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Distinctiveness and correlates of maladaptive behaviour in children and adolescents with Smith–Magenis syndrome

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Abstract

This two-part study examines the distinctiveness and correlates of maladaptive behaviour in 35 children and adolescents with Smith–Magenis syndrome, a developmental disorder caused by an interstitial deletion of chromosome 17 (p11.2). Study 1 compares Child Behavior Checklist scores in 35 children with Smith–Magenis syndrome to age- and gender-matched subjects with Prader–Willi syndrome and mixed intellectual disability. Subjects with Smith–Magenis syndrome had significantly higher levels of maladaptive behaviour than the other groups. Although some problems were shared across groups, 12 behaviours differentiated the three groups with 100% accuracy. Study 2 assessed the frequency and correlates of self-injurious and stereotypical behaviours, including unusual features such as nail-yanking, inserting objects into bodily orifices, self-hugging and a 'lick-and flip' behaviour. Nail-yanking and bodily insertions were less common than other types of self-injury, and self-hugs and the 'lick-and flip' stereotypies were seen in about half the sample. Although age and degree of delay were correlated with problem behaviours, sleep disturbance emerged as the strongest predictor of maladaptive behaviour. The implications are discussed for clinical diagnostic ambiguities between the Smith–Magenis and Prader–Willi syndromes, and for intervention.

Keywords Smith–Magenis syndrome, maladaptive behaviour, self-injurious behaviour

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Introduction

Many studies now document the increased risk of behavioural dysfunction in people with intellectual disability in general (e.g. Fletcher & Dosen 1993; Borthwick-Duffy 1994). However, relatively few studies have examined behavioural problems in people with distinctive genetic intellectual disability syndromes (Hodapp & Dykens 1994; Dykens 1995), especially newly described disorders such as Smith–Magenis syndrome. Smith–Magenis syndrome was identified just 15 years ago (Smith *et al.* 1982), and is emerging as a relatively common, although under-diagnosed disorder, with distinctive physical and behavioural features.

Affecting about one in 25 000 births, Smith–Magenis syndrome is caused in most cases by an interstitial deletion of chromosome 17 (p11.2) (Smith *et al.* 1986; Greenberg *et al.* 1991). Many

genes are now mapped to this region, although none explain the syndrome's complex physical and behavioural phenotype (Chen *et al.* 1996). The physical features of Smith–Magenis syndrome include: craniofacial abnormalities such as a flat midface; prominent forehead; a broad nasal bridge and flat head shape; short hands and stature; visual problems (e.g. strabismus and myopia); a hoarse voice; and speech delay with or without hearing loss (for a review, see Smith *et al.* 1998a).

Although the behavioural features of Smith–Magenis syndrome have yet to be widely studied, certain problems are consistently seen. These include: impulsivity, aggression, tantrums, attention-seeking, hyperactivity, sleep disturbance, stereotypies and self-injurious behaviour (Stratton *et al.* 1986; Colley *et al.* 1990; Finucane *et al.* 1994; Dykens *et al.* 1997; Greenberg *et al.* 1996a; Smith *et al.* 1998a). Several key aspects of these behaviours remain unknown.

Firstly, it is unclear how behavioural difficulties in people with Smith–Magenis syndrome differ from others with intellectual disability. Families and professionals alike report that maladaptive behaviour in people with Smith–Magenis is extraordinarily disruptive and hard to manage; indeed, these behaviours are the single best predictor of family stress (Hodapp *et al.* 1998). Even so, comparative studies have not yet determined if behavioural disturbance in Smith–Magenis syndrome is high relative to others with intellectual disability.

Furthermore, some behaviours shown by people with Smith–Magenis syndrome, such as impulsivity, aggression and tantrums, are also seen in groups with heterogeneous or non-specific intellectual disability (e.g. Rojhan & Tasse 1996). Given this overlap, the present study uses a comparison group with heterogeneous intellectual disability, a widely used group in behavioural research (Hodapp & Dykens 1994; Dykens 1995, 1996). Behaviours in people with Smith–Magenis syndrome also overlap with other genetic syndromes. Indeed, more than a dozen cases have recently been reported at professional meetings that were either Smith–Magenis cases misdiagnosed with Prader–Willi syndrome or Prader–Willi cases misdiagnosed with Smith–Magenis syndrome.

The Smith–Magenis and Prader–Willi syndromes are genetically distinct, yet these conditions share certain phenotypic traits which may create diagnostic confusion, especially among clinicians who are unfamiliar with either condition. As noted by Greenberg *et al.* (1996b), overlapping features of the Prader–Willi and Smith–Magenis syndromes include: infantile hypotonia, hyperphagia, short stature, small hands and feet, skin-picking, and sleep and behaviour problems. Although behaviour problems are globally noted in both syndromes, more detailed comparisons are necessary if behaviour is to play a meaningful role in the differential diagnosis of these disorders.

In addition to between-group approaches, studies are needed which assess the correlates of maladaptive behaviour within Smith–Magenis syndrome, including some unusual self-injurious and stereotypical behaviours. Many people with Smith–Magenis syndrome reportedly pull out their finger- or toenails (onychotillomania), and insert foreign objects into bodily orifices (polyembolokoilamania) (Greenberg *et al.* 1991, 1996a). Two unusual stereotypies have also been reported in this population. The 'upper body spasmodic squeeze', or self-hug, is seen primarily when people are happy or excited, and is viewed as one of the syndrome's more endearing features (Finucane *et al.* 1994). Another stereotypy involves sequential hand-licking and page-flipping, seen with remarkable consistency across subjects during their cognitive test sessions (Dykens *et al.* 1997).

It is unknown how these or other behaviours relate to age, gender or the level of developmental delay. In addition, sleep disturbance may also prove to be an important correlate of maladaptive behaviour in Smith–Magenis syndrome. Seen in 75% or more of samples, sleep disturbance in Smith–Magenis syndrome primarily involves reduced REM, frequent and prolonged night-time arousal, early awakening, and daytime sleepiness (Greenberg *et al.* 1991, 1996a; Smith *et al.* 1998b).

In this two-part study, the present authors first compare maladaptive behaviour in children and adolescents with Smith–Magenis syndrome to age- and gender-matched subjects with Prader–Willi syndrome, and heterogeneous intellectual disability. In study 2, they assess how maladaptive, self-injurious and stereotypical behaviours in Smith–Magenis syndrome relate to age, gender, level of delay and sleep disturbance.

Study 1

Method

Subjects

The participants were 105 children and adolescents (45 males and 60 females) with intellectual disability, aged 4–20 years (mean = 9.57 years). Subjects belonged to one of three aetiological groups: Smith–Magenis syndrome, Prader–Willi syndrome and heterogenous intellectual disability. There were 35 subjects (15 males and 20 females) in each aetiological group, and subjects were matched across groups on gender and age. The mean age in each aetiological group was 9 years. All subjects lived at home with their families.

Subjects with Smith–Magenis syndrome were recruited through the parent group, Parents and Researchers Interested in Smith–Magenis Syndrome (PRISMS), and also during the 1997 First National Conference on Smith–Magenis Syndrome. As per parental report (including test date, and name of laboratory or medical centre), all subjects had cytogenetically confirmed diagnoses of Smith–Magenis syndrome (del 17p11.2).

Subjects with Prader–Willi syndrome were obtained through the Prader–Willi California Foundation and through the University of California, Los Angeles Prader–Willi Syndrome Clinic. The majority of subjects, 83% (n = 29) had genetic testing confirming the Prader–Willi syndrome diagnosis. Six subjects met the clinical criteria for Prader–Willi syndrome (Holm *et al.* 1993) and were awaiting results of genetic testing at the time of the study. None of these six subjects were suspected of having Smith–Magenis syndrome.

Subjects with heterogeneous intellectual disability were recruited through public schools and recreation programmes for children with disabilities in California and the north-east. As per parent and teacher reports, subjects did not have Smith–Magenis or Prader–Willi syndrome, nor any other commonly known genetic disorder. This group may have included subjects with undetected genetic anomalies, yet this was considered acceptable as the goal of the present authors was to compile a group of individuals with mixed or unknown aetiologies.

The subjects' level of delay was ascertained through a combination of IQ and adaptive behaviour standard scores. For participants with Prader–Willi syndrome or mixed intellectual disability, the level of delay was identified by parental report of previously administered IQ tests. For subjects with Smith–Magenis syndrome, level of delay was identified by standard composite scores on the Vineland Adaptive Behaviour Scales (Sparrow *et al.* 1984). The Vineland Scales were administered to parents over the telephone by trained, advanced-level psychology graduate students. A high correlation is found between IQ and Vineland composite standard scores in people with Smith–Magenis syndrome (r = 0.92; Dykens *et al.* 1997).

The level of delay differed significantly across groups [F (2, 93) = 22.49; $P < 0.001$]. Consistent with previous studies, participants with Smith–Magenis syndrome showed moderate delays (mean = 50.38; SD = 14.33). These scores were significantly lower than the Prader–Willi syndrome (mean = 69.46; SD = 5.89) or the heterogeneous groups (mean = 59.85; SD = 10.49). As such, the level of delay was used as the covariate in all between-group analyses.

Given the phenotypic overlap between Smith–Magenis and Prader–Willi syndromes, the subjects' body mass indices (BMIs) were calculated: BMI = weight (kg)/[height (m)]^2. The Smith–Magenis syndrome group had lower mean BMIs than the Prader–Willi syndrome group [F (1, 60) = 8.63; $P < 0.005$; mean = 19.96 and 27.50, respectively]. The BMIs were not correlated with maladaptive behaviour or level of delay in either group.

Forty per cent of subjects with Smith–Magenis syndrome were taking psychotropic medication(s) at the time of the present study. An additional 14% were taking a psychotropic agent along with melatonin (to aid sleep) and 8% were on melatonin alone. As determined by one-way analyses of variance (anovas), subjects on or off medication did not differ in age or in their global CBCL domains, or specific maladaptive or sleep behaviours. Only 14% of the subjects with Prader–Willi syndrome were on medication and medication data were not available for the heterogeneous group.

Procedures and measures

Parents were invited to participate in an ongoing study on the behaviour and development of people with Smith–Magenis syndrome, Prader–Willi syndrome or

intellectual disability in general. The parents of all subjects were asked to complete a packet of questionnaires that included a demographic sheet and the Child Behavior Checklist.

Demographic sheet

Parents were asked to note their affected child's age, gender and previous intelligence test results on a demographic sheet. The parents of children with Smith–Magenis syndrome and Prader–Willi syndrome were also asked to supply genetic testing information (where and when testing was conducted, by whom, and the results), as well as their child's height and weight.

Child Behavior Checklist

The Child Behavior Checklist (CBCL; Achenbach 1991) is a widely used standardized measure which asks parents to rate 112 problem behaviours on a three-point scale (0 = not true; 1 = somewhat or sometimes true; 2 = very true or often true). In addition to a total score, the CBCL contains two broad domains, internalizing and externalizing, as well as nine sub-domains. The internalizing domain has three sub-domains (Withdrawn, Anxious/Depressed and Somatic Complaints) and two sub-domains comprise the externalizing domain (Delinquent Behaviour and Aggressive Behaviour). The remaining sub-domains include Social, Thought, Attention and Other Problems. The CBCL raw scores were used in data analyses.

Results

Between-group analyses

CBCL domains. The internalizing, externalizing and total domain scores of the CBCL were compared across the three groups in a MANCOVA, with the level of delay as the covariate. The MANCOVA was significant [F (6, 178) = 14.20; $P < 0.0001$], with univariate analyses showing significant differences in all three domains. NeumanKeuls *post hocs* revealed that the Smith–Magenis group had significantly higher scores than remaining groups in the externalizing and total domains. The Smith–Magenis syndrome and Prader–Willi syndrome groups had higher scores than the mixed group in the internalizing domain. Table 1 summarizes each group's mean CBCL domain scores, standard deviations and univariate F-values.

The CBCL total scores were converted to t-scores, and based on cut-points established by Achenbach (1991), the percentage of subjects in each group was calculated which showed clinically

Table 1 Group means (standard deviations in brackets) and **F-values for the CBCL domains and sub-domains**

CBCL	Smith–Magenis syndrome	Prader–Willi syndrome	Mixed intellectual disability	F-value
Domains				
Internalizing†	10.43 (7.71)	13.25 (6.94)	5.77 (6.11)	11.32***
Externalizing‡	23.28 (9.46)	18.03 (8.74)	12.74 (12.94)	6.27**
Total CBCL‡	74.82 (27.87)	61.31 (19.94)	36.51 (27.29)	17.81***
Sub-domains				
Somatic Complaints†	2.74 (2.61)	3.60 (2.46)	0.37 (.84)	19.49****
Social Problems†	7.51 (3.18)	7.80 (3.18)	4.86 (3.33)	11.24****
Thought Problems†	3.40 (2.27)	3.28 (1.77)	1.71 (2.35)	8.24***
Other‡	18.85 (3.94)	11.77 (3.68)	3.51 (3.30)	90.85****
Aggressive Behaviour§	19.80 (7.63)	14.08 (6.53)	10.74 (10.65)	6.42***
Withdrawn¶	3.48 (2.80)	5.63 (3.32)	2.23 (2.61)	13.40****
Anxious/Depressed	4.20 (4.24)	4.02 (3.30)	3.17 (4.38)	0.85
Attention Problems	11.37 (4.21)	7.17 (2.45)	7.91 (3.98)	2.57
Delinguent Behaviour	3.48 (2.64)	3.94 (2.85)	2.00 (2.08)	5.01

** $P < 0.01$; *** $P < 0.001$; **** $P < 0.0001$.

†SMS, PWS > Mixed; ‡SMS > PWS > Mixed; §SMS > PWS, Mixed; ¶PWS > SMS, Mixed.

elevated scales. Clinically elevated scores were seen in 89% (n = 34) of subjects with Smith–Magenis syndrome, as opposed to 71% of the Prader–Willi syndrome group and 28% of the mixed group [χ^2 (2) = 28.63; $P < 0.0001$].

CBCL sub-domains. Significant CBCL domain findings were further examined with a MANCOVA using the nine CBCL sub-domains, with the degree of delay as the covariate. The MANCOVA was significant [F (18, 166) = 12.32; $P < 0.0001$]. Using a Bonferonni correction of $P < 0.001$, significant differences were found in six out of the nine sub-domains. As shown in Table 1, NeumanKeuls *post hocs* revealed that the Smith–Magenis syndrome group was significantly higher than remaining groups in the Aggression and Other sub-domains. The Smith–Magenis syndrome and Prader–Willi syndrome groups were higher than the mixed group in Social Problems, Thought Problems and Somatic Complaints, and the Prader–Willi syndrome group was singularly high in the Withdrawn domain.

CBCL behaviours. To identify specific behaviours associated with Smith–Magenis syndrome, frequently occurring behaviours were tabulated or those CBCL items which occurred in 50% or more of any group; 35 behaviours occurred frequently. Among the subjects with Smith–Magenis syndrome, 94–100% showed disobedience, hyperactivity, tantrums, attention-seeking and sleep disturbance. Other frequent behaviours in this group included: lability (89%), property destruction (86%), impulsivity (86%), bed wetting (80%), argumentative (80%), nail-biting (72%), nervousness (66%), physical aggression (57%), and daytime wetting or soiling (54%).

Using mean scores, a MANCOVA was conducted with the 35 frequently occurring behaviours, with the level of delay as the covariate. This was significant [F(74, 110) = 12.67; $P < 0.0001$], and adopting a Bonferroni-corrected value of $P < 0.001$, 23 behaviours showed significant group differences. An exploratory, step-wise discriminant function analysis was then conducted to identify which of these 23 behaviours best differentiated the three groups. The discriminant analysis yielded 12 differentiating behaviours, and Table 2 shows the means, similarities and differences in these 12 behaviours across groups. The 12 behaviours correctly predicted membership into the three groups with 100% accuracy; in other words, none of the 105 cases were misclassified.

Study 2

Methods

To examine correlates of maladaptive behaviour in children with Smith–Magenis syndrome, the 35 parents of these subjects were administered the Self-Injury and Stereotypy Checklists, and the Sleep Questionnaire. Within-group analyses also used the CBCL.

Table 2 Means (standard deviations in brackets) of the 12 behaviours which best predicted group membership in the discriminant function analysis

Behaviour	Smith–Magenis syndrome	Prader–Willi syndrome	Mixed intellectual disability
Sleeps less than others†	1.63 (0.60)	0.31 (0.72)	0.06 (0.24)
Wets bed†	1.26 (0.78)	0.37 (0.60)	0.00 (0.00)
Bowel movements outside toilet‡	0.69 (0.80)	0.03 (0.17)	0.00 (0.00)
Demands a lot of attention‡	1.91 (0.28)	1.06 (0.80)	1.20 (0.87)
Sudden changes in mood, feeling‡	1.51 (0.70)	0.89 (0.63)	0.71 (0.75)
Underactive, slow moving§	0.46 (0.74)	1.46 (0.61)	0.29 (0.57)
Withdrawn§	0.26 (0.51)	0.71 (0.71)	0.11 (0.32)
Skinpicks¶	1.34 (0.68)	1.63 (0.49)	0.06 (0.24)
Sleeps more than others¶	0.37 (0.69)	1.31 (0.72)	0.06 (0.24)
Overeats¶	0.60 (0.81)	1.23 (0.77)	0.23 (0.60)
Obsessions¶	1.00 (0.87)	1.40 (.65)	0.49 (0.82)
Bites nails††	1.34 (0.87)	1.00 (0.91)	0.17 (0.38)

†SMS > PWS > Mixed; ‡SMS > PWS, Mixed; §PWS > SMS, Mixed; ¶PWS > SMS > Mixed; ††SMS, PWS > Mixed.

Stereotypy Checklist

The Stereotypy Checklist (Bodfish *et al.* 1995) is a 36-item Yes/No answer checklist which asks parents to indicate any stereotypies shown by their child. Stereotypies are defined as repetitively showing certain movements, with or without an object, and usually repeating these movements twice or more in quick succession. Two stereotypies specific to Smith–Magenis syndrome were added to the Checklist: hugging or squeezing upper body; and licks hand/fingers and or flips pages repetitively. Raw scores, ranging from 0 to 36, were used in data analyses.

Self-Injury Checklist

The Self-Injury Checklist (Powell *et al.* 1996) asks parents to indicate if their child currently shows 10 different self-injurious behaviours, defined as repetitive movements or behaviours which have the potential to cause redness, bruising, soreness or other self-injury. Two self-injurious behaviours specific to Smith–Magenis syndrome were added: pulls out finger or toe nails, and inserts fingers or objects into bodily openings. Raw scores range from 0 to 10.

Sleep History Questionnaire

The Sleep History Questionnaire (Smith *et al.* 1998b) was specifically designed to identify sleep problems in people with Smith–Magenis syndrome. This Questionnaire asks parents to note the presence or absence of sleep problems in four domains: Getting to Sleep (e.g. 'repeatedly gets out of bed' and 'refuses to sleep alone'); During Sleep (e.g. 'snores' and 'awakens in night'); Awakening (e.g. 'wakes up looking tired); and Daytime Naps. Results from the Sleep History Questionnaire have previously been reported (Smith *et al.* 1998b). In this study, the present authors assessed if sleep scores were correlated with maladaptive behaviour.

Results

Within Smith–Magenis syndrome analyses

Self-injurious and stereotypical behaviours. The vast majority of subjects with Smith–Magenis syndrome (92%) showed one or more self-injurious behaviours on the Self-Injury Checklist. The mean number of self-injurious behaviours was 3.69 (SD = 2.07). The most frequently noted self-injurious behaviours were bites self (77%) and hits self (71%). Only 29% of subjects showed nail-yanking and 25% inserted foreign objects into bodily openings.

All Smith–Magenis syndrome subjects (100%) showed one or more stereotypies on the Stereotypy Checklist; the mean number of stereotypies was 10.66 (SD = 5.73). The most frequent stereotypies were: inserting hands (69%) or objects (54%) into the mouth, and grinds teeth (54%). On the two items specific for Smith–Magenis syndrome, 51% showed the 'lick and flip' repetitive page turning, and 46% showed the self-hug.

Self-injury and stereotypy checklist correlates. No significant correlations were found between the total number of self-injurious or stereotypical behaviours, and age, gender, level of delay and the total number of sleep problems. However, nail-yanking was positively correlated with age [r (34) = 0.39; $P < 0.01$] and negatively associated with level of delay [r (34) = 0.48; $P < 0.01$].

CBCL correlates. Age was positively correlated with the CBCL total score [r (34) = 0.47; $P < 0.01$]. No gender differences in CBCL scores were found. The level of delay was negatively correlated with total CBCL scores [r (34) = 0.57; $P < 0.001$].

The total CBCL domain was positively correlated with the total number of sleep problems (r = 0.57; $P < 0.001$) and with three sleep domains: Awakening, During Sleep and Daytime Naps (r = 0.54, 0.51 and 0.38, respectively; $P < 0.001$ and 0.05). Only a few items in each domain related to CBCL scores. As shown in Table 3, laboured breathing at night was correlated with the total CBCL, as well as with aggressive and acting out behaviour. Although snoring was also positively associated with aggressive behaviours, snoring correlated the strongest with attentional problems. Awakening at night with a bad dream correlated best with the anxious/depressed domain. Increasing length of day time naps was associated with decreased CBCL total scores, aggressive behaviour and attentional problems.

To further assess the strength of these correlations, a step-wise regression was conducted with the CBCL total score as the dependent

Table 3 Correlations between various CBCL and sleep domains in subjects with Smith–Magenis syndrome

CBCL	Total sleep problems	Nap length	Snoring	Laboured breathing	Awakens with bad dream
Total CBCL	0.57***	−0.38*	0.36*	0.49**	0.40*
Aggressive Behaviour	0.55***	−0.45*	0.35*	0.52**	0.43**
Attention Problems	0.47**	−0.52**	0.47**	0.39*	NS
Anxious/Depressed	0.44**	NS	NS	NS	0.57***
Delinquent Behaviour	0.42**	NS	0.37*	0.50**	NS
Social Problems	0.49**	NS	0.37*	0.39*	0.53***
Other	0.51**	NS	NS	0.39*	0.55***

* $P < 0.05$; ** $P < 0.01$; *** $P < 0.001$; NS, not significant.

variable. Three predictors were entered into the analysis: age, the level of delay and total sleep problems. Sleep problems emerged as the strongest predictor of CBCL maladaptive behaviour, accounting for 37% of the variance [F (1, 28) = 16.44; $P < 0.0001$]. The level of delay was the second strongest predictor, accounting for an additional 16% of the variance [F (1, 28) = 15.20; $P < 0.0001$]. Collectively, sleep problems and level of delay accounted for 53% of the variance.

Discussion

Relative to their counterparts, children and adolescents with Smith–Magenis syndrome had significantly higher levels of maladaptive behaviour. Indeed, all but four subjects with Smith–Magenis syndrome (89%) had clinically elevated CBCL *t*-scores. Subjects with Smith–Magenis syndrome had particularly high rates of temper tantrums, disobedience, attention-seeking, property destruction, impulsivity, aggression, hyperactivity, distractibility, toileting difficulties, sleep disturbance, stereotypies, and self-injurious behaviours. Although some of these problems were shared across groups, the findings point to a relatively distinct and complex Smith–Magenis syndrome behavioural phenotype.

Despite some overlap, the three groups differed in their patterns of behavioural problems, especially in two areas. Firstly, differences emerged in the regulation of basic bodily functions: sleeping, modulating activity and affect, eating, and toileting. Subjects with Smith–Magenis syndrome slept less than others and were prone to hyperactivity, whereas those with Prader–Willi syndrome slept more than others and were underactive. All three groups showed problems regulating affect, yet the Smith–Magenis syndrome group was more emotionally labile than their counterparts. Not surprisingly, the Prader–Willi syndrome group was highest in overeating and they also had higher BMIs than Smith–Magenis syndrome subjects. Unlike either group, the Smith–Magenis group was singularly high in symptoms of enuresis and encopresis.

The second pattern of findings is less clear cut, and involves social and repetitive behaviours. Socially, members of all three groups demanded attention from others, yet Smith–Magenis subjects were more demanding than their counterparts. The Prader–Willi syndrome group was singularly high in a proneness for social withdrawal. Repetitive skin-picking was highest in the Prader–Willi syndrome group, as were obsessions about food. Subjects with Smith–Magenis syndrome also showed obsessive thinking, yet they were apt to think about specific topics or events as opposed to food.

Thus, both similarities and differences are found across groups, with 12 behaviours predicting group membership with 100% accuracy. Such accurate classifications have important clinical implications. When faced with uncertainties between the Smith–Magenis and Prader–Willi syndromes (e.g. Greenberg *et al.* 1996b), diagnosticians may find it helpful to probe their patients for information regarding regulatory behaviours of sleep, affect, activity level, eating and toileting, and to some extent, social and repetitive behaviours as well.

Further insight into the Smith–Magenis syndrome phenotype is gained through the within-syndrome analyses of self-injurious, stereotypical and other behaviours in study 2. The majority of Smith–Magenis subjects, 92%, showed self-injurious behaviours, especially self-biting and hitting. These behaviours are likely related to clinical signs of peripheral neuropathy, seen in up to 75% of people with this disorder (Greenberg *et al.* 1996a). Relatively few subjects (25–29%) showed nail-yanking or inserting objects into body orifices, and nail-yanking was associated with advancing age and lower levels of delay. Although self-hugs were seen in about half the sample, the most frequent stereotypies involved the mouth in some way: placing fingers or objects in the mouth, teeth-grinding, and the 'lick-and-flip' behaviour. Nail-biting was also highest in the Smith–Magenis group. All these seem to be oral variants of bodily insertion behaviours.

Many correlations were found between maladaptive behaviour in general and sleep problems. Although age and the level of delay correlated with maladaptive behaviour, sleep problems emerged as the strongest predictor of total CBCL scores. In particular, aggressive, acting out behaviours and attentional problems correlated with sleep difficulties, especially with snoring and laboured nocturnal breathing. These findings are consistent with studies of other groups of children. Children with intellectual disability of mixed aetiologies and comorbid sleep problems show high rates of aggression and temper tantrums relative to children without sleep problems (Wiggs & Stores 1996). Furthermore, non-retarded children with attention deficit hyperactivity disorder (ADHD) show more sleep-disturbance compared with non-ADHD children (Ball *et al.* 1997), and snoring in particular has been associated with hyperactivity in non-retarded children (Ali *et al.* 1994).

Thus, the relationships between sleep disturbance and maladaptive behaviour in Smith–Magenis syndrome are in keeping with other studies, yet several aspects of these relations are unclear. Without longitudinal data, for example, it is unclear to what extent sleep disturbance causes or perpetuates behavioural problems. This issue is complicated by environmental factors. Many parents report less patience in dealing with their child's daytime behaviours when they are up at night attending their child and are themselves sleep-deprived. Less-than-optimal parental interventions may, in turn, exacerbate problem behaviours.

Nevertheless, a causal role for sleep disturbance in this sleep-behaviour-intervention cycle is suggested by the finding that increased nap length is associated with decreased aggressive and attentional problems. Although preliminary, these findings provide encouraging grounds for future psychotropic or behavioural studies which aim to reduce sleep disturbance in people with Smith–Magenis syndrome, with an eye toward possible improvements in the frequency or severity of maladaptive behaviour.

As work in this newly delineated syndrome is just now underway, future studies are also needed on adults with Smith–Magenis syndrome, presently an underdiagnosed group relative to children. As more genes in the Smith–Magenis syndrome region (del 17p11.2) are discovered, studies are also needed which link gene function to behaviour. However, in the meantime, these findings underscore a pressing need for pharmacological and other interventions aimed at reducing a wide range of problem behaviours in children with Smith–Magenis syndrome.

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