., 2004; Wood et al., 2005; Weeber & Sweatt, 2002).

Finally, it was found that some repetitive behaviors, namely, stereotypy, repetitive phrases/signing and echolalia may be partly related to degree of intellectual disability. This is

consistent with previous studies that have explored the impact of ability level (McClintock, Hall & Oliver, 2003; Smith & VanHouren, 1996). However, it is worth noting these behaviors are unlikely be related solely to ability level given the variation found in repetitive behavior in the matched sample. For example, there were significant differences between the matched samples for stereotypy. There is also some variation across syndrome groups in respect to the degree to which repetitive behaviors correlate with ability level (only object stereotypy and echolalia are related to ability level in RTS). Furthermore, a large number of repetitive behaviors do not correlate with age or ability in any of the groups although they relationships are in the anticipated direction.

There are a number of implications of these findings. One potential mechanism that might underpin specific repetitive behavior in RTS and may warrant further investigation is an executive functioning deficit. Turner (1997) found that adherence to routine in individuals with ASD was related to difficulties shifting set on a rule based card game. Woodcock, Oliver and Humphreys (2009a, 2009b, 2009c) conducted a series of studies with individuals with Prader-Willi syndrome to demonstrate a link between set-shifting deficits and resistance to change. They used a broad range of methodologies including a questionnaire study, naturalistic observations, manipulating environmental contingencies and brain scanning to demonstrate this link. There is also evidence that suggests repetitive behaviors emerge in individuals with Alzheimer's patients alongside executive function impairments (Cullen et al. 2005). Given that repetitive behavior profiles can be described at a fine-level of description further investigation should explore how specific executive function deficits might map on to these profiles (Turner, 1997).

Fine-grained description of repetitive behavior has implications for clinical practice. Repetitive behavior profiles provide a description of what is generally 'typical' within a syndrome group and may help highlight potential key areas of need for particular groups, such as an importance of routine. Clinicians may be able to use this information to target interventions towards particular repetitive behaviors, or to predict which behaviors may pose a challenge for families over time. Some families may find it useful to know that a particular behavior, such as repetitive questioning, is common in a genetic disorder. In addition, these profiles may help clinicians differentiate between individuals who are displaying repetitive behavior that is characteristic of a genetic disorder and behavior that is related to an

underlying mental health issue such as the presence of Obsessive Compulsive Disorder, or the presence of a co-morbid neurodevelopmental disorder such as ASD.

The possibility of a potential dissociation of the triad of impairments should influence how ASD phenomenology is studied in the future and more may be learnt about the underpinnings the triad of impairments from cross syndrome comparisons. This conclusion would be strengthened in RTS by an empirical investigation of the social motivation of individuals with RTS syndrome relative to other syndrome groups. To date, extensive research is being conducted into the cognitive underpinnings of the social phenotype of RTS (Powis, Apperly, Waite & Oliver, personal communication; Nelson, 2010) and it is hoped that this research will lend further support to these conclusions.

There are a number of limitations of this study. The first limitation relates to the low response rate from families of individuals with ASD and DS. This might have occurred because families of individuals with these neurodevelopmental disorders are more frequently invited to participate in research studies. A further possibility is that the families that did respond were those with fewer care-giving demands due to supporting individuals with less severe difficulties. If this occurred it could have reduced the differences between FXS and ASD and increased the chances of finding significantly less behavior in DS. This seems unlikely for DS as the results presented in this paper concur with the previous reports of behavior in DS (Hepburn & MacLean, 2009). In addition, it could be argued that the opposite pattern of responding should be expected from families of individuals with ASD in that families with the greatest need are more likely to be motivated to respond.

Additional limitations include relying on the self-help score from the Wessex scale to assess intellectual functioning. The self-help score gives an estimate of adaptive behavior; however, a measure of cognitive non-verbal intelligence may have yielded different associations. Therefore, further studies using direct assessment of intellectual functioning are needed to validate the current findings. In addition, direct assessment of intellectual functioning and child characteristics such as repetitive behavior would help overcome potential bias that arises when one informant reports on multiple aspects of behavior.

A final caveat is that while behavior appears to occur at a similar frequency in ASD and FXS it may be that differences would emerge if severity of the behavior was measured or if the

scale on the RBQ was expanded to allow for more frequent behavior to be reported (i.e. more than once a hour). Despite this there still appears to be a good range of scores on the RBQ, which suggests that the measure is capturing important differences between the groups.

In conclusion, RTS has a varied profile of repetitive behavior and certain behaviors are elevated in this group (e.g. repetitive questioning, body stereotypy). In RTS these repetitive behaviors appear to be unrelated to social-communication, and ability level does not fully explain the profile. This supports the possibility of differing underlying aetiology and developmental trajectories for specific repetitive behaviors that is distinct from the aetiology of other ASD phenomenology. It is too simplistic to argue that all repetitive behaviors decrease with age or that the presence of a repetitive behavior guarantees another. These findings converge with those of Moss et al. (2009) who demonstrated the heterogeneous nature of repetitive behavior profiles and lend support to Turner (1997) who argued that grouping behaviors together serves to mask subtle differences that need to be observed if the underlying mechanisms of these behaviors are to be understood.

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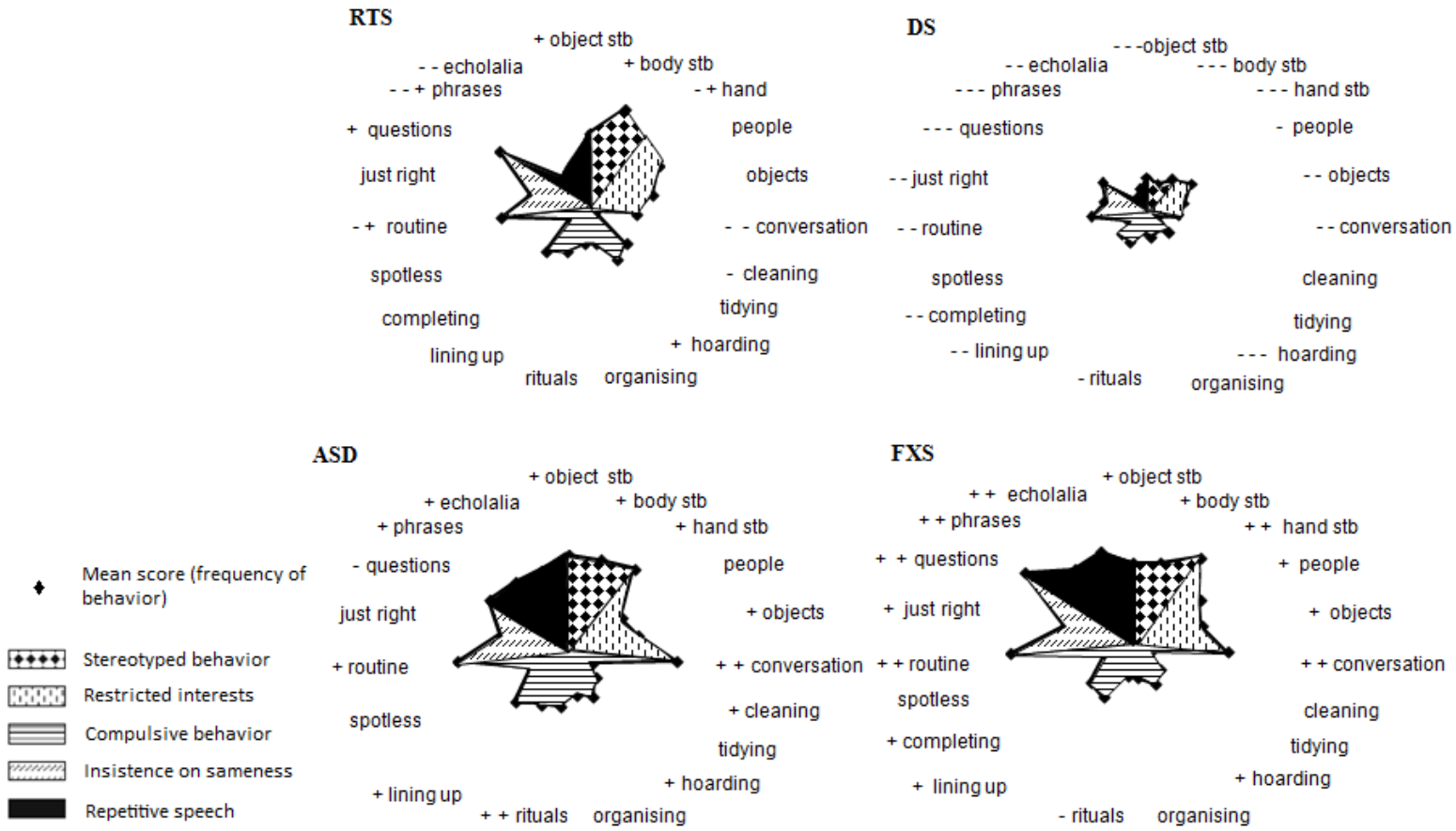
Figure Caption Sheet

Figure. 1 The repetitive behavior profiles of ASD, FXS, RTS and DS at item level. A plus sign indicates that the group had a significantly higher mean score on that item than another group, whereas a minus sign indicates that the group had a significantly lower mean score than another group on that item.

Note. 5 point likert scale: 0 = never, 1 = once a month, 2 = once a week, 3 = once a day, 4 = more than once a day.

Note. Behaviors listed from top clockwise are as follows: object stereotypy, body stereotypy, hand stereotypy, attachment to people, attachment to objects, restricted conversation, cleaning, tidying, hoarding, organising objects, rituals, lining up objects, completing behavior, spotless behavior, adherence to routine, just right behavior, repetitive questions, repetitive phrases/signs, echolalia.


RTS
+ object stb
- - echolalia
+ body stb
- - + phrases
- - + hand
+ questions
people
just right
objects
- - + routine
- - conversation
spotless
- cleaning
completing
tidying
+ hoarding
lining up
rituals
organising
DS
- - -object stb
- - echolalia
- - - body stb
- - - phrases
- - - hand stb
- - - questions
- people
- - just right
- - objects
- - routine
- - conversation
spotless
cleaning
- - completing
tidying
- - lining up
- - - hoarding
- rituals
organising
ASD
+ object stb
+ echolalia
+ body stb
+ phrases
+ hand stb
- questions
people
just right
+ objects
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+ + rituals
organising
FXS
+ object stb
+ + echolalia
+ body stb
+ + phrases
+ + hand stb
+ + questions
+ people
+ just right
+ objects
+ + routine
+ + conversation
spotless
cleaning
+ completing
tidying
+ lining up
+ hoarding
- rituals
organising
Mean score (frequency of behavior)
Stereotyped behavior
Restricted interests
Compulsive behavior
Insistence on sameness
Repetitive speech

Figure 1.

Table 1. Demographic characteristics of the total group and matched samples broken down by syndrome group

		Total Group Analysis								Matched Group Analysis							
		Syndrome group								Syndrome group							
		A ASD	B FXS	C RTS	D DS	*Df*	χ^2/ Kruskal Wallis*	*p* value	Post hoc analyses (<.005)	A ASD	B FXS	C RTS	D DS	χ^2/ Kruskal Wallis*	*p* value	Post hoc analyses (<.005)
N[a]		228	196	87	132					42	42	42	42			
Age[b]	Mean	12.01	17.48	19.98	23.91	3	103.52*	<.001	B,C,D>A D>B	15.55	15.50	15.86	15.90	0.30*	.960	-
	SD	5.78	8.93	11.45	12.61					8.26	7.02	7.06	7.47			
	Range	4.10-45.84	6.30-47.49	4.24-59.41	4.37-62.00					6.61 – 45.84	6.31-34.06	6.60-32.80	4.95-34.63			
Gender	% male	86.0	100[f]	54.0	43.2	3	180.83	<.001	B>A,> C,D	83.3	100	57.1	38.1	44.91	< .001	B>A,C,D A>D
Ability[c]	% able or partly able[d]	89.9	90.8	77.0	93.1	3	16.03	.001	A,B,D>C	90.5	90.5	83.3	95.2	3.34	.342	-
Mobility[c]	% mobile[e]	95.2	72.0	77.9	92.4	3	54.52	<.001	A,D>B,C	90.5	71.4	82.9	88.1	6.49	.090	-
Verbal ability [c]	%verbal	92.5	96.3	84.9	96.2	3	14.55	.002	B,D>C	90.5	95.2	88.1	97.6	3.59	.309	-
Hearing[c]	% normal hearing	96.9	97.4	85.1	65.9	3	101.68	<.001	A,B>C>D	97.6	97.6	78.6	61.9	27.67	< .001	A,B>C,D
Vision[c]	% normal vision	96.5	88.1	85.1	63.4	3	75.66	<.001	A>B,C>D	97.6	88.1	78.6	61.9	19.42	< .001	A> C, D B>D
SCQ	Mean	26.36	21.00	17.15	9.84	3	255.28	<.001	A>B>C>D	27.72	24.11	17.33	11.45	71.81	< .001	A > B > C > D
	SD	5.48	6.79	5.51	7.07					5.44	5.42	5.53	8.27			

Groups: *ASD* Autism Spectrum Disorder, *FXS* Fragile X Syndrome, *RTS* Rubinstein-Taybi Syndrome, *DS* Down Syndrome

[a] *N* may vary across analyses due to missing or incomplete data

[b] in years

[c] information obtained from the Wessex self help scale (Kushlick et al, 1973)

[d] Those scoring 6 or above on the self help subscale. Self help is derived from summing three items regarding independent feeding, washing and dressing. Items are scored between one and three resulting in a total score ranging between three and nine.

[e] defined as scoring 6 on the Wessex mobility subscale

[f] due to the X linked nature of the disorder 100% of FXS participants were male.

Note. FXS data previously presented in Moss et al. (2009). FXS group contains five additional participants who were added to dataset.

Table 2.
Total Group Analyses. Mean score, standard deviation, statistical analyses and post hoc analyses at subscale and full scale level of the RBQ.

	Total Group (N = 643)								Matched Group (N = 162)						
	A ASD	B FRX	C RTS	D DS	df	χ^2	*p* value	Post hoc analyses	A ASD	B FRX	C RTS	D DS	χ^2	*p* value	Post hoc analyses
	Mean (SD)								Mean (SD)						
Stereotyped behavior	6.56 (4.14)	6.47 (4.10)	6.21 (4.27)	2.39 (3.63)	3	96.85	< .001	ABC>D	7.42 (4.21)	6.95 (4.17)	5.97 (4.20)	3.07 (3.98)	24.04	<.001	ABC> D
Compulsive behavior	8.42 (7.73)	7.03 (6.93)	7.11 (6.48)	4.29 (6.22)	3	37.53	< .001	ABC>D	8.07 (6.70)	7.99 (7.25)	7.26 (6.48)	3.39 (4.95)	16.82	=.001	ABC> D
Restricted preferences[a]	5.23 (3.63)	5.51 (3.71)	4.57 (3.45)	2.76 (3.02)	3	50.88	< .001	AB>D	5.91 (3.67)	5.51 (3.71)	4.48 (3.20)	3.28 (3.29)	14.51	=.002	AB > D
Insistence on sameness	3.96 (2.79)	4.3 (2.73)	3.46 (3.07)	2.29 (2.78)	3	43.85	< .001	AB>D	4.49 (2.78)	4.49 (2.67)	3.30 (3.23)	1.88 (2.63)	21.05	<.001	AB > D
Repetitive speech	5.96 (4.00)	7.14 (3.67)	4.67 (3.69)	2.03 (2.84)	3	121.68	< .001	B>AC>D	6.47 (4.49)	7.58 (3.21)	4.81 (3.45)	2.76 (2.72)	30.57	<.001	AB > D B > C
Verbal total score[a]	29.33 (17.00)	30.08 (15.50)	26.01 (15.56)	13.25 (14.46) 9.88	3	94.82	< .001	ABC>D	32.09 (15.91)	31.35 (14.52)	25.87 (15.09)	13.87 (13.87)	32.14	<.001	AB C>D
Nonverbal total score[b]	22.41 (13.64)	21.80 (12.72)	19.34 (11.82)	(11.51)	3	93.03	< .001	ABC > D	23.84 (12.80)	23.44 (13.17)	18.85 (12.14)	9.78 (10.47)	32.69	<.001	ABC> D

[a] Analysis only includes participants who are verbal

[b] Score calculated using nonverbal items for all participants

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

Note. A letter missing from the post hoc analyses column indicates that this group was not different from other groups.

Note. FXS data previously presented in Moss et al. (2009). FXS group contains five additional participants who were added to dataset.

Table 3 *Matched Sample Analyses. Mean score, standard deviation, statistical analyses and post hoc analyses at item level of the RBQ.*

Group:	A ASD	B FXS	C RTS	D DS	χ^2	*p* value	Post hoc
Stereotyped behavior							
Q1 Object stereotypy	2.54 (1.67)	1.86 (1.72)	1.67 (1.78)	1.24 (1.72)	11.07	ns	
Q2 Body Stereotypy	2.24 (1.78)	2.29 (1.72)	2.45 (1.88)	0.86 (1.54)	20.55	<.001	A B C > D
Q3 Hand stereotypy	2.64 (1.69)	2.81 (1.58)	1.86 (1.88)	0.98 (1.60)	25.85	<.001	A B > D
Compulsive behavior							
Q4 Cleaning	0.64 (1.36)	0.62 (1.27)	.02 (.15)	0.28 (0.97)	10.55	ns	
Q5 Tidying	0.81 (1.31)	0.88 (1.29)	1.05 (1.48)	.43 (1.13)	6.45	ns	
Q6 Hoarding	0.88 (1.45)	0.79 (1.39)	1.21 (1.68)	.31 (.98)	9.61	ns	
Q7 Organising objects	0.81 (1.38)	0.86 (1.44)	0.93 (1.49)	.60 (1.23)	1.12	ns	
Q12 Rituals	1.26 (1.70)	0.81 (1.55)	0.86 (1.52)	.39 (1.07)	7.40	ns	
Q16 Lining up objects	1.21 (1.65)	1.42 (1.70)	1.43 (1.70)	.78 (1.35)	4.26	ns	
Q18 Completing behavior	1.60 (1.67)	1.60 (1.75)	1.36 (1.68)	.68 (1.39)	8.34	ns	
Q19 Spotless behavior	0.86 (1.49)	1.02 (1.63)	.40 (1.04)	.27 (.90)	7.54	ns	
Restricted preferences							
Q8 Attachment to people[a]	1.41 (1.73)	1.82 (1.63)	1.77 (1.56)	1.38 (1.57)	2.06	ns	
Q10 Attachment to objects	1.73 (1.87)	1.51 (1.81)	1.48 (1.80)	1.19 (1.63)	2.20	ns	
Q13 Restricted conversation[a]	2.71 (1.61)	2.64 (1.60)	1.23 (1.65)	0.79 (1.42)	30.07	< .001	A B > C D
Insistence on sameness							
Q15 Preference for routine	2.74 (1.62)	2.78 (1.59)	1.97 (1.80)	1.05 (1.56)	23.70	< .001	A B > D
Q17 Just right behavior	1.68 (1.66)	1.67 (1.56)	1.33 (1.76)	0.83 (1.39)	8.27	ns	
Repetitive Speech							
Q9 Repetitive questions[a]	2.44 (1.76)	3.27 (1.23)	2.61 (1.70)	1.87 (1.67)	12.33	= .006[b]	B > D
Q11 Repetitive phrases/signing	2.02 (1.79)	2.43 (1.71)	0.58 (1.16)	0.24 (0.89)	48.09	< .001	A B > C D
Q14 Echolalia[a]	1.88 (1.74)	2.12 (1.62)	1.55 (1.80)	0.68 (1.32)	15.12	< .005	A B > D

[a] Analysis only includes participants who are verbal.

[b] Differences for the repetitive question item were approaching significance at .006. Due to the exploratory nature of the analysis, post hoc analyses were performed.

Table 4

Pearson's partial correlations between subscales of the RBQ and the communication and social interaction subscales of the SCQ for total groups (ASD, FXS, RTS & DS). Exploring the relationship between repetitive behavior and Autistic Phenomenology (controlling for self help score)

Group	Subscales of the Social Communication Questionnaire (SCQ)	RBQ: Stereotyped Behavior Subscale	RBQ: Compulsive Behavior Subscale	RBQ: Restricted Preferences Subscale	RBQ: Insistence on Sameness Subscale	RBQ: Repetitive Use of Language Subscale	RBQ Total Nonverbal Score
ASD	SCQ: Communication Subscale	.16	.19**	.11	.20**	.17	.23***
	SCQ: Social Interaction Subscale	.11	.19**	.13	.17	.08	.21**
FXS	SCQ: Communication Subscale	.14	.17	.13	.17	.25**[a]	.21*[b]
	SCQ: Social Interaction Subscale	.20*[c]	.28***	.20	.32***	.21*[d]	.33***
RTS	SCQ: Communication Subscale	.14	-.02	.15	-.10	.27	.00
	SCQ: Social Interaction Subscale	.08	-.10	.17	-.05	-.05	.00
DS	SCQ: Communication Subscale	.53***	.27*[e]	.28*[f]	.23	.34**[g]	.42***
	SCQ: Social Interaction Subscale	.51***	.21	.14	.15	.16	.35***

*** Significant at < .001, ** significant at <.005, * significant at <.01

[abcdefg] = .001, .005, .007, .007, .005, .006, & .001 respectively

Table 5
Total Group Analyses. Spearman's correlations between degree of intellectual disability and repetitive behavior at item level of the RBQ.

	Group			
	ASD	FXS	RTS	DS
Stereotyped behavior				
Q1 Object stereotypy	-.37***	-.33***	-.35**	-.40***
Q2 Body Stereotypy	-.41***	-.38***	-.12	-.32***
Q3 Hand stereotypy	-41***	-.41***	-.26	-.23*[a]
Compulsive behavior				
Q4 Cleaning	.12	-.08	.13	.18
Q5 Tidying	-.09	-.06	-.10	-.02
Q6 Hoarding	-.03	-.02	.13	.08
Q7 Organising objects	-.12	-.05	-.01	.05
Q12 Rituals	-.22**[b]	-.14	-.00	-.08
Q16 Lining up objects	-.22**[c]	-.10	-.14	.01
Q18 Completing behavior	-.12	-.07	-.14	-.09
Q19 Spotless behavior	-.13	.09	-.02	.04
Restricted preferences				
Q8 Attachment to people[a]	-.08	-.25**	.16	-.08
Q10 Attachment to objects	-.24***	-.19*[d]	-.02	-.28**
Q13 Restricted conversation[a]	-.03	-.06	-.12	-.07
Insistence on sameness				
Q15 Preference for routine	-.10	-.12	-.04	.10
Q17 Just right behavior	-.17	-.06	-.06	-.04
Repetitive Speech				
Q9 Repetitive questions	-.13	-.23**	-.16	-.24*[e]
Q 11 Repetitive phrases/signing	-.22**[f]	-.27***	-.14	-.31***
Q 14 Echolalia	-.31***	-.29***	-.36**	-.28**

*** Significant at <.001** Significant at <.005
* Significant at < .01
[a b c d e f] Significant at .008, .001, .001, .009, .008 . & .001 respectively

Table 6
Descriptive summary of the key findings for ASD, FXS, RTS & DS

	Group			
	ASD	FXS	RTS	DS
Degree of repetitive behavior	High	High	Moderate	Low
Degree of social/communication deficit (SCQ Score)	High	High	Moderate	Low
Relationship between repetitive behavior and social/communication deficits	Present	Present	Absent	Present