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Investigation of adaptive and maladaptive behaviour in people with Wolf Hirschhorn Syndrome

Peter McGill and Paul Langthorne



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Summary

Methods

Wolf Hirschhorn syndrome (WHS) is caused by a deletion to chromosome 4p16. A number of studies have described the physical characteristics associated with the syndrome; however, with only a handful of exceptions, the behavioural profile of WHS has to date escaped systematic attention. The current study aimed to provide an overview of the behavioural profile associated with WHS.

Information was collected by survey and interview on 49 people with a reported diagnosis of WHS (aged 2-37 years). Interviews were conducted over the telephone with parents/caregivers and included questions examining: the form, severity and communicative function of any challenging behaviours displayed by the person with WHS; social impairment (i.e. symptoms of autistic spectrum disorder (ASD)); and levels of adaptive behaviour across communication, daily living skills, socialization and motor skill domains. Questions were also asked regarding family experiences of the support provided by local services.

Findings

Adaptive behaviour

- People with WHS, generally, have severe deficits in adaptive behaviour. The average developmental age equivalent was just over 2 years and the majority of the sample scored in the profoundly intellectually disabled range;
- Deficits in adaptive behaviour were, on average, more severe than those of comparison groups consisting of people with Fragile X Syndrome, Smith-Magenis Syndrome and for people with intellectual disabilities of a mixed etiology;

- A minority of the sample presented much less severe deficits in adaptive behaviour more consistent with a moderate or mild intellectual disability;
- The extent of variability in adaptive behaviour was greater in the sample of people with WHS than in the comparison groups. That is, in a number of areas the WHS group included both the least able and the most able of all those for whom scores were available;
- Significant within-group differences were found in the profile of scores for the WHS group. Scores on the socialization subscale were significantly higher than other adaptive behaviour domains.

Social Impairment

- Detecting the presence of ASD in people with WHS is more difficult with respect to the more disabled individuals. At a minimum, it appeared that close to a third of participants met criteria for possible ASD including a smaller number meeting the criteria for Autism itself.

Challenging behaviour

- On average people with WHS were reported to display less, and less serious, challenging behaviour than people with Prader-Willi, Smith-Magenis and Cri du Chat Syndromes;
- There was considerable variability in scores relating to challenging behaviour. Almost a third sometimes displayed aggressive behaviour and over a fifth sometimes behaved destructively;
- Nearly half of the sample displayed at least 1 form of self-injury (SIB). The most common topographies were teeth grinding, self-biting and head banging;
- In relation to behavioural function, significant within-group differences were found for all three forms of challenging behaviour. It appeared that people with WHS presented with relatively high levels of attention-maintained challenging

behaviours and relatively low levels of challenging behaviour maintained by other functions such as escaping from demands or gaining access to preferred objects or activities;

- Physical discomfort/pain may be a relatively frequent contributing factor to the self-injurious and aggressive behaviour of people with WHS;
- Overall, challenging behaviour was more likely to be reported in people with WHS who had lower levels of adaptive behaviour and higher levels of social impairment;
- Those displaying SIB were substantially more disabled with poorer daily living, socialization and motor skills and higher levels of social impairment than those who did not display SIB.

Family support

- The most commonly provided form of support was ‘other’ (e.g., physiotherapy, occupational therapy). The least common type of support came from psychologists;
- The most ‘helpful’ source of support was ‘other’, the least helpful was psychology;
- Families reported relatively high levels of satisfaction with all kinds of support other than psychological input though a number of families reported having had to fight to obtain support.

The findings of the current study are compared with those from other research, their limitations considered and their possible implications for people with WHS and their families described. Suggestions for future research are made.

Introduction

Wolf Hirschhorn syndrome (WHS), first described in 1961 by Cooper and Hirschhorn, is caused by a deletion to part of chromosome 4 (4p16.3). The syndrome is a contiguous genetic syndrome (involving multiple genes) and has an estimated incidence of 1/50,000-1/20,000 births, occurring more commonly in females than males, with a predilection of 2:1 (Battaglia, Filippi, & Carey, 2008). WHS is a recognised cause of intellectual and developmental disability.

Several clinical descriptions now exist suggesting a highly characteristic physical phenotype associated with the syndrome. Battaglia et al (2008), for example, note that of a sample of 87 patients with WHS all had a distinctive “Greek warrior helmet” craniofacial appearance¹. The authors also note that over 80% of the sample had pre-natal onset growth deficiency followed by short stature and slow weight gain; over 75% were reported to have had feeding difficulties (50% of which required a gastrostomy). A number of enduring health issues also appear to be related to WHS. For example, Battaglia et al (2008) note that 50% of their sample suffered from congenital heart defects, some 33% of the sample suffered from urinary tract defects, 40% had hearing loss and some 60% of their sample had skeletal anomalies. Seizures also appear to be common in WHS, and represent a major health issue (Battaglia, Carey, & Wright, Updated 2009; Worthington, Rigby, & Quarrell, 2008).

Despite having been first described over forty years ago relatively little is known about the behavioural profile of WHS. Only a handful of studies have explicitly examined cognitive and behavioural aspects of the syndrome (e.g., Fisch, Battaglia, Parrini, Youngbloom, & Simensen, 2008; Sabbadini, Bombardi, Carlesimo,

¹ Interestingly, Battaglia et al note that these physical features were less marked in children with smaller deletion sizes on chromosome 4.

Rosato, & Pierro, 2002). In their study, involving 11 people with WHS (ages 7-20 years), Sabbadini et al reported that 7 participants were reported to have 'behavioural disorders' that affected participation in learning or play. In regards to adaptive behaviours, Sabbadini et al noted considerable within-syndrome variability but despite this the motor skills of individuals with WHS in their sample were stronger relative to communication skills or daily living skills (as measured by a bespoke questionnaire). A recent paper by Fisch et al (2008) presented preliminary data for 12 children with WHS (ages 4-17 years). Using the Childhood Autism Rating Scale only 1 child met criteria for autistic spectrum disorder (ASD). Using the Child Behavior Checklist as a measure of maladaptive behaviour, 5/12 children were reported to have social problems, 4/12 attention problems, and 1/12 thought problems. In relation to adaptive behaviour, Fisch et al noted significantly higher scores on the *Socialization* subscale of the Vineland than on the *Communication* or *Daily Living Skills* subdomains. The children sampled in the Fisch et al study all had expressive language, so the extent to which the findings generalised to people with WHS without expressive communication skills is unknown.

In the current study we aimed to extend this line of work by examining adaptive and challenging behaviours in a sample of 49 people with WHS. Caregivers rated the severity of the person's challenging behaviour, and, where relevant, data were also gathered on the form of any self-injurious behaviours and the context in which challenging behaviours tended to occur. A screening tool for ASD was also employed. Adaptive behaviour profiles were measured allowing for a direct comparison of the findings with Fisch et al (2008). Caregivers were also asked questions regarding their experience of different types of support from local services.

Methodology

Participants

A total of 49 participants took part in the study. All participants were parents or caregivers of people with a reported diagnosis of WHS (ages ranged from 2 to 37 years of age).

Participants were recruited from the Wolf Hirschhorn Syndrome Trust's (WHST) mailing list. Potential participants were initially sent a letter of support from the WHST. Information packs about the study were then sent to a total of 133 people across three separate mail outs. Reminder letters were sent to all people who did not respond within 3 weeks of the initial mail out. This led to a total of 49 people returning consent forms, a response rate of 36.8%. In some cases it proved difficult to arrange interviews so that not all measures were completed for the whole sample. Characteristics of the 49 people with WHS are presented in Table 1 below.

Table 1 Characteristics of 49 children and adults with WHS

Chronological Age	
Mean (minimum to maximum)	17 years 6 months (2y3m-37y3m)
Gender	
Male	24
Female	25

Measurement Instruments

A range of measures were used as part of the current study. The following measures were used to measure the severity, form, and function of challenging behaviour respectively: the *Aberrant Behavior Checklist-Community* version (ABC-C; Aman, Borrow, & Wolford, 1995), the *Self-Injurious Behavior* subscale of the

Behavior Problems Inventory (Rojahn, Matson, Lott, Esbensen, & Smalls, 2001) and the *Questions About Behavioral Function* scale (QABF; Matson & Vollmer, 1995). The *Social Communication Questionnaire* (M. Rutter, Bailey, & Lord, 2003) was used to measure ASD symptomology. The *Vineland Screener* (Sparrow, Carter, & Cicchetti, 1993) was used to measure profiles of adaptive behaviour. Finally a measure of the support experienced by families from local services was completed (McGill, Papachristoforou, & Cooper, 2006).

The *Aberrant Behavior Checklist-Community (ABC-C)* was designed to measure the severity of challenging behaviour displayed by individuals in community settings. The *ABC-C* consists of 58 items, each scored on a 4-point scale (0: not a problem, through to 3: problem is severe in degree). The items fall into five subscales: (1) *Irritability, Agitation, Crying* (15 items), (2) *Lethargy, Social Withdrawal* (16 items), (3) *Stereotypic Behavior* (7 items), (4) *Hyperactivity, Non-Compliance* (16 items), and (5) *Inappropriate Speech* (4 items). The *ABC-C* is a reliable and valid behaviour rating instrument (Rojahn, Aman, Matson, & Mayville, 2003).

The *Questions About Behavioral Function* scale (*QABF*) is a 25-item questionnaire designed to identify the variables that maintain challenging behaviour. The *QABF* is scored on a 4 point Likert scale from never (0) to often (3). If an item does not apply then the N/A option may be endorsed. Its five subscales (*escape, attention, non-social, tangible, physical discomfort*) have been confirmed via factor analysis and the scale has good reliability (Paclawskyj, Matson, Rush, Smalls, & Vollmer, 2000).

The *Self-Injurious Behavior* subscale of the *Behavior Problems Inventory (BPI)* was used where respondents indicated that the person displayed self-injurious behaviour. The *BPI* has been found to be a reasonably reliable and valid measure

(Rojahn, et al., 2001). The Self-Injurious Behavior subscale consists of 14 items which are rated on a 7 point frequency scale (never to more than hourly).

The *Social Communication Questionnaire (SCQ)*, formerly known as the *Autism Screening Questionnaire*, was used to examine the presence of ASD symptomology. The total score can range from 0 to 39. The *SCQ* may be used to screen for the likelihood that an individual has an ASD and to measure the severity of autistic symptoms in comparison to other groups. *SCQ* scores have good reliability, correlate highly with established measures of autism and discriminate well between ASD and other disorders (Howlin & Karpf, 2004; M. Rutter, et al., 2003).

The *Vineland Screener (VSC)* is a tool used to assess domains of adaptive behaviour derived from the full Vineland Scale and is intended for research purposes only. The *VSC* consists of 15 items in each of the following three domains: *Communication, Daily Living Skills, and Socialization*. There are also a smaller number of items for the *Motor Skills* domain. Respondents are asked several probe questions to explore their perception of the child's ability for each item. Items are scored on a Likert-type scale from 0-2; an N can also be scored if the individual does not have the opportunity to engage in certain behaviours. Age equivalent scores for each sub-domain can be derived from the Vineland Screener. A mean age equivalent score across the Communication, Daily Living Skills and Socialization subdomains has been used in prior research to provide a global measure of intellectual functioning (e.g., Chadwick, Kusel, Cuddy, & Taylor, 2005; Charman, Howlin, Berry, & Prince, 2004). Sparrow et al (1993) report that the *VSC* has high inter-rater reliability and each domain has high convergent validity with the respective domains on the full Vineland.

The *Family Support Questionnaire* includes questions on the extent and quality of support received by parents and has been used in previous studies (McGill, et al., 2006). The questionnaire consists of 11 items scored on Likert-type scales and respondents are asked to indicate which supports they have accessed in the past and their satisfaction with each.

Procedure

All parents/caregivers were provided with an information sheet describing the aims of the study, a consent form, a copy of the *ABC-C* together with a pre-paid envelope. Each participant also received a covering letter from the WHST detailing their support for the research.

All participants were asked to return a completed copy of the *ABC-C* and a completed consent form indicating a convenient time for the interviews to be conducted. Participants were then sent a confirmation of the scheduled interview time together with copies of the remaining measures that were to be completed over the telephone. Parents were advised to have the questionnaires to hand for the telephone interview.

All telephone calls began with the researcher introducing himself and establishing whether this was a convenient time for the interview. If necessary, interviews were rearranged for an alternative time. All interviews began with a brief description of the aims and structure of the interview. At the end of each measure, participants were offered the opportunity to take a break and resume the interview at a later date. For participants who had indicated that the person displayed self-injurious behaviour the *Self-Injurious Behavior subscale* of the *BPI* was completed to gain more detailed information on the form of self-injury displayed by each person. The *QABF* was then completed for each general topographical category of behaviour that

had been rated as problematic in the *ABC-C* (aggression, self-injury, destructive behaviour). The *SCQ* was then completed followed by the *VSC*. The *VSC* consists of several age ranges (0-2yrs, 3-5yrs, 6-12yrs, and 13-18yrs). All participants were assigned to the age range immediately beneath the person's chronological age. For example, if the person was 15 years old then the respondent was asked to complete the 6-12 yrs old age range. The appropriateness of this age range was then assessed, using the following protocol. If the participant achieved a score of 9 or 10 (max score = 10) after the first five items or 18 to 20 (max score = 20) after the first 10 items then a higher age range was selected. Conversely if the participant achieved scores of 0 to 1 after five items (min score = 0) or 2 or less after 10 items (min score = 0) then a lower age-range was selected. This process was repeated for each sub domain until the assessment was completed.

Results

All results were compared for male vs female participants. As no significant differences were found, all results are presented for males and females combined.

Adaptive Behaviour

Adaptive behaviour profiles for participants with WHS were examined using the Vineland Screener for a total of 44 participants. Standard scores from the full Vineland manual were derived from Vineland Screener equated raw scores. These standard scores provide an indication of developmental level with typical developmental level being 100 (as with IQ) and are shown in Table 2. Table 3 presents age equivalent scores for the sample in comparison with other syndrome groups.

Table 2 Vineland Standard Scores (where 100 would be average development)

	Mean	Range
Overall Mean	24.3	<20 - 62
Communication Skills	23.0	<20 - 59
Daily Living Skills	22.5	<20 - 91
Socialization	31.8	<20 - 84
Motor Skills	33.2	<20 - 102

Table 3 Vineland Age Equivalent Scores (in months²)

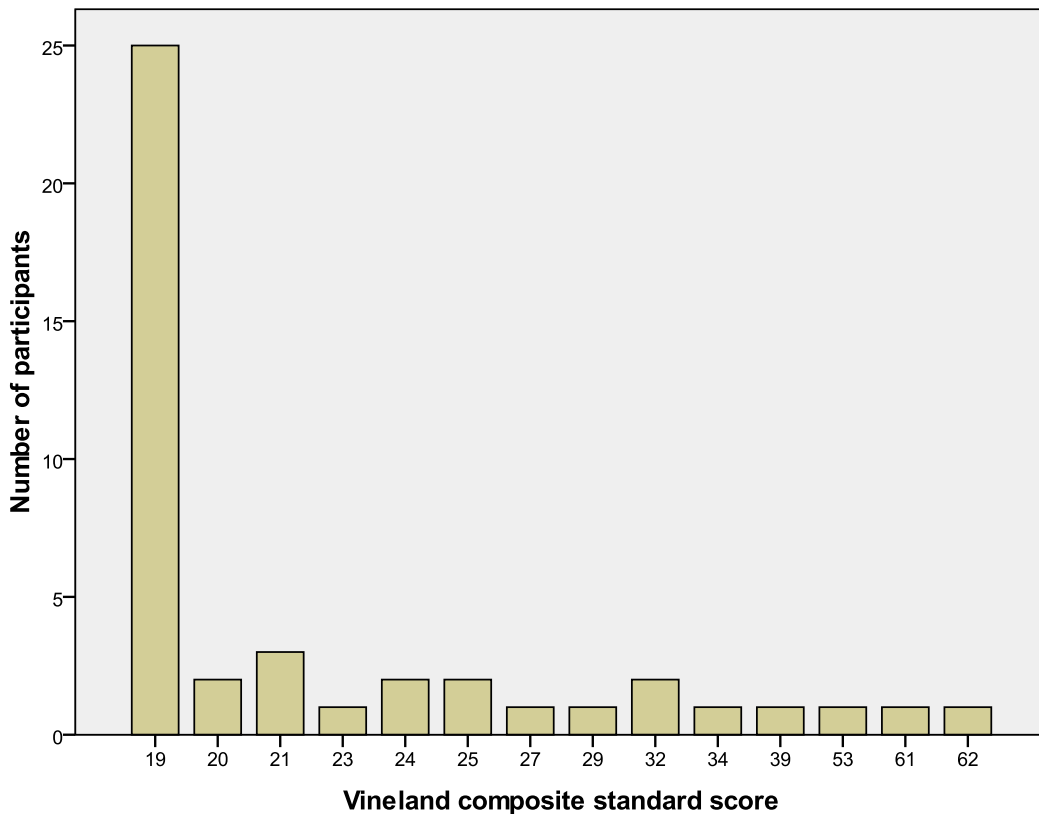
	WHS	FXS	SMS	Mixed
	N = 44	N = 35	N = 24	N = 30
	Mean (Range)	Mean (Range)	Mean (Range)	Mean (Range)
Overall Mean	24.8 (1-149)	46.9 (10-118)	53.9 (19-102)	42.3 (13-96)
Communication Skills	21.7 (0-103)	50.6 (11-189)	65.3 (15-135)	40.4 (12-95)
Daily Living Skills	21.3 (2-198)	42.1 (12-105)	48.7 (18-89)	42.8 (11-124)
Socialization	31.4 (0-147)	48.0 (6-81)	47.8 (11-90)	43.8 (6-83)
Motor Skills	17.5 (0-63)	49.9 (20-71)	56.9 (23-71)	48.7 (8-71)
Age (years)	17.5 (2-37)	11.1 (5-16)	11.2 (5-21)	10.1 (5-15)

A number of observations arise from the above Tables. First, it is clear that the WHS sample is severely disabled with an overall mean standard score of 24.3 in the severe

² Comparison data are taken from previously conducted work by Paul Langthorne and Peter McGill with participants who had Fragile X Syndrome (FXS), Smith-Magenis Syndrome (SMS) or learning disability of mixed etiology (Mixed). Groups from this study exclusively included individuals who displayed challenging behaviour.

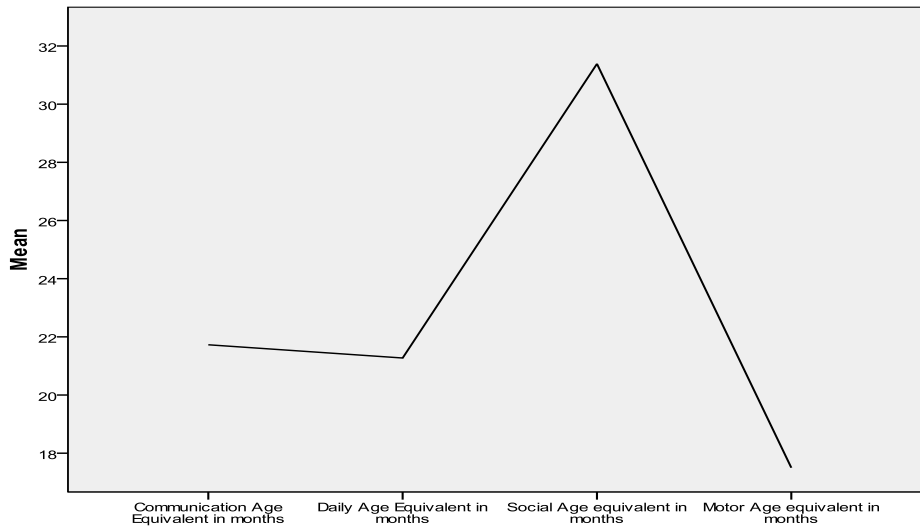
learning disability range. Second, it is clear that, the sample is highly variable. Figure 1 illustrates this more clearly – 25 participants had a mean score of less than 20 (in the profoundly disabled range), 15 in the 20-34 range (severely disabled), 1 in the 35-50 range (moderately disabled) and 3 over 50 (mildly disabled).

Figure 1 Vineland Composite Standard Scores



Third, the sample is generally more severely learning disabled than the comparison samples, especially when it is considered that the comparison samples were significantly younger. Fourth, within the WHS sample socialization skills were relatively higher (see Figure 2). In fact, socialization skills were highest in over half of the sample's individual skills profiles.

Figure 2 Profile of Vineland Subscale Scores.



Within-group analysis of variance (ANOVA) revealed significant differences in the profile of Vineland scores ($F(3, 129) = 9.056; p = 0.00$). Further statistical comparisons between subscales showed that Socialization subscale scores were significantly higher than all other subscale scores ($p < 0.05$).

Challenging Behaviour

Aberrant Behavior Checklist.

Table 4 shows descriptive statistics for the overall and sub domain scores of the ABC-C. Comparison data is from Clarke and Boer's (1998) between-syndrome study of people with Prader-Willi syndrome (mean age = 25.1, range = 6-43.5), Smith-Magenis syndrome (mean age = 14.5, range = 5.5 – 32.5) and Cri Du Chat syndrome (mean age = 17.3, range = 5-40).

Table 4 Aberrant Behavior Checklist Scores

ABC-C	WHS (N = 49)	PWS (N=55)	SMS (N=21)	CdC (N =38)
	Mean	Mean	Mean	Mean
Overall Score	32.2	34.7	68.4	48.1
Irritability	8.6	14.6	21.7	13.2
Lethargy	6.0	6.7	7.5	5.2
Stereotypical	5.3	1.6	7.0	5.3
Hyperactivity	10.5	7.6	26.1	21.7
Inappropriate Speech	2.0	4.2	6.1	2.4

There was a wide spread of scores on the ABC with approximately 50% having scores of 30 or less and 50% over 30. Comparison with the other syndrome groups shown suggests that people with WHS display less challenging behaviour overall with the exception of stereotypical behaviour and lethargy where levels are comparable. As a result of completion of the ABC and discussion during the telephone interview 27 participants were identified who displayed one or more of self-injury (SIB), aggression and destructiveness:

- 21 displayed SIB including 11 who also displayed aggression and/or destructiveness;
- 14 displayed aggression including 10 who also displayed SIB and/or destructiveness;
- 10 displayed destructiveness. In all but one they also displayed SIB and/or aggression;
- 12 displayed at least two of the three behaviours of whom 6 displayed all three.

Behaviour Problems Inventory - SIB subscale

The BPI was completed for all participants who were reported to display SIB. 21 participants with WHS (48% of those interviewed) were reported to display some form of self-injurious behaviour. In only 10 of these participants was self-injury noted in responses to the ABC.

The percentage of endorsement of topographies of SIB in rank order were: teeth grinding (86% of those who displayed SIB); self-biting (43%); hitting head against object (38%); hitting body with object (33%); self-punching (29%); self-scratching (24%); hitting body (19%); stuffing fingers in body openings (14%); head hitting (10%), mouthing or pica (10%); hair pulling (10%); object stuffing (5%); extreme drinking (5%); other (5%); air swallowing (0%); self-induced vomiting (0%).

The frequency of teeth-grinding was particularly notable. Of the 18 participants who engaged in it, it was a daily or more frequent occurrence for 14. Dental abnormalities have been reported in more than 50% of people with WHS (Battaglia et al, 2006) and these may well be related to teeth-grinding.

Challenging Behaviour - Function

Table 5 shows descriptive statistics depicting the profile of scores on the QABF for participants across self-injurious behaviour, aggression and destructive behaviours.

Table 5 QABF Scores

QABF Total Scores	Self-Injury (N=21)		Aggression (N=14)		Property Destruction (N=10)	
	Mean	Range	Mean	Range	Mean	Range
Attention Subscale	6.5	0-14	7.5	0-15	11.0	2-15
Tangible Subscale	3.5	0-15	5.9	0-13	2.9	0-11
Demand Subscale	3.8	0-15	2.9	0-9	1.6	0-6
Physical discomfort Subscale	8.4	0-15	6.3	0-15	1.4	0-12
Automatic Subscale	6.0	0-15	0.9	0-6	6.2	0-15

A within-group ANOVA was conducted for each topographical class of challenging behaviour. Significant differences were found for SIB ($F(4, 80) = 3.80$; $p = 0.07$), aggression ($F(4, 52) = 5.65$; $p = 0.01$) and property destruction ($F(4, 36) = 9.32$; $p = 0.00$).

Post hoc Bonferroni comparisons revealed no pairwise differences for SIB, attention & tangible > automatic for aggression, attention > demand and physical discomfort for property destruction.

The small numbers involved make it difficult to reach definitive conclusions. A number of observations, however, can be made about these findings:

1. Physical discomfort/pain may be relatively frequent contributors to self-injury and aggression. The possible connection noted above between high levels of teeth-grinding and dental abnormalities may be seen in this light though a number of other sources of discomfort/pain (related to other forms of SIB) were also mentioned in interviews, such as sleep disturbance, colds and allergies.

2. There is evidence across all forms of challenging behaviour that the attention of others is an important motivator for people with WHS. That is, a common function of challenging behaviour may be to help obtain the attention of others.
3. There is no evidence that escape from demand acts as a common function of challenging behaviour.
4. As illustrated in Figures 3-7 the pattern of scores may be compared with those patterns found in previous work with people with FXS/SMS. Like people with SMS (but not people with FXS), the challenging behaviours of people with WHS appear to be often attention-maintained and related to physical discomfort. Unlike both groups their challenging behaviours appear to be less frequently maintained by escape from demands or access to tangibles.

Figure 3 QABF attention scores across syndromes

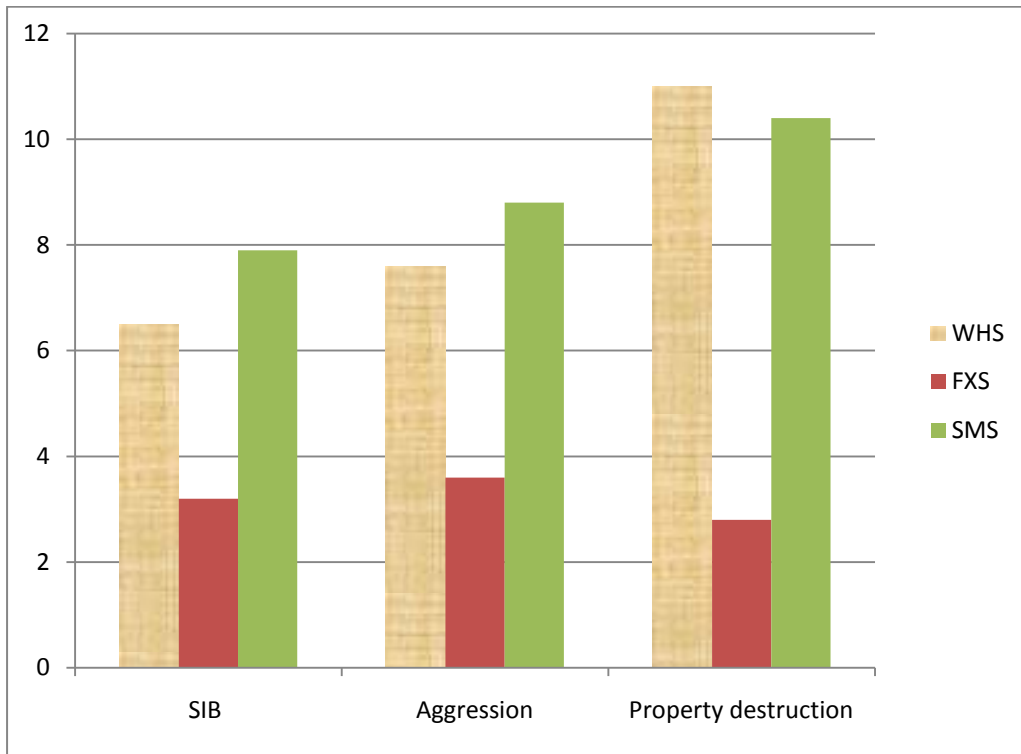


Figure 4 QABF tangible scores across syndromes

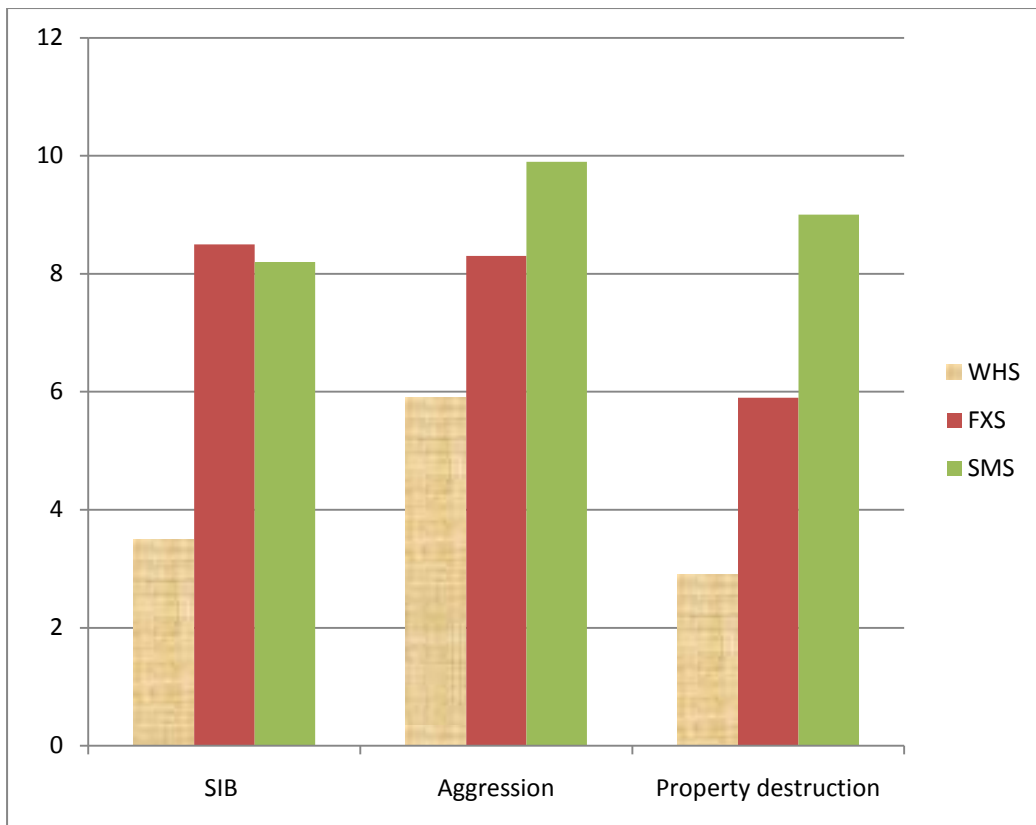


Figure 5 QABF demand scores across syndromes

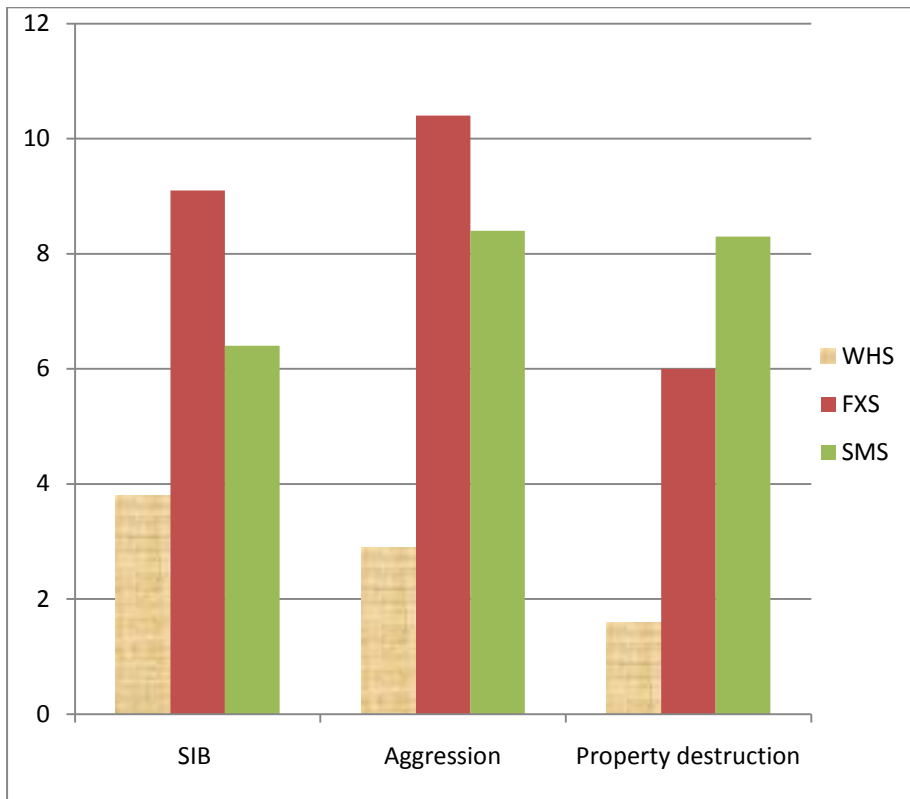


Figure 6 QABF physical discomfort scores across syndromes

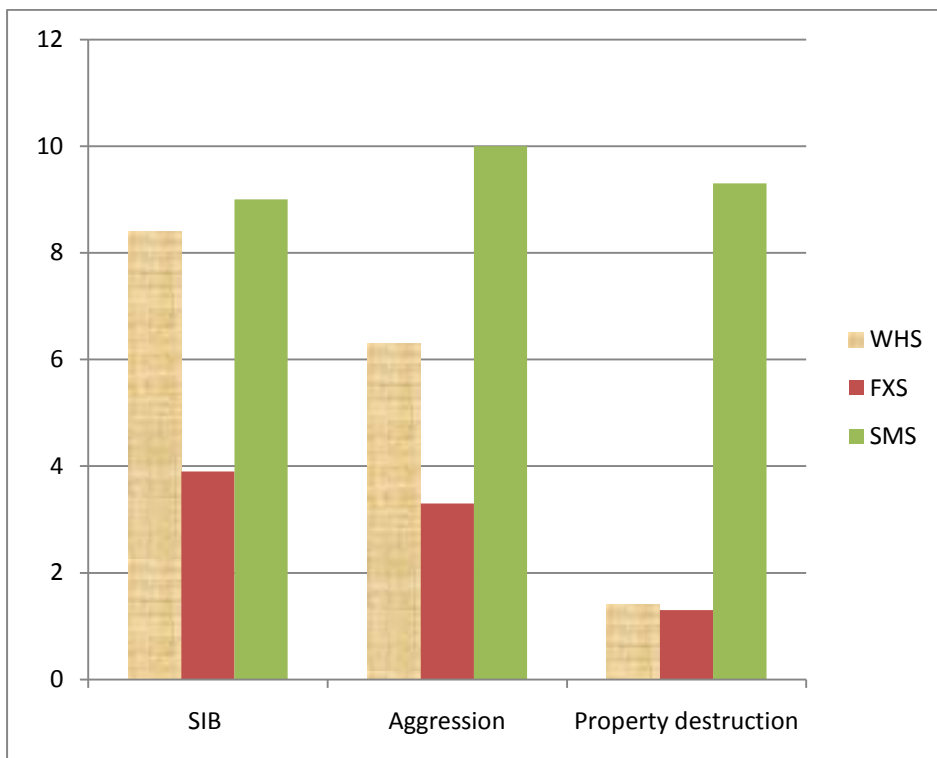
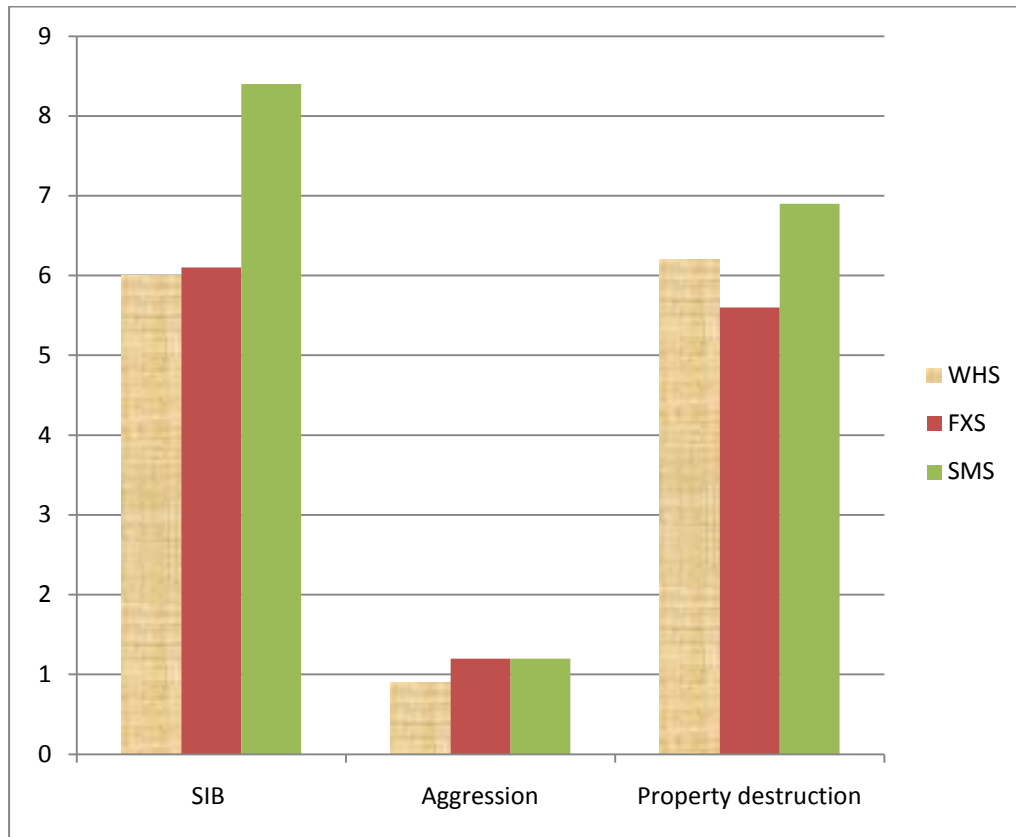


Figure 7 QABF automatic scores across syndromes



Social Impairment (Autistic Spectrum Disorder symptoms)

The SCQ was completed for 40 participants. The mean score was 18 (range: 4-28) and 28 participants (70%) scored 15 or over, the recommended cutoff for indication of possible ASD. Of these, 16 (40% of the whole sample) scored 22 or more, the recommended cutoff for possible Autism. This may, however, overestimate the presence of ASD/Autism as the SCQ score is likely to be artificially high in people with severe learning disabilities without ASD. The mean SCQ score for those participants who had an overall Vineland age equivalent score of 24 months or over (leading to the inclusion of 17 participants) was 12.3 (range: 4-28). Of these, a total of 5 participants (29%) met criteria for the presence of ASD, 1 of whom met the criteria for Autism. Table 6 compares WHS scores with those in other groups including other

genetic syndromes. It should be noted that the Vineland age equivalent scores of the Cornelia de Lange and Cri du Chat groups were substantially higher than the overall WHS group and a comparison with the smaller WHS group (N=17) is more appropriate. Recent research (e.g., Moss, Oliver, Berg, Kaur, & Jephcott, 2008) has concluded that the prevalence of ASD is heightened in people with CdL while it is not thought to be heightened in CdC. The figures presented suggest that people with WHS fall somewhere between the two syndrome groups and that the prevalence of ASD might be usefully investigated further.

Table 6 Comparison of SCQ scores across groups

SCQ Total Scores	WHS N=40	WHS N=17	Autism ³ CdC ⁶		Children ⁴	CdL ⁵
Mean	18.0	12.3	24.2	5.2	14.6	10.0
% 15 or more	70	29	96	0	41	9
% 22 or more	40	6	75	0	8	0

Family Support

Family Support Questionnaires were completed for 44 people. Descriptive statistics are presented in Tables 7 and 8.

³ Children with a diagnosis of Autism – from Berument, Rutter, Lord, Pickles & Bailey (1999)

⁴ Adopted children from the general population – from Rutter et al (1999)

⁵ Children and young people with Cornelia de Lange syndrome – from Moss et al (2008)

⁶ Children and young people with Cri du Chat syndrome – from Moss et al (2008)

Table 7 Nature of Support provided to families

	% received support	Av number of inputs	% rated as helpful
Medication	90.9%	2.3	79.5%
Psychological	50.0%	1.1	54.5%
Communication	95.5%	1.3	61.9%
Other Supports	97.7%	2.6	93.0%
Respite Care	77.3%	N/A	87.1%

Table 8 Average Satisfaction on a scale from 1 (very dissatisfied) to 7 (very satisfied)

	M	% satisfied (ratings of 5-7)	% dissatisfied (ratings of 1-3)
Overall satisfaction.	4.9	68%	16%
Medical.	5.4	75%	14%
Psychological	3.4	7%	34%
Communication	4.1	41%	30%
Other	5.4	74%	9%
Respite	4.9	61%	20%

The tables suggest relatively high levels of satisfaction with services received.

However, satisfaction with psychological input amongst WHS families was rather

lower. Only half the families were offered any input and, of these, only half found it helpful. It should also be noted that the most frequent comment made by families in the open section of the interview related to the struggle/fight/battle they had had in order to get some of the support they needed.

Associations between measures

Adaptive Behaviour and Challenging Behaviour

Table 9 shows correlations between Vineland and ABC scores.

Table 9 Correlations between adaptive and challenging behaviour scores

Vineland ABC	Composite	Communication	Daily Living	Social	Motor
Total	-0.32*	-0.16	-0.15	-0.15	-0.04
Irritability	-0.24	-0.13	-0.13	-0.05	0.04
Lethargy	-0.30*	-0.39**	-0.28	-0.43**	-0.29
Stereotypy	-0.40**	-0.43**	-0.40**	-0.49**	-0.46**
Hyperactivity	-0.21	0.00	-0.01	0.01	0.16
Inappropriate speech	0.08	0.48**	0.35*	0.52**	0.48**

* significant at $p < 0.05$ ** significant at $p < 0.01$

A number of observations can be made from the Table:

1. Most of the correlations are negative. That is, overall, higher levels of challenging behaviour are reported in participants with lower levels of adaptive behaviour/lower developmental levels.

2. This pattern is exemplified by the significant negative correlations between the Lethargy/Stereotyped behaviour scales and Vineland scores for communication, daily living, socialization and motor skills.
3. The exception to this overall pattern, however, is that correlations between the Vineland and the Inappropriate speech scale were significant and positive. That is, participants with higher Vineland scores also scored more highly in Inappropriate speech. This is likely to be a result of all of the relevant ABC items requiring the participant to speak so that participants with lower developmental levels (who do not speak) will not score highly on these items.
4. Most of the more “challenging” behaviours on the ABC (e.g. aggression, destruction, disruption) are on the Irritability and Hyperactivity scales. It is notable that there were no significant correlations (positive or negative) between adaptive behaviour and scores on these scales.

Those participants who displayed self-injury (and for whom the self-injury scale of the BPI was completed) were compared with those who did not. Those displaying SIB were substantially more disabled overall showing significantly poorer daily living, socialization and motor skills.

Challenging behaviour and social impairment (ASD symptoms)

Correlations between SCQ and ABC scores are shown in Table 10. The scores are presented both for the entire WHS group and for the smaller group (N=17) with a developmental age of 24 months or more.

Table 10 Correlations between SCQ and ABC scores

	WHS N=40	WHS N=17
ABC total	0.28	0.44
Irritability	0.18	0.20
Lethargy	0.41**	0.47
Stereotyped	0.50**	0.25
Hyperactivity	0.09	0.34
Inappropriate speech	-0.22	0.58*

The pattern of correlations is similar in the larger and smaller groups with the exception of those with Inappropriate speech. In the larger group the negative correlation likely reflects the absence of speech in more disabled people with WHS while, in the smaller group, the significant positive correlation probably reflects some overlap in the items used by the two scales e.g. in respect of repetitive speech. The overall positive pattern of correlations is consistent with existing knowledge that people with ASD (or ASD like symptoms) are more likely to display challenging behaviour.

Those participants who displayed self-injury (and for whom the self-injury scale of the BPI was completed) were compared with those who did not. Those displaying SIB had significantly higher scores on the SCQ and 89% scored above the cut-off for possible ASD.

Discussion

The current research has provided a description of the behavioural profile associated with WHS. To date, only a small number of studies have examined adaptive and challenging behaviours associated with the syndrome (e.g., Fisch, et al., 2008; Sabbadini, et al., 2002). The current study extends this line of work through the use of additional measures of the form and function of challenging behaviour and the larger sample size than that used in other studies. The current study also collected data on the nature of supports received from local services as reported by families.

Summary of findings

Adaptive behaviour

- People with WHS, generally, have severe deficits in adaptive behaviour. The average developmental age equivalent was just over 2 years and the majority of the sample scored in the profoundly intellectually disabled range;
- Deficits in adaptive behaviour were, on average, more severe than those of comparison groups consisting of people with Fragile X Syndrome (FXS), Smith-Magenis Syndrome (SMS) and people with learning disabilities of a mixed etiology;
- A minority of the sample presented much less severe deficits in adaptive behaviour more consistent with a moderate or mild intellectual disability;
- The extent of variability in adaptive behaviour was greater in the sample of people with WHS than in the comparison groups. That is, in a number of areas the WHS group included both the least able and the most able of all those for whom scores were available;

- Significant within-group differences were found in the profile of scores for the WHS group. Scores on the socialization subscale were significantly higher than other adaptive behaviour domains.

Social Impairment

- Detecting the presence of ASD in people with WHS is more difficult with respect to the more disabled individuals. At a minimum, it appeared that close to a third of participants met criteria for possible ASD including a smaller number meeting the criteria for Autism itself.

Challenging behaviour

- On average people with WHS were reported to display less, and less serious, challenging behaviour than people with Prader-Willi Syndrome (PWS), SMS and Cri du Chat Syndrome;
- There was considerable variability in scores relating to challenging behaviour. Almost a third sometimes displayed aggressive behaviour and over a fifth sometimes behaved destructively;
- Nearly half of the sample displayed at least 1 form of self-injury (SIB). The most common topographies were teeth grinding, self-biting and head banging;
- In relation to behavioural function, significant within-group differences were found for all three forms of challenging behaviour. It appeared that people with WHS presented with relatively high levels of attention-maintained challenging behaviours and relatively low levels of challenging behaviour maintained by other functions such as escaping from demands or gaining access to preferred objects or activities;
- Physical discomfort/pain may be a relatively frequent contributing factor to the self-injurious and aggressive behaviour of people with WHS;

- Overall, challenging behaviour was more likely to be reported in people with WHS who had lower levels of adaptive behaviour and higher levels of social impairment;
- Those displaying SIB were substantially more disabled with poorer daily living, socialization and motor skills and higher levels of social impairment than those who did not display SIB.

Family support

- The most commonly provided form of support was ‘other’ (e.g., physiotherapy, occupational therapy). The least common type of support came from psychologists;
- The most ‘helpful’ source of support was ‘other’, the least helpful was psychology;
- Families reported relatively high levels of satisfaction with all kinds of support other than psychological input though a number of families reported having had to fight to obtain support.

Comparison with other studies

Few studies have discussed the presence of challenging behaviour in WHS. Sabbadini et al (2002) noted that 7/11 participants in their study were reported to have behavioural disorders. However, little detail was provided on the form and function of those behaviours. As such the current study has provided the most comprehensive current description of the phenomenology of challenging behaviour in WHS.

Fisch et al (2008), using the CARS as an ASD screener, reported that only 1/12 participants met criteria for possible ASD. However, his study only included more able people with WHS. The measure used in the current study probably overestimates the presence of ASD but the sample was much more reflective of the

overall population of people with WHS. It is likely that “true” levels of ASD lie somewhere between the figures reported above and those reported by Fisch.

The finding that people with WHS have a relative strength in social skills relative to other domains of adaptive behaviour (such as communication skills, daily living skills, and motor skills) is consistent with those reported by Fisch et al (2008), adding to its robustness. Overall, however, the sample was more severely disabled than that reported by Battaglia et al (2008) who found 65% to have a severe learning disability (91% in the current study), 25% a moderate disability (2% in current study) and 10% a mild disability (7% in current study). These differences may be to do with the nature of the samples (see section on *Limitations* below) or may be to do with the assessment measures used or definitions of degree of disability applied.

Families reported relatively high levels of satisfaction with services received by comparison with data using the same measure in a study of family carers of children and young people with learning disabilities and challenging behaviour (McGill, et al., 2006). In that study, for example, overall satisfaction averaged 2.8 (vs 4.9 in the current study) and, in every category, the percentage of families dissatisfied was substantially higher than the percentage satisfied. However, overall ratings of psychological input were much more comparable to those found by McGill. For example, current satisfaction with psychological input averaged 3.4 (2.8 in challenging behaviour study) and 34% of families were dissatisfied as against only 7% satisfied (66% vs 19% in challenging behaviour study). The theme of interaction with services as a constant battle was also echoed in the findings reported by McGill.

Limitations

As always in research this study has a number of limitations which should be noted:

- While the sample was relatively large (by comparison with previous research) it remains uncertain how representative it is of the larger population of people with WHS. Families were recruited through the WHS Trust. Not all families in the UK, of course, are linked to the Trust and it is not known if there are differences between families linked and not linked. For example, it might be possible that families linked to the Trust would have a child with (on average) more severe disabilities than families not linked to the Trust. This carries the clear implication that future research should seek to recruit families who are not linked to the Trust as well as those that are;
- Confirmation of a WHS diagnosis was only available for 20 out of the 49 families. While it is highly unlikely that a substantial number of the sample did not have WHS it remains possible that, in a very small number of cases, the diagnosis (perhaps made some time ago) was problematic;
- Although no gender differences were found in the current study it should be noted that the roughly equal gender distribution of the participants was surprising. Previous research (e.g., Battaglia, et al., 2008) has suggested that females with WHS outnumber males with WHS by approximately 2:1.

Suggestions for further research

- Very little is known about the developmental trajectory of people with WHS. The current study could only provide a snapshot of behavioural characteristics at one point in time. It would be very useful to follow up the participants looking at changes over the course of time. Such follow-ups would focus on both development during childhood and change during adulthood (e.g., the extent to which anecdotal reports of “premature aging” are supported). A longitudinal

programme of research would also help to establish a large, well-described sample from which smaller samples could be drawn for the more detailed investigation of particular issues;

- It is clear from the current study that a significant number of people with WHS present challenging behaviours, perhaps especially of self-injurious and stereotypic kinds. More research is needed into the severity and impact of the behaviours they present and the causes of these behaviours. It might be particularly useful to investigate the possible relationship between challenging behaviour and physical health issues;
- Previous research (Battaglia, et al., 2008) has suggested that some of the characteristics of WHS are related to the size of the chromosomal deletion. It would clearly be useful for future research to consider this issue in respect of adaptive and maladaptive behaviour;
- The relatively high prevalence of ASD found in the sample suggests that it would be useful to investigate this with more detailed assessment measures.

Implications of the research for people with WHS and their families and for supports provided

- The measures used in the current study were not the most sensitive to the severity and impact of challenging behaviours presented. There was some suggestion, however, that self-injury was relatively frequent and something of a “hidden” problem, not always identified immediately as “challenging”. When coupled with the apparently limited psychological input to families this carries the possible implication for some families that more professional input should be sought and it suggests that psychological or behaviour support services should, routinely, investigate possibility of a link to physical health issues;

- Many families probably already appreciate their son/daughter's relatively good social skills and liking for their and others' attention. While deficits in other areas should not be ignored it is obviously also important to build on strengths. This would imply (amongst other things) using (e.g. as a reward) the liking for attention to aid in the development of other skills, encouraging participation in activities which allow the use of social skills and encouraging the greatest development possible in the area of social skills;
- It is often forgotten that different disorders can co-exist and the "problems" an individual has are attributed to his or her "primary" disorder. Should families of someone with WHS suspect that their family member may also have Autism or a related disorder they should not hesitate from asking for a diagnostic evaluation.

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