COGNITIVE AND BEHAVIOURAL CHARACTERISITCS OF CHILDREN WITH SMITH-MAGENIS SYNDROME

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by

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ABSTRACT

Cognitive and Behavioural Characteristics of Children with Smith-Magenis Syndrome

by Carolyn Webber

The study aimed to identify behavioural and cognitive characteristics in 29 children with Smith-Magenis syndrome. Cognitive assessments were undertaken on the children, and detailed interviews assessing sleep patterns, maladaptive behaviours, self-injury, hyperactivity and autism were carried out with their parents and teachers.

The study identified high levels of sleep problems, aggression, self-injury, distractibility and autism in the sample, in comparison with rates reported for other groups of children with learning disabilities. These were associated with high levels of stress in the parents.

It is concluded that the **combination** of difficulties and abilities identified in the present sample of children with SMS is indicative of a behavioural phenotype for the syndrome, and that there is an urgent need for intervention studies on the challenging behaviours posed by this group of children.

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1.1 INTRODUCTION

Recent technological advances in molecular and clinical genetics have stimulated renewed interest in research focusing on genotype-phenotype associations in the field of learning disabilities. However, of 1000 known genetic causes of learning disabilities (Sarimski, 1997), most studies have concentrated on dysmorphic features of the physical phenotype, associated conditions and physical disabilities, and relatively few have detailed phenotypic workups (Sarimski, 1997). The new genetics and recent discoveries in specific syndrome groups highlight the importance of studies looking at the behavioural phenotype of various syndromes, and relating these findings to specific genetic features. For example, in fragile X syndrome, the most common inherited form of developmental delay, it has been suggested that variations in gene protein repeats are associated with within-syndrome variations in behaviour (Dykens, Hodapp & Leckman, 1994). It has been argued that behavioural investigations of specific syndromes could have far reaching implications:

'with accurate and thorough behavioural data, refined mappings can be made of specific genes associated with particular behaviours (Epstein et al., 1991)...(and that)...eventually, these mappings of gene functions may help develop various forms of gene therapies (Anderson, 1994)'.

Smith-Magenis syndrome, a multiple congenital abnormality syndrome associated with learning disabilities and developmental delay, is caused by an interstitial deletion

of chromosome 17p11.2, and was first proposed by Smith, McGavran, Magenis et al. (1986). Although more than 100 cases have now been described in the literature, most published reports take the form of either single or small case series, describing variously the physical phenotype, and to a lesser degree some commonly found behavioural and cognitive characteristics. The largest systematic evaluation of individuals with the syndrome (n=27), reports the physical, medical and developmental characteristics but does not address behavioural characteristics (Greenberg, Lewis, Potocki et al., 1996). The present study seeks to add to currently available information about the cognitive and behavioural characteristics associated with the syndrome in childhood. Before reviewing the current literature on Smith-Magenis syndrome in more detail, it is useful to look at the theoretical applications of behavioural phenotypes.

This chapter will examine various definitions of behavioural phenotypes, and consider methods for studying behavioural phenotypes, their use and clinical application. The chapter will conclude with a review of the current literature on Smith-Magenis syndrome and relate this to the aims and hypotheses of the current study.

1.2 Definition of Behavioural Phenotypes

A *phenotype* is the "observable or measurable expression of a gene or genes" (Berini & Kahn, 1987). This broad definition applies to many groups, such as to "normal" individuals, those with psychiatric disorders, learning difficulties, and so on. The

study of behavioural phenotypes, however, is typically limited to "measurable expressions" in individuals with genetic disorders (O'Brien, 1992).

Nyhan (1972) was the first to use the term 'behavioural phenotype'. He suggested an association between an inborn error of purine metabolism and self-mutilatory behaviour in Lesch-Nyan syndrome. The behaviour seemed to relate to the underlying biological diagnosis and was relatively independent of environmental influences.

Flint and Yule (1994) propose that the term behavioural phenotype refers to a characteristic pattern of motor, linguistic and social abnormalities which is consistently associated with a biological disorder. In some cases, the behavioural phenotype may constitute a psychiatric disorder; in others, behaviours which are not usually regarded as symptoms of psychiatric disorders may occur. Thus, a behavioural phenotype refers to the specific and characteristic behavioural repertoire exhibited by individuals with a given genetic or chromosomal disorder. These authors require two conditions to be fulfilled: firstly, there must be a 'distinctive behaviour that occurs in almost every case of the condition and rarely in other conditions', and secondly, a 'direct and specific' relationship to the genetic anomaly causing the condition's physical manifestations must be demonstrated. Flint and Yule (1994) noted that using their strict criteria would only allow three behavioural phenotypes to be identified (Lesch-Nyhan, Prader-Willi and Rett's syndrome).

There is considerable evidence in the literature that a broader definition of the term might be acceptable. Relative to other causes of learning disability, there may be more physical or behavioural homogeneity among individuals with a syndrome because, by definition, they have a common genetic anomaly. However, this is not to say that **all** people with a syndrome share the same characteristic appearance or behaviour. Dykens (1995) suggests that a phenotype may best be described as the 'heightened *probability* or *likelihood* that people with a given syndrome will exhibit certain behavioural and developmental sequelae relative to those without the syndrome' (p.523). Similarly, Turk and Hill (1995) considered a behavioural phenotype as a "pattern of behaviour present in many or most individuals with that condition".

There is a great deal of variability in both the physical and behavioural manifestations of syndromes. For example, approximately 40% of babies with Down syndrome do not have the "classic" epicanthal folds around the eyes (Peuschel, 1983). This variability in expression may be explained in very general terms as the product of an individual's entire genetic endowment (not just their genetic anomaly), their environment, and the complex interactions between the two (Plomin & Rende, 1991; Scarr & McCartney, 1983).

Although there may be within-syndrome variability, certain behaviours may be quite prevalent and striking. These behaviours may characterize a syndrome, although they are not necessarily unique to a given syndrome. For instance, sociability and

friendliness are found in individuals with Williams and Down syndrome (Dilts, Morris, & Leonard, 1990; Gibbs & Thorpe, 1983); and self-mutilation is seen among those with Cornelia de Lange and Lesch-Nyhan syndromes (Nyhan, 1977; Shear, Nyhan, Kirman, & Stern, 1971). There is also overlap in behaviours that are common among individuals with learning disabilities in general, such as aggression, temper tantrums, and stereotypies (Dykens, 1995).

An important issue to consider, however, is that there are often *qualitative* differences in these shared behaviours across syndromes. Many individuals with fragile X and Prader-Willi syndromes are aggressive and have temper tantrums. Boys with fragile X syndrome may hit or bite others with or without a clear precipitant; some of this aggression seems to be associated with sensory sensitivities. In contrast, individuals with Prader-Willi syndrome may have tantrums because of an inability to gain access to food, or because of an idea they obsessively get "stuck on" (Dykens, Hodapp, Walsh & Nash, 1992).

Behavioural phenotypes, then, are characterized by variable expressions within syndromes and by overlap and qualitative differences between syndromes. These properties create many challenges to the study of phenotypes. Many difficulties arise in the choice of research strategies to study behavioural phenotypes, and these will be explored in the following section along with an examination of how behavioural phenotypes can best be studied.

1.3 Methods for Studying Behavioural Phenotypes

Dykens (1995) suggests three approaches to the study of behavioural phenotypes; the psychiatric approach, the psychometric approach, and syndrome specific observations. Phenotypic research may be optimized by combining all three methods. This section will review each approach, along with methodological complications in relation to phenotypic work, including developmental issues. The use of appropriate control or comparison groups will also be examined.

The psychiatric approach focuses on the use of psychiatric diagnosis to describe a particular population. One advantage of this approach is that it addresses 'diagnostic overshadowing', or the tendency for clinicians to attribute maladaptive behaviour to an individual's learning disability (Reiss, Levitan, & McNally, 1982). Psychiatric illness in the population with learning disabilities is therefore often overlooked. There are certain disadvantages to a psychiatric approach. Of primary concern is the "goodness of fit" in applying certain criteria established for individuals without a learning disability to the population of individuals with a learning disability. For example, poor judgement and impulse control are common problems for the latter population, yet these features may or may not be associated with psychiatric illness (Sovner,1986). A further problem with this approach is that accurate diagnosis relies on the individual's ability to label and express certain internal states, which can be difficult for this population.

These diagnostic problems are exemplified by the controversial association between fragile X syndrome and autism. Early case reports of a few boys with fragile X syndrome who met DSM-III criteria for autism created support for the hypothesis that fragile X was a common genetic cause of autistic disorder (Dykens et al., 1994). Only after careful, in-depth studies of the behaviours shown by males with fragile X syndrome was it discovered that most of these individuals do relate to others, are attached to their caregivers, and do not show the profound indifference to others that is a hallmark of autism. Instead of autism per se, most of these males can be placed on a continuum of social anxiety and shyness (Bregman, Leckman, & Ort, 1988), and mutual gaze aversion (Cohen, Vietze, Sudhalter, Jenkins, & Brown, 1991).

The psychometric approach moves the study of phenotypes beyond easily observed traits that are often not measured, to a wide range of behavioural domains that are systematically assessed. These behavioural domains include cognition, developmental level, adaptive skills, and social competence, among others. Standardized, reliable, and valid instruments have been developed to assess these domains, and therefore profiles of strengths and weakness can be established in each area and conveyed in a uniform manner. Psychometric methods have already been used to establish behavioural profiles in several syndromes. It should be noted that some have been normed on populations with learning disabilities , while others, such as the Child Behaviour Checklist (Achenbach, 1991) have not been. A problem with these instruments is that they might skip over uncommon or highly unusual behaviours that might characterize a given syndrome, for example food identification by individuals

with Prader-Willi syndrome or hand ringing by people with Rett syndrome. Within-syndrome differences might also be overlooked. There are clearly limitations in the sensitivity of this approach, especially if it is the sole approach taken.

The third approach is syndrome-specific observations, or detailed observations of behaviour. They may be particularly helpful in describing qualitative differences in shared behaviours across syndromes. For example, Finucane et al. (1994) carried out careful classroom observations of unusual motor movements in individuals with SMS, resulting in a description of an "upper body spasmodic squeeze", which was exhibited when the individuals were excited or happy. This characteristic behaviour would not have been identified had standardized questionnaires alone been done.

A multi-method approach incorporating the three approaches is a useful strategy to apply to the study of genetic syndromes.

The issue of comparison groups is an important matter to consider in the study of behavioural phenotypes. There are two possibilities in terms of choice of population from which to draw a comparison group in a study of behavioural phenotype: a general learning disability population, or other groups with learning disabilities affected by different, discrete and diagnosable biological conditions. Methodological and practical difficulties exist in respect of both approaches. In the first group, in order to choose a representative sample from such a group one would need to consider that such a sample would contain a variety of conditions, some of which will

have their own behavioural characteristics. In order to control for any chance effects of clustering of behaviours, a large control group would need to be employed (O'Brien & Yule, 1995, p.40). Obtaining an equivalent number of individuals in the group under study is not always possible in the case of newly identified and rare genetic conditions. It has been suggested that a preferable control group would be drawn from a population for which no diagnosable disorder has been identified. However, such a control group would simply represent those for whom 'no causal diagnosis has *as yet* been demonstrated' (O'Brien & Yule, 1995, p.40).

The second strategy, which has been favoured, is to make comparisons of the behavioural characteristics of individuals affected by different, discrete and diagnosable biological conditions. This approach has several advantages: First, it is possible to select disorders which have similar impact in terms of general intellectual or developmental disability, and to carry out behavioural comparisons between them. Where certain behaviours are found to be common across a number of disorders, it is most likely that these behaviours are a reflection of the degree of intellectual or developmental disability in question. However, where one condition is found to have a quite different set of behavioural concomitants, this is likely to suggest a specific behavioural phenotype (O'Brien and Yule, 1995).

A decision about the type of control and comparison groups needed depends on the aims of the particular study. Control and comparison groups were rarely used in the early stages of the description of the physical and behavioural phenotype of

biologically distinct syndromes (e.g. Williams syndrome, Udwin, Yule & Martin, 1987).

1.4 Clinical applications of behavioural phenotype research

Professionals often attribute maladaptive behaviours in children with learning disabilities to poor parenting. Turk and Sales (1996) discuss the potential impact of findings from behavioural phenotype research on clinical practice. Professionals working in the field of child and adult mental health need to have an appreciation of the underlying biological abnormalities which might be contributing to challenging behaviours. Such knowledge is essential in order to plan interventions, make appropriate referrals to specialist agencies, maximise the chances for individuals with a disability to reach their full potential, and provides then with enough knowledge to advise parents and carers about the complex interplay between psychological state, environmental influences and social contributors.

Detection and longitudinal follow-up is important, in terms of informing parents and carers about prognosis, looking ahead at possible challenges that are likely to be encountered, as well as giving information on useful interventions and educational approaches that have proved to be effective for others with the same condition. Moreover, if a child has an identifiable genetic or chromosomal abnormality there can be crucial implications for future pregnancies, and genetic counselling is advised. Parents and carers receive invaluable support from syndrome organisations and parent support groups; receiving a diagnosis facilitates membership to these groups. When

planning the present study, contact was made with the Smith-Magenis Parents Support Group (UK) which expressed an urgent need for more information on the syndrome, and on effective strategies that could be usefully employed to manage challenging behaviours in affected individuals. Receiving a diagnosis of a specific condition allows families access to the invaluable support and help that membership of a parent support group offers.

1.5 Examples of syndromes with a distinct and specific behavioural phenotype

The following are examples of syndromes which have a behavioural phenotype associated with them, demonstrating how knowledge of a syndrome can assist greatly with intervention strategies.

Lesch-Nyhan syndrome was first reported in two brothers by Lesch and Nyan in 1964. It is caused by a deficiency of the enzyme hypoxanthine phonsphoribosykltransferase (Stout and Caskey 1989). The syndrome is almost only manifest in males and estimates of incidence range from 1:380,000 live births to 1:10,000 male births. Affected individuals appear normal at birth, but then motor delay and hypotonia became apparent within the first few months.

Severe motor impairment and poor gross and fine motor co-ordination are characteristic, and represent a mixture of abnormal movements, spasticity and ataxia. Delayed speech development and dysarthria are common. Most individuals have moderate or severe learning difficulties though a few fall into the low-average range

and attain age appropriate reading skills. Anecdotal reports describe affected individuals as charming, and with a sophisticated sense of humour. Anderson et al. (1992) found that they had an excellent memory for recent and past events, good concentration with good communication skills.

Lesch-Nyhan syndrome has a striking behavioural phenotype in terms of compulsive self-injurious behaviour. As soon as the children develop teeth they start to bite themselves and others. The biting can be so severe that they may sever fingers, and parts of their lips and tongues. Christie et al. (1982) found that all 19 individuals with the syndrome they examined had extensive loss of lip tissue, and sometimes required arm splints or teeth extraction to prevent further mutilation. In 12 of the cases the diagnosis of the condition was only made after the onset of self-mutilation, which illustrates the diagnostic value of the behavioural phenotype. Self mutilation generally becomes less pronounced after ages 10 to 12.

Although self-injurious behaviour is not unusual in individual's with severe learning disabilities, various features of the behaviour in Lesch-Nyhan syndrome suggest that it is a specific abnormality: Firstly, the self-injurious behaviour in Lesch-Nyhan syndrome is extremely severe and resistant to behavioural modification, and in some cases intervention has worsened the behaviour. This suggests that the behaviour is less environmentally determined than is commonly the case (Anderson et al., 1994). A further interesting point is that although the individuals are aware of their behaviour they express having little control over it and often indicate a preference for

being restrained. Taking these features into account it is evident that Lesch-Nyhan syndrome is qualitatively different from self-injurious behaviour in most other learning disability syndromes.

Prader-Willi syndrome was first described by Prader, Labhart and Willi in 1956. The incidence is estimated at about 1:15,000 live births. Males and females are equally affected. Prader-Willi syndrome is characterized by small stature, hypogonadism, infantile hypotonia and mental retardation. There is an increased prevalence of scoliosis and other orthopaedic abnormalities. This condition is due to a chromosomal abnormality affecting part of the long arm of chromosome 15.

Prader-Willi syndrome is characterized by several distinctive psychological and behavioural features. In early childhood, motor and language development are significantly delayed. It is common to find difficulties with gross motor skills, coordination and balance through childhood. The degree of learning difficulties is variable (IQ 20-90), although it is mainly in the borderline to moderate range, however average IQ or above is reported in at least 3 per cent of cases. Severe learning difficulties are unusual (Sulzbacher et al., 1981; Greenswag, 1987; Curfs and Fryns, 1992). Individuals with Prader-Willi syndrome are found to have an unusual cognitive profile, with particular strengths in visual organization and perception. Auditory information processing and sequential processing are relatively weak. Very few adults with Prader-Willi syndrome achieve a fully independent lifestyle, even

those with IQs in the normal range, probably due to the severe behavioural characteristics associated with the syndrome (Greenswag, 1987).

During the first few years of life children with Prader-Willi syndrome tend to be passive, quiet and characteristically fail to thrive. However, behavioural difficulties become increasingly apparent and severe as the child gets older and are far more frequent in this population when compared with other children matched on age and ability levels (Curfs et al., 1992). From about 2 years of age (range 1-6 years) overeating and an insatiable appetite which are so characteristic of the syndrome, become manifest. The individual begins foraging for food, and if it is withheld they are said to steal food, even consuming 'inedible' material, such as frozen food (Zellweger & Schneider 1968; Holm & Pipes, 1976; Taylor & Caldwell, 1985). Extreme obesity is often the result, with one-third of individuals weighing more than 200% of ideal body weight (Schoeller et al., 1988).

Dietary management is essential to try and control the obesity. Carers have to limit access to food and take over control of food intake. This approach to weight control, and behaviour modification methods which involve reinforcement and self-monitoring as part of weight control and exercise programmes, have been effectively used with Prader-Willi individuals (Descheemaeker et al. 1994).

Hall and Smith (1972) reported that 71% of the 32 individuals they assessed had violent outbursts. General findings are that emotional lability, outbursts of rage and

aggression, stubbornness and belligerence are found in at least 50 per cent of cases. These become more marked in later childhood. Greenswag (1987) and Clarke at al. (1989) noted that carers reported such behaviours persisting into adulthood, with a high rate of belligerence, stubbornness, irritability and impulsiveness, along with compulsive/obsessive behaviours and anxiety-based difficulties, excessive worrying and skin picking. More than 90% of affected individuals self-injure by deliberately picking or scratching their skin, which can lead to infection or cellulitis.

These behaviours are thought to be the result of the combination of the physical aspects to the syndrome (e.g. central nervous system abnormalities, metabolic defects), the insatiable hunger, and the psychosocial pressures of being obese, sexually immature and limited cognitively. In addition, a higher than usual pain threshold is often noted in individuals' with Prader-Willi syndrome.

In between the episodes of difficult behaviour individuals with Prader-Willi syndrome are good natured, placid and co-operative. They tend to have difficulties in social relationships with peers and are often described as being immature, lonely and isolated.

Sleep disorders are common, including sleep apnoea, which could contribute to excessive daytime sleepiness (in over 90 per cent of cases). This could be secondary to obesity (Clarke et al. 1989), or may be due to anatomical narrowing of the upper airway and severe muscle hypotonia.

Williams syndrome (or idiopathic infantile hypercalcaemia). The incidence is estimated at 1:20,000 to 1:25,000 live births. Affected individuals have a characteristic face (wide mouth, long philtrum, thick lips, turned up nose) and heart defects; (Burn, 1986; Morris et al., 1988), notably, supravalvular aortic stenosis and peripheral pulmonary artery stenosis.

Studies have identified a distinctive psychological profile and unusual personality and behavioural characteristics that are associated with Williams syndrome (Udwin et al. 1987, Udwin and Yule 1990). About 55 per cent of individuals with this condition have severe learning difficulties, 40 per cent have moderate learning difficulties, and 5 per cent fall in the borderline to low-range of abilities. Most need special schooling (Udwin et al. 1987) . During the pre-school years language is slow to develop, however by school-age verbal abilities are mostly superior to visuospatial abilities and to gross and fine motor skills. Comprehension is usually far more limited than expressive language. Most individuals have a well developed and precocious vocabulary, with extensive use of clichés and stereotyped phrases. The children present as being chatty, with well developed auditory memory, mimicry skills and social use of language (Bellugi et al. 1994). In contrast they show deficits in the integration of visual-perceptual information and in motor coordination and constructional tasks.

The majority of children with Williams syndrome have poor relationships with peers, but tend to be outgoing, socially disinhibited and excessively affectionate toward adults, including strangers.

Children with Williams syndrome display higher rates of emotional and behavioural disturbance when compared with the rates reported for other children with learning difficulties, especially in terms of overactivity, poor concentration and distractibility, eating and sleeping problems, excessive anxiety and attention seeking behaviours (Udwin et al. 1987). There are high rates of preoccupations and obsessions with particular activities, objects or topics. A hypersensitivity to particular sounds, including electrical noises such as vacuum cleaners and drills has been found in over 90 per cent of the children, but a physical cause for this hyperacusis has yet to be identified.

These distinctive psychological characteristics, behavioural and emotional difficulties in people with Williams syndrome have been found to persist into adulthood with similar, and sometimes increased frequency (Udwin, 1990). As a result, most adults with Williams syndrome are unable to live independently, despite sometimes low average or borderline cognitive abilities.

Udwin et al. (1987) and others argue that the specific cognitive and linguistic profile and hyperacusis in Williams syndrome constitute a behavioural phenotype. It would be difficult to explain the emergence of this pattern in affected individuals from

different backgrounds, exposed to different environments, except that they would appear to share the same biological disorder.

Fragile X syndrome is the commonest cause of inherited learning disabilities, affecting 1:1250 males and 1:2000 females (Reiss & Freund, 1990). The physical features are an untrustworthy guide to diagnosis, hence the distinctive cognitive and behavioural profile is often more help diagnostically, especially in prepubertal males (Hagerman and Silverman 1991, Turk 1992). Approximately 80 per cent of fragile X males have learning difficulties; which fall primarily into the mild to moderate range, while 30 per cent have severe learning difficulties. Verbal abilities seem to be superior to performance abilities.

Language difficulties range from an absence of speech to mild communication difficulties. A characteristic speech pattern referred to as 'cluttering' is observed, whereby the rate of talking is fast and fluctuating. Dysfluent conversation is common (Dykens, Hodapp & Leckerman, 1994).

The behavioural characteristics of fragile X are striking, and are thought to be problems for 80 per cent of cases. They include hyperactivity, impulsivity and marked concentration problems, even in higher functioning individuals. Irritability, tantrums and aggressive outbursts are also reported to be characteristic, and seem to be precipitated by overstimulation in the environment.

Among males with fragile X there is a spectrum of autistic like behaviours, including gaze aversion (in up to 90 per cent) (Reiss and Freund, 1992). Adult males with fragile X show strengths in daily living skills, compared to their communication and socialization abilities (Dykens et al. 1994).

Females with fragile X show less severe, but similar, learning and behavioural abnormalities, than those seen in males (i.e. Freund et al. 1993). There are also indications of increased prevalence of psychiatric problems in adult females with fragile X.

While there is increasing information on behavioural phenotypes and appropriate interventions for many syndromes, including those described above, very little is known about Smith-Magenis syndrome. This study arose from a need, expressed by parents of affected children, to gather more information on the cognitive and behavioural phenotype of this condition, and appropriate interventions.

1.6 Smith-Magenis Syndrome

Aetiology

Smith-Magenis syndrome is a multiple congenital anomaly and learning disability syndrome caused by an interstitual deletion of chromosome 17p11.2. The first original description of Smith-Magenis syndrome appeared in an article by Smith et al. (1982), and since then over 100 patients with the syndrome have been reported (Allen, Phelan, & Stevenson,1991: Colley, Leversha, Voullaire & Rogers, 1990: Fan

and Funnel, 1994: Finucane, Kurtz, Babu & Scott, 1993a.b. 1994: Fischer, Oswald, Duba, Doczy et al., 1993: Greenberg, Guzetta, de Oca-Luna, Magenis et al., 1991: Hamill, Roberts, Maguire, & Laurence et al., 1990: Lockwood, Hecht, Dowman, Rizkallah, Goodwin et al., 1988: Masuno, Asano, Arai, Kuwahara et al., 1992: Meinecke,1993: Moncla et al., 1991: Patil and Bartley, 1984: Popp, Johnson, Stratton, 1987: Smith et al., 1982, 1986: Stratton, Dobyns, Greenberg, De Sanu et al., 1986: Zori, Lupski, Heju, Greenberg et al., 1993). Physical features may depend on the extent of the deletion.

Prevalence

The majority of individuals with Smith-Magenis syndrome have been identified in the last 5 years as a result of improved cytogenetic banding techniques. The incidence of Smith-Magenis syndrome is estimated to be 1:25,000 births (Greenberg et al., 1991), though this could well be an underestimate. There is an equal sex ratio (Moncla et al., 1991). A diagnosis of Smith-Magenis syndrome is usually made cytogenetically, with detectable deletions of 17p11.2 ranging from 2 to 9 megabases. Trask et al. (1996) suggested that the average deletion spans ~4-5 Mb and is likely to contain a large number of genes, each of which could potentially contribute towards the Smith-Magenis syndrome phenotype.

The cause of the deletion is unknown. The vast majority of the cases occur de novo, which suggests a low recurrence risk for parents. There has been one reported case of vertical transmission of del 17p11.2 from mother to daughter ; the mother was

confirmed to be mosaic for the deletion (Zori et al., 1993). The random parental origin of the 17p deletion in 15 patients with Smith-Magenis syndrome suggests that imprinting does not play a role in the expression of Smith-Magenis syndrome phenotype (Greenberg et al., 1991).

Clinical Phenotype

Greenberg et al. (1996) carried out a multidisciplinary clinical, cytogenetic, and molecular study in 27 children and adults with Smith-Magenis syndrome. They reported the following features in over two-thirds of cases: brachycephaly with a characteristic craiofacial appearance; occular abnormalities; short stature; brachydactyly; a hoarse, deep voice; a history of infantile hypotonia and failure to thrive; speech delay with or without hearing loss; and signs of peripheral neuropathy.

Facial features include midface hypoplasia, prominent forehead, upslanting palpebral fissures, epicanthal folds, broad nasal bridge, downturned mouth with cupid's bow, ear anomalies, and relative prognathism. Allanson, Smith & Greenberg (1998) carried out anthropometric Z-scale pattern profiles on 32 individuals with Smith-Magenis syndrome. The facial pattern profile supported past subjective impressions, with mandibular dimensions consistently exceeding the maxillary counterparts. Craniofacial widths were found to be greater than depths and heights. It has become apparent that several features are age-dependent, these include the prominent forehead, prognathism, brachycephaly, hoarse voice, and opthalmologic findings, in particular high myopia with and without retinal detachment (Finucane et al., 1993a;

Chen et al., 1996). Greenberg et al. (1996) found clinical signs of peripheral neuropathy in approximately 75 per cent of their sample of 27 individuals with Smith-Magenis syndrome, which included decreased deep tendon reflexes, decreased sensitivity to pain or temperature, and reduced leg muscle mass.

Eye abnormalities are common in persons with Smith-Magenis syndrome, and include iris anomalies, microcornea, myopia and strabismus, "apparent" telecanthus, cataracts, and myopia (Chen et al., 1996; Finucane, 1993a). Less consistent features are genital abnormalities, up-slanting palpebral fissures, Brushfield spots and abnormal palmar creases. Finucane et al. (1993) have described strabismus, high myopia and retinal detachment as less common features. They suggest that the combination of high myopia, self-injurious head banging, aggression and hyperactivity means that individuals are particularly prone to retinal detachment. Other findings include cardiac defects (37 per cent); renal abnormalities (35 per cent); thyroid abnormalities (29 per cent); seizures (11-30 per cent); abnormal EEG without seizures (21 per cent); forearm abnormalities (16 per cent); and facial clefts (9 per cent).

Genetics

DNA markers linked to the gene for Charcot-Marie-Tooth disease type 1A (CMT1A), a dominantly inherited peripheral neuropathy, also map to 17p11.2 (Lupski et al., 1991; Patel et al., 1990). Moncla et al. (1993) have suggested that an unstable region, located between the Smith-Magenis syndrome locus and CMT1A, could be a 'hot

spot' for rearrangements leading proximally to SMS microdeletions and distally to CMT1A duplications. Chevillard et al. (1993) have proposed a candidate gene. Greenberg et al. (1991) showed five markers to be deleted in almost all of the 31 patients with Smith-Magenis syndrome investigated. So far 12 genes have been localized to the Smith-Magenis region of 17p11.2, but their exact role in Smith-Magenis syndrome is far from clear.

Cognitive characteristics

There have been very few systematic studies investigating the cognitive and behavioural phenotype of Smith-Magenis syndrome (Greenberg et al., 1996: Dykens, Finucane & Gayley, 1997: Dykens & Smith, 1998: Smith et al., 1998). So far the evidence suggests that all individuals with Smith-Magenis syndrome have some degree of learning disabilities. On average they show Moderate levels of learning disability (IQ 50-70), with speech and language delays. Greenberg et al. (1996) found that IQ scores spanned across the Mild, Moderate and Severe ranges, with most falling in the moderate range. Dykens (1997) examined the cognitive profiles in 10 children/adults with Smith-Magenis syndrome, and found relative weaknesses in sequential processing and short-term memory and relative strengths in long-term memory and perceptual closure.

Speech delay (with or without associated hearing loss) occurs in 96 per cent of cases (Smith et al., 1998). Receptive language skills tend to be generally higher than expressive language. The use of sign language has been found to be extremely

useful along with speech and language therapy (Smith et al., 1998), and helps to reduce the levels of frustration linked with the expressive language problems.

Behavioural characteristics

There are very few studies of the behavioural characteristics associated with Smith-Magenis syndrome. Moreover, the studies that have been undertaken are characterized by small sample sizes and failure to use standardized or systematic methodologies.

The studies undertaken to date have described aggression, temper tantrums, impulsivity, outbursts, repetitive behaviours, attention deficits, and attention seeking behaviours in more than 60-80% of individuals (Dykens et al., 1997; Greenberg et al., 1996; Smith et al., 1986).

Self-injurious behaviours have been observed in children as young as 18 months, and can be extreme, including head banging, wrist biting, skin picking, onychotillomania (pulling out of finger and toe nails) and polyembolokolamania (insertion of foreign objects into bodily orifices) (Greenberg et al., 1991), although no study has used systematic investigation to obtain this information. Individuals with Smith-Magenis syndrome appear to show an insensitivity to pain, and may cause injury to themselves by persistent picking or biting or from an uncontrollable rage. McNaught and Turk (1993) describe one case where parents were reported to social services for suspicion of child abuse because of their child's self-injurious

behaviours. This child was taken into care with a putative diagnosis of deprivation and neglect, before the correct diagnosis of Smith-Magenis syndrome was made, with the accompanying recognition that the self-injurious behaviour is characteristic of the syndrome (McNaught and Turk 1993).

A characteristic spasmodic upper-body squeeze, or "self-hug" has also been described in Smith-Magenis syndrome (Finucane et al., 1994). This behaviour was observed to be more prevalent in the young children and adolescents than in the adults, seemed quite involuntary and was almost tic-like in quality. It was described to occur when individuals are happy or excited. Dykens et al. (1997) described a stereotypic behaviour involving hand licking and page flicking.

Stratton et al. (1986) and McNaught and Turk (1993) have found that some children with SMS fulfil the diagnostic criteria for **autism**, however, there is no information on the prevalence of autism in this population.

Reviewing several earlier studies, de Riijk-van Andel et al. (1991) report a 100% prevalence of **hyperactivity** and a 69% prevalence of self-mutilation, but again this information had not been gathered systematically. Studies included in the review report cases spanning the child to adult range, and include small samples (maximum n=9), (Smith et al., 1986; Stratton et al.,1984; Patil et al., 1984; Stallard et al., 1984; Rao et al., 1985; Viera et al., 1985; Hamill et al., 1988).

Sleep disturbance is of particular importance and was identified in 65 to 100 per cent of cases by Smith et al. (1998), who examined the sleep behaviours of 39 individuals with Smith-Magenis syndrome ranging from 1.6 to 32 years of age. The sleep problems identified included difficulties falling asleep, shortened sleep cycles, frequent and prolonged nocturnal awakenings, excessive daytime sleepiness, daytime napping, snoring, and bed wetting.

There is no study published as yet on the physical and clinical phenotype of SMS that uses a control group. The current study aims to expand on anecdotal reports of the behavioural phenotype. The focus of the study has been to look for internal consistency of behaviours within the study sample. Where possible, previously published comparison data on the prevalence of characteristics has been taken from studies on both the general learning disabilities populations and those disorders where there is a diagnosable biological cause.

Parent support groups for the various syndromes are often approached as a potential source of participants in studies on behavioural phenotypes. This was also the case for this study. Although there are advantages in drawing a sample from such groups, especially because the parents/carers are often highly motivated to take part in research, there are also disadvantages. For instance, the sample might not be representative of the entire population under investigation. It is a possibility that these cases have come to the notice of services, and then of parent support groups,

because they are at the extreme end of the behavioural phenotype continuum. As such findings from studies using such samples need to be treated with caution.

The limited information, and small sample sizes of studies undertaken to date, limit the conclusions that can be drawn about cognitivie and behavioural characteristics associated with SMS. The present study had the following aims:

1.7 Aims and Hypotheses of the study

Aims

1. To obtain systematic information on the cognitive abilities, educational needs and behavioural difficulties of children with Smith-Magenis syndrome.
2. To examine factors associated with severity of behavioural difficulties in this group, including level of cognitive ability and age.
3. Given the high levels of behavioural difficulties reported to be associated with Smith-Magenis syndrome, to gather information about effective interventions for managing maladaptive behaviours in individuals with Smith Magenis syndrome, which can be shared with parents, other carers and teachers.
4. To raise awareness of the condition amongst professionals who may come into contact with children with Smith-Magenis syndrome.

The **hypotheses** of the study are as follows:

1. There is a constellation of particular behaviours and cognitive characteristics which commonly co-occur and are strongly associated with an interstitial deletion on the short arm of chromosome 17 (Smith-Magenis syndrome), and which differentiates individuals with this chromosomal deletion from other groups of children with learning difficulties. In particular that:

i. Children with Smith-Magenis syndrome have a distinctive pattern of cognitive abilities not shared by other groups of children with learning disabilities and other genetic syndromes.

ii. There are higher rates of sleep disturbance, self-injurious behaviour and hyperactivity in children with Smith-Magenis syndrome compared with other groups of children with learning disabilities matched for cognitive level.

The present study is a preliminary investigation aimed at a systematic description of the cognitive and behavioural characteristics of children with Smith-Magenis syndrome, as a step towards establishing whether a behavioural phenotype exists in this population. The findings will be compared to the current literature on the cognitive abilities and behavioural characteristics found in other syndromes and in the population of individuals with learning disabilities in general. Given the limited number of affected children in the country, and the limited information about the syndrome, parents and teachers are eager for guidelines on effective management and

educational strategies. Therefore, a further aim of the study is to pool the information in order to produce guidelines and advice booklets for parents and teachers of children with Smith-Magenis syndrome, with information concerning appropriate ways of managing their cognitive, educational and behavioural difficulties.

The British SMS parent support group has expressed an urgent need for more information about the syndrome and effective strategies for managing the maladaptive behaviours associated with the condition. At present the syndrome is underdiagnosed; by raising its profile and highlighting information about the cognitive and behavioural characteristics associated with the syndrome, the study will hopefully contribute to better recognition of Smith-Magenis syndrome.

2 METHOD

2.1 PARTICIPANTS

Smith-Magenis Syndrome

The participants were drawn from two sources: membership of the Smith-Magenis syndrome Parent Support Group (UK), and Regional Clinical Genetics Centres throughout the UK. These centres and the Support Group distributed letters and consent forms (see Appendix 1) to all the parents of all children with SMS aged between 6-16 years who were known to them. Approximately 35 families were contacted via the Parent Support Group, of who16 responded and gave consent for their children to be included. A further 13 positive responses were received from parents via the Clinical Genetics Centres. All of the participants had documented evidence of SMS, in the form of a letter from a Consultant Clinical Geneticist verifying the presence of the SMS gene deletion.

Comparison Group

Where possible comparisons were made with findings from the literature on sleep disorder, self-injurious behaviours, aggression, hyperactivity and autism in both general learning disability populations and in samples of children with specific biological disorders.

2.2 PROCEDURE

Ethical approval for the study was sought and obtained through St.George's Healthcare NHS Trust Local Research Ethics Committee.

Parents who returned consent forms agreeing to take part in the study were contacted by telephone, and consent forms asking for permission to contact the child's school were then sent. Parents were interviewed in their own homes, and the teachers at their schools, at a time convenient to them. Interviews and assessments took place between May 1997 and June 1999, with the interviewer traveling around the United Kingdom to meet with, assess and interview the children and their carers.

In most cases the assessments and interviews were carried out on consecutive days. In some cases the time between assessment and interview was greater (but not more than one week) if the child was in residential accommodation a long distance from the parents.

The semi-structured interview with parents took between 3-4 hours to complete, and parents were encouraged to take breaks if they wished. The semi-structured interview with teachers took approximately 1 hour to complete. Prior to the interview, parents and teachers were reminded of the aims of the study, and reassured that the information they gave would remain confidential. Prior to the interviewer's visit a copy of the clinical geneticist's letter confirming the diagnosis of Smith-Magenis syndrome had been requested. These were obtained when parents were interviewed. Parents were given the General Health Questionnaire, and Developmental Behaviour Checklist - Parents, and teachers were given the Developmental Behaviour Checklist-

Teachers, to fill in and return to the interviewer in the stamped addressed envelope provided.

Assessment of the child's intellectual functioning and attainment took between 1-2 hours. Before beginning the assessment the participants were asked whether they wished to participate, and thus were reminded that they could take a break or end the assessment whenever they wanted.

2.3 MEASURES

2.3.1 Cognitive and Educational Abilities

General intellectual ability and educational attainment were assessed using the following measures:

(i) Wechsler Intelligence Scale for Children (III) (WISC III) (Wechsler, 1992)

The WISC-III was used to assess intellectual abilities and general cognitive functioning. The scale consists of 2 subscales providing a Verbal and Non-verbal IQ score, from which a Full Scale IQ may be derived. The scale is known to have high levels of reliability and validity. Subtests have been shown to have an average reliability of 0.96r. The scale has been shown to have criterion-related validity through satisfactory correlations with the WISC-R, and a Full Scale IQ score correlation of 0.89.

(ii) Wechsler Objective Reading Dimensions (WORD) (Wechsler, 1993)

The WORD assesses literacy skills by yielding scores in three areas of attainment: Basic Reading, Spelling, and Reading Comprehension. It has average split-half reliabilities of 0.95 (Basic Reading), 0.92 (Spelling) and 0.91 (Reading Comprehension), and test-retest reliabilities of 0.94 (Basic Reading), 0.94 (Spelling), and 0.85 (Reading Comprehension). The WORD has satisfactory construct, content, and criterion-validity (Rust, Golombok & Trickey, 1990; Roid, Twing, O'Brien & Williams, 1992).

2.3.2 Semi-Structured Interview with Parents

Measures of intellectual abilities and attainment were obtained during an assessment with participants with Smith-Magenis syndrome. Semi-structured interviews conducted with parents and teachers were devised incorporating open-ended questions about specific behavioural problems and management strategies specifically addressing the Smith-Magenis syndrome group, and standardised instruments.

The semi-structured interview included questions about the following areas:

(i) **Demographic details:** demographic information included the age, sex, and birth order of the child participant, parental marital status and the socio-economic status of the family, coded according to the occupation of the chief wage earner in the family. The Standard Occupational

Classification (HMSO, 1995) was used to establish categories based on occupation.

(ii) **Diagnosis of Smith-Magenis syndrome:** age at diagnosis, satisfaction with diagnosis, what additional information parents would have liked at the time of diagnosis.

(iii) **Health history and medication:** details of medication participants were taking, current and past health problems, their severity, type and effectiveness of treatments. Parents were further asked about specific eating problems their child with Smith-Magenis syndrome might have.

(iv) **History of schooling**: including nurseries and schools attended, as well as details about the current school, appropriateness and problems encountered. Whether participants had a Statement of their Special Educational Needs and the appropriateness of the Statements.

(v) **Social and emotional functioning**: parent's were asked about the social and emotional functioning of their child with Smith-Magenis syndrome, including anxiety, peer and adult relationships, leisure activities, preoccupations and obsessions and sexual behaviour.

(vi) **Impact on the family:** carers were invited to reflect on the way caring for an individual with Smith-Magenis syndrome had affected their family life and social life, and to describe their current and future concerns with regard to their son or daughter.

(vii) **Participants' current and previous involvement with mental health services and support agencies:** parents were asked whether they have or

had contact with one or more mental health professional, who they had seen and how often, reasons for referral, details of any treatment implemented, its effectiveness and satisfaction with the service. Parents were also asked about the extent of the families' contact with Social Workers and about the help and advice they had received from social services and voluntary organizations, for example in obtaining access to respite care provision. In addition, they were asked about the amount and quality of respite care available to them.

(viii) **General concerns:** which aspects of behaviour parents found most difficult to manage, and what they thought was the greatest challenge in terms of rearing a child with Smith-Magenis syndrome.

(ix) **Areas of strength:** the positive characteristics of the child with Smith-Magenis syndrome; including whether they are affectionate, caring, have a good sense of humour, and particular skills and abilities the child has.

In addition to the above areas, the following standardized measures were also used:

- **Schedule of Handicaps, Behaviour and Skills (HBS) (Wing, 1980)**

The HBS is a semi-structured interview schedule developed for use with children and adults who have difficulties with reciprocal social interaction and/or communication. It describes the participant's level of functioning and present behaviour. Items divide into those indicating developmental stage reached (e.g. use of language, ability to walk, dress and feed) and those concerning abnormal or difficult behaviour (e.g. sleep

disturbance, echolalia, stereotyped movements, physical aggression). The Vineland Social Maturity Scale (Doll, 1935, 1965) can be completed from the information obtained on the HBS. Discrete diagnoses of DSM-III-R typical autism and pervasive developmental disorder can be made using the accompanying Autistic Disorders Diagnostic Checklist (Wing, 1987). The schedule has been used in large-scale community studies and satisfactory reliability has been obtained (Wing and Gould, 1979).

- **Developmental Behaviour Checklist (DBC) (Einfeld & Tonge, 1989)**

The DBC was used to screen for behavioural and emotional problems. This scale was developed specifically to assess behavioural and emotional problems in children and adolescents with developmental disabilities, and as such was considered an appropriate screening instrument for the present sample. The DBC is a 96 item instrument which is completed by parents (DBC-P) and teachers (DBC-T). Each behavioural description is scored on a 0,1, 2 rating where 0= 'not true as far as you know', 1= 'somewhat or sometimes true', and 2= 'very true or often true'. Both versions of the checklist were used: the Parent/Carer Version (DBC-P), and the Teacher's Version (DBC-T). The questionnaires were left with the families and teachers to complete together with a stamped addressed envelope.

The cut-off of 46 on the Total Behaviour Problems score has been shown to predict what 'experts in diagnosing psychiatric disorders and learning disabilities' rate independently as "major behavioural and emotional disturbance" or a "definite

psychiatric case". The specificity and sensitivity for the Developmental Behaviour Checklist is high (i.e. 92%)

Einfeld & Tonge (1994, 1995) presented the following summary of the psychometric properties of the Developmental Behaviour Checklist: interrater reliability parent-parent (n=42), intraclass correlation (ICC) = 0.80, 99% confidence interval (CI) = 0.59 to 0.90, and teachers-aides (n=110), ICC = 0.60, 99% CI =0.42 to 0.74; internal consistency (total score, n= 1, 093) = 0.941; clinician-parent agreement-item meaning (n=70), 97%, criterion group validity (n=70), t=7.783, p<0.001; concurrent validity DBC/Adaptive Behaviour Scale (Nihira et al., 1975) (n=40),0.86, p,.001, DBC/Scales of Independent Behaviour Problems section (Bruininks et al., 1984) (n=40),0.70, p< .001, and clinician ratings/DBC (n=70), r=0.81, p<0.001; readability of the DBC, Flesch Index = 76.2.

The DBC comprises of 5 subscales: Disruptive, Self-absorbed, Language disturbance, Anxiety, Autistic relating and Antisocial.

- **The Modified Overt Aggression Scale** (Kay, Wolkenfield & Murrill, 1988)

This scale was used to assess the level of aggression displayed by the children in the present study. It is a revision of the Yudofsky et al. (1986) method that was developed to assess the nature and prevalence of aggression in a psychiatric population. The behavioural checklist consists of 16 items that are grouped according to four categories of aggressive behaviour (verbal aggression, aggression against

property, autoaggression, and physical aggression). Each item has a five-point rating system (0-4) that represents increasing levels of severity; the instrument provides definitions for each category of aggression; zero represents absence of such behaviours. It also has a weighted total score that reflects the overall seriousness of aggression. The following is an example of one of the categories:

Aggression against property:

0. *No aggression against property*
1. *Slams door angrily, rips clothes, urinates on floor*
2. *Throws objects down, kicks furniture, defaces walls*
3. *Breaks objects, smashes windows*
4. *Sets fires, throws objects dangerously*

The internal reliability of the MOAS is supported by the consistency of the scale scores across psychiatric units. Good inter-rater reliability of the total score was found between a psychologist and social worker (Pearson r =0 .85, $p<.001$ for a secure care unit, and Pearson r = 0.94, $p<.001$ for an admissions unit) (Kay et al. 1988). The scale accurately distinguished patients from three psychiatric units that had different expected rates of aggression.

As the scale was devised for use with psychiatric patients there are no norms for children. However, it was felt that the scale would provide a useful measure of aggression for the present sample.

Answers to the questions about aggression were classed as follows: 'occasionally, once a week or less, 'regularly, every few days', 'once a day', or 'more than once a day'. Open-ended questions were asked about the causes of outbursts of aggressive behaviour. Owing to the suggested high rate of self-injury in SMS, these behaviours were looked at in detail. A list of 10 types of self-injury was read out to the parent who rated frequency and severity of each behaviour. Parents were also asked to identify triggers for the various behaviours . Open-ended questions were asked about both formal and informal strategies that were found to be helpful in managing aggressive and self-injurious behaviours.

- **Overactivity and Attention Difficulties**

The Hyperactivity Scale from the **Parental Accounts of Children's Symptoms** (PACS) (Taylor and Schachar, 1993) was used in the present study. The questions are semi-structured and address the following areas; attention span (as expressed by the duration of an activity), activity level/restlessness (shown by getting up and down or moving around during an activity) and fidgetiness (an inability to keep still while sitting down). These dimensions are examined in a number of age appropriate situations like watching television, reading, solitary play, play with other children, mealtimes, shopping trips and family outings. Ratings are made for severity, on a four point scale (0-3), and frequency in the previous week. Taylor et al. (1986) found inter-rater reliability values ranging from 0.92 to 0.95. The internal consistency of the hyperactivity scale was 0.89.

In addition, parents were asked about effective strategies for managing the overactivity and concentration difficulties exhibited by their children, as well as questions regarding medication for such problems.

- **Sleep Difficulties**

Parents were asked detailed questions about their child's sleeping pattern and any difficulties in this area. Questions were included from a modified version of a specially developed semi- structured interview based on items in a sleep questionnaire for parents developed by Simonds and Parraga (1982). The questionnaire covers the main categories described in the International Classification of Sleep Disorders (1990). It is a two part questionnaire: Part 1 is concerned with the quantity and quality of the child's sleep. Items require yes/no answers or a reply in hours (e.g. 'Does your child generally sleep soundly?' and 'How many hours does you child sleep?'). Part 2 of the questionnaire is concerned with identifying sleep disorders. Four broad categories of disorders are covered and expressed in lay language; disorders of initiating and maintaining sleep; parasomnias; sleep-related breathing problems; and daytime sleep-related features (i.e. drowsiness or increased activity). Parents rated the frequency of occurrence of the 25 behaviours on a five-point scale of: (1) 'Never' (2)'Less than once a month' (3) 'Two or four times a month' (4) 'Many times a week'; or (5) 'Daily'.

Part 1 of the questionnaire also yields information on settling, night waking and early waking, but to allow for the use of standardized definitions of severe settling and

night waking difficulties (Richman & Graham, 1971), two further questions were added: 'How many times per week do you have difficulty settling your child?' and 'How long does it take to resettle your child if they wake in the night?'. Open-ended questions were included for more qualitative information about the settling and waking problems (e.g. 'If your child will not go to bed or settle to sleep what do you do about it?').

Parents were asked who attended to their child (e.g. mother, father, siblings), if they were disturbed during the night, , and whether they thought they received enough sleep themselves.

Test-retest reliability for individual items over a two week period was reported by the authors to range from 0.83 to 1.00. The scale was also found to have high face validity; as there were 19r values of 1.00 and 4 between 0.90 and 0.99.

- **Impact on the Family/ Parental Stress**

Parents were administered the **General Health Questionnaire** (GHQ) (Goldberg & Hillier 1979; Goldberg & Williams 1988), which is a self-administered screening instrument used to detect psychiatric disorders in the general population. The questionnaire covers two major areas: inability to continue to carry out one's normal 'healthy' functions, and the appearance of new phenomena of a distressing nature. Each of the 60 items consists of a question asking whether the respondent has

recently experienced a particular symptom or item of behaviour on a scale ranging from 'less than usual' to 'much more than usual'. The GHQ was found to have good split-half reliability (0.95), test-retest reliability (0.76), specificity (87.8%) and sensitivity (95.7%) (Goldberg & Williams 1988).

The questionnaire was left with the parents who were asked to complete and return it by post in a stamped addressed envelope.

2.3.3 Semi-Structured Interview with Teachers

Interviews with teachers took approximately 1 hour. Teachers were asked questions about the child's academic abilities; including areas of strength and weakness, concentration difficulties, and the appropriateness of the educational placement. They were also asked open-ended questions about aggressive behaviours, and the causes of outbursts of aggression. Open-ended questions were asked about teaching strategies found to be helpful. Questions addressing areas of social functioning were administered, which included questions about social responsiveness (incorporating questions about interactions with peers and adults, and bullying behaviour).

2.4 DATA ANALYSIS

Summary and descriptive statistics were performed on the data. Nonparametric and parametric statistical analyses were performed as appropriate, on interval and ordinal data to test for associations between: age, gender, severity of aggressions and self-injury, severity of sleep difficulties and diagnosis of autism. Comparisons were made

between the frequency of behavioural characteristics in the SMS group and other groups of children with learning disabilities from the literature. Given the limited sample sizes across studies along with the use of different methodologies and measures, these comparisons relied on observable trends in reported frequencies of behaviours rather than on statistical analysis.

3 RESULTS

3.1 DEMOGRAPHIC DATA

Demographic information on the participants is presented in Table 1.

Table 1. Demographic information

	% Total group (n= 29)	
	n	**(%)**
Gender		
Male	13	(44.8)
Female	16	(55.2)
Age (months)		
Mean (SD)	116.8 (34.7)	
Range	75-192	
Socio-economic status (based on occupation of main breadwinner)		
1 - Professional	2	(6.9)
2 - Managerial & technical	12	(41.4)
3 - Skilled occup., non-manual	6	(20.7)
4 - Skilled occup., manual	4	(13.8)
5 - Partly skilled occupation	0	(0)
6 - Unskilled occupation	1	(3.4)
7 - Housewife/Unemployed	4	(13.7)
Birth order		
Only child	4	(13.8)
Youngest	7	(24.1)
Middle	3	(10.3)
Oldest/older	15	(51.7)

As Table 1 shows, the gender ratio was roughly equivalent. Socio-economic status of the main breadwinner/head of household, was determined using the Standard Occupational Classification (Government Statistical Service, Second Edition, 1995) and is weighted towards the higher end with 12 families (41%) falling into the Managerial & Technical category; this is not unexpected given previous findings that

members of parent support groups are predominantly middle class. Twenty-five (86%) of the parents were married/living together, 4 (14%) were single mothers. The majority of children (21 (72%)) were aged 6-11 years, with the remainder aged between 12-16 years. Three quarters of the sample were living at home with their families. Parents were asked to rate their satisfaction with these living arrangements. The results suggest a high degree of satisfaction (see Table 2).

Table 2. Living arrangements

	% Total group (n= 29)	
	n	**(%)**
Living arrangements		
At home with family	22	(75.9)
Residential/boarding school	7	(24.1)
Satisfaction with living arrangements		
Very satisfied	23	(79.3)
Mixed, but mostly satisfied	4	(13.8)
Mixed, mostly dissatisfied	2	(6.9)
Very dissatisfied	0	(0)

Parents were asked about the types of school the children were attending. As can be seen in Table 3, 90% of the sample (26 children) were in Special Schools, mostly for children with Severe Learning Disabilities or Moderate Learning Disabilities. Only 3 children (10%) were in mainstream school, one of whom was in a class for children with special needs, and the other two received between 17-18 hours of help from a learning support assistant per week. The teachers of both these children felt that full-time support for them, covering break-times, was required.

Table 3. Type of school attending

	% Total group (n= 29)	
Type of school	**N**	**(%)**
Ordinary	2	(6.9)
Remedial/special class in ordinary school	1	(3.4)
Severe Learning Disabilities	10	(34.5)
Moderate Learning Disabilities	6	(20.7)
Mixed ability school	3	(10.3)
Residential/boarding (Learning Disabilities)	5	(17.2)
Residential/boarding (Autism)	1	(3.4)
Language unit	1	(3.4)

When asked to rate their satisfaction with the education their children were receiving; 23 parents (79%) stated that they were 'very satisfied', or 'satisfied with most aspects' of their child's schooling. The most common reasons for dissatisfaction were that their child had better communication skills compared to the others in the school, and was therefore not able to develop these skills further, as well as becoming frustrated when others could not respond (4), that the class sizes were too big(1), and that the ratio of teacher/assistant to child was not high enough(1).

Diagnosis

Information was gathered on age at diagnosis of Smith-Magenis syndrome, and carers were asked to rate their satisfaction with the information they had received at the time of diagnosis, and whether they felt that they had a satisfactory understanding of the syndrome. Age at diagnosis ranged from 1 month to 14 years. The mean age at diagnosis was 4.8 years and the median was 4 years. Eighteen parents (62%) said

they were satisfied with the way the diagnosis had been given; the remainder expressed dissatisfaction, and the most common reason was the lack of information available on Smith-Magenis syndrome at the time they received the diagnosis.

Medical history

Parents were asked about any health difficulties their children experienced in the past and/or currently, as well as the severity of the condition. Ninety percent of the sample (26 children) were reported to have health problems at the time of interview, and 97% (28 children) had had health problems in the past. The most common of these were ear infections, dry skin, and chest infections (see Table 4). In addition, 86% of the children (25 children) were reported by their parents to have reduced sensitivity to pain/heat/cold. Seventy-six percent of parents (22 parents) reported continuing feeding difficulties in their children, in particular with chewing solid foods (especially meat), and a third reported mild to moderate scoliosis. Epilepsy was reported in only 10% of cases (3 children).

Table 4. Medical problems

Medical problems	% Total group (n= 29)	
	N	(%)
Ear infection		
No	7	(24.1)
Mild (no treatment required)	1	(3.4)
Moderate (requires regular treatment)	9	(31.0)
Severe (requires regular hospitalization)	3	(10.3)
In past	9	(31.0)
Chest infection		
No	11	(38.0)
Mild (no treatment required)	5	(17.2)
Moderate (requires regular treatment)	10	(34.5)
Severe (requires regular hospitalization)	0	(0)
In past	3	(10.3)
Heart problems		
No	22	(76.1)
Mild (no treatment required)	2	(7.1)
Moderate (requires regular treatment)	1	(3.4)
Severe (requires regular hospitalization)	1	(3.4)
In past	3	(10.3)
Scoliosis		
No	19	(66.0)
Mild (no treatment required)	6	(21.0)
Moderate (requires regular treatment)	3	(10.3)
Severe (requires regular hospitalization)	1	(3.4)
In past	0	(0)
Epilepsy		
No	26	(90.0)
Mild (no treatment required)	0	(0)
Moderate (requires regular treatment)	3	(10.3)
Severe (requires regular hospitalization)	0	(0)
In past	0	(0)
Dry skin		
No	8	(28.0)
All over body	7	(24.0)
Just on hands and feet	14	(48.0)
Sensitivity to pain/heat/cold		
Normal reaction	1	(3.4)
Reduced sensitivity	25	(86.2)
Increased sensitivity	3	(10.3)

3.2 COGNITIVE ABILITIES AND ATTAINMENT

3.2.1. Cognitive Abilities

Cognitive assessments were carried out on all 29 participants, using the Wechsler Intelligence Scale for Children-III (WISC III) (Wechsler, 1992). For 7 children (24%) Full Scale IQs on the WISC-III fell below the basal level of 40. The IQs of the remaining 22 children ranged from 42 to 60, with a mean of 48.5, and a median of 48.0. The distribution of Full Scale IQ scores is presented in Table 5. Considering performance on the WISC-III Verbal and Performance Scales separately, it was found that 9 children (31%) failed to score above the floor of the Verbal Scale, and for the rest Verbal IQs ranged from 48 to 62, with a mean of 53.9, and a median of 54.0. On the Performance Scale 11 children (38%) scored below the floor of the test, and the remaining children obtained IQs ranging from 47 to 64, with a mean of 52.6, and a median of 50.0. The sums of the scaled scores on the WISC-III Verbal and Performance Scales were compared statistically. There was no significant difference between summed scores on these two scales ($t = 0.85$). Since significant numbers of children scored below the floor of the WISC-III on one or both scales, the comparison was repeated including only those children with Full Scale IQs of 40 or above. Once again, there was no significant difference between scores on the two scales ($t = 0.85$).

Table 5. Distribution of IQ scores on the WISC-III

	Full Scale		Verbal Scale		Performance Scale	
IQ	n	(%)	n	(%)	n	(%)
40-45	13	(44.8)	-	-	-	-
46-50	9	(30.9)	15	(51.7)	21	(72.3)
51-55	4	(13.7)	8	(27.5)	2	(6.8)
56-60	3	(10.3)	3	(10.3)	4	(13.7)
61-65	-	-	3	(10.3)	2	(6.8)
n	29		29		29	

The children's performance on the individual WISC-III subtests was also examined. Table 6 presents the mean scaled score and standard deviation for each of the 10 subtests.

Table 6.
Mean scaled scores and standard deviations of the subtests of the WISC-III

Verbal Scale	Mean	SD	Performance Scale	Mean	SD
Information	2.48	1.77	Picture Completion	2.97	2.58
Similarities	1.72	1.19	Coding	1.52	0.91
Arithmetic	1.14	0.44	Picture Arrangement	1.69	1.37
Vocabulary	1.76	1.68	Block Design	1.24	0.58
Comprehension	1.21	0.56	Object Assembly	1.41	1.09

A repeated measures ANOVA was carried out on the data provided by the WISC-III subtest scores. This yielded a highly significant variation between subtest scores ($F= 21.852$ for df 1 & 28; $P<.001$). Paired comparisons were then made between individual subtest scores using the Tukey HSD test. The mean subtest difference to be significant at the 5% level was calculated to be 1.88, and as can be seen from Table 6

the largest difference in mean subtest scores, that between Arithmetic and Picture Completion, is 1.83 which fails to reach significance at the 5% level.

There was no significant association between age and IQ (based on the sum of scaled scores) (Pearson r = -0.18, p=0.36), nor between gender and IQ (chi-square = 0.03, p = 0.86).

3.2.2 Attainment in reading and spelling

Sixteen (55%) of the children were able to obtain some score on the Wechsler Objective Reading Dimensions (WORD) (Wechsler, 1993) for Basic Reading and Reading Comprehension, and 9 (31%) for Spelling (see Table 7). Eight (28%) children obtained scores on all three tests, while 8 (28%) were readers but non-spellers, and one child obtained some score on the spelling test but not on the reading test. The 16 readers had a mean chronological age of 10 years 10 months (range 7 years 1 month – 16 years), and they obtained a mean age for Reading Accuracy of 7 years 2 months (range 6 years – 11 years 8 months) and a mean age for Reading Comprehension of 6 years 4 months (range 6 years – 7 years 7 months). The Reading Ages for Accuracy were significantly higher than the Reading Ages for Comprehension (t = 2.47, p=0.02). The 9 spellers had a mean chronological age of 11 years 2 months (range 7 years 1 month – 16 years), and obtained a mean Spelling Age of 7 years (range 6 years – 7 years 8 months).

Table 7. Age-equivalent Scores on Attainment Tests

Test (*)	Mean attainment age	
	Years (SD)	Range
Basic Reading (n = 16)	7.2 (19.3)	6.0 - 11.8 years
Reading Comprehension (n = 16)	6.4 (5.8)	6.0 - 7.7 years
Spelling (n = 9)	7.0 (15.5)	6.0 - 7.8 years

*No. of participants able to score

Table 8 presents comparisons between the readers and non-readers, and spellers and non-spellers, in terms of their ages and IQ (scaled scores) using independent samples tests. As expected, the children who were reading and spelling were significantly older when compared with the children who obtained no scores on the reading and spelling tests. In addition, children who obtained some score on the spelling test had significantly higher full scale, and verbal scale IQs (scaled score), compared to non-spellers. In contrast, the readers and non-readers did not differ significantly in terms of IQ.

Table 8.
Ages and IQ (scaled scores) of the readers versus non-readers, and spellers versus non-spellers

	Readers (n=16) Mean (SD)	Non-readers (n=13) Mean (SD)	t/p		Spellers (n=9) Mean (SD)	Non-spellers (n=20) Mean (SD)	t/p
Age	136.8 (30.8)	92.2 (21.1)	t =4.44 p=<0.00		138.9 (35.8)	106.9 (30.1)	t=-2.50 p=0.02
Full Scale IQ (scaled scores)	18.6 (9.4)	15.3 (5.4)	t=-1.13 p=0.27		21.3 (9.9)	15.3 (6.3)	t=-2.01 p=0.05
Verbal IQ (scaled scores)	8.8 (4.2)	7.7 (3.0)	t=-0.8 p=0.43		10.3 (4.4)	7.4 (3.0)	t=-2.1 p=0.04
PerformanceIQ (scaled scores)	9.8 (5.8)	7.6 (3.0)	t=-1.23 p=0.22		11.0 (6.2)	7.9 (3.8)	t=-1.7 p=0.11

Parents were asked whether their child has a Statement of Special Educational Needs. All of the children attending English schools had a Statement. The one child who did not was from Ireland where the Statementing process is not available. The mean age at which children were Statemented was 4 years 4 months (range 3 years – 12 years 7 months).

Special Skills

Parents were asked whether their child showed any special skills or abilities. If a special skill was noted, parents were asked to rate whether, in their judgment, it was better than might be expected from children of similar ages, or whether it was just above what might be expected of the children relative to their general level of ability. As can be seen in Table 9, in 21 children with SMS (72%) memory was rated as superior to that of even children with normal intelligence. In particular, parents described extremely good long-term memory for past events, for example being able to recall the exact detail of what someone was wearing when the child with SMS met them several years ago. Four of the children (14%) were described as having a special skill using computers, and a further 12 (41%) were rated as having computational skills beyond their general developmental level.

Table 9. Special Skills

Skill	Skill better than general developmental level, but not above chronological age		Skill above chronological age	
	n	(%)	n	(%)
Visuospatial (e.g. puzzles)	7	(24)	0	(0)
Computers	12	(41)	4	(14)
Musical	1	(3)	1	(3)
Memory	6	(21)	21	(72)
Drawing	0	(0)	0	(0)

Teaching approaches

Teachers were asked which teaching approaches they found most effective for their pupils with SMS. Table 10 presents these and shows that rewarding/giving praise, 1child to 1 teacher/assistant support, and using the mornings for teaching the most difficult work (as the children tend to concentrate better then) were the most effective techniques. In addition, several teachers reported that being firm and consistent, teaching in short blocks of a few minutes (depending upon the child) with breaks in between, repeating work, and using computers were also useful strategies to aid learning.

Table 10. Effective teaching strategies

Teaching approaches	N	%
1:1 ratio of teacher/child support	12	41
Distraction	3	10
Rewards, praise	19	66
Short teaching blocks(several minutes)	4	14
Firm, consistent structure	5	17
Ignore, time-out	7	24
Repetition of work	4	14
Establishing eye contact	1	3
Visual cues and gestures	1	3
Computers	4	14
Makaton	2	7
Home-school diary of good behaviour	1	3
Small group work	3	10
Most effective learning in the morning	12	41

*For some children more than one approach was used

3.3 SLEEP PATTERN AND DIFFICULTIES

3.3.1 Sleep Problems

One of the aims of this study was to obtain information on the sleep behaviours of children with SMS, and to compare these with the sleep patterns and difficulties exhibited by other groups of children with learning disabilities, so that conclusions about the existence of an emerging behavioural phenotype may be drawn.

All the participants were reported by their parents to exhibit sleep problems; in 5 cases (17%) the sleep problem was rated as 'mild', in 6 cases (21%) as 'moderate' and in 18 cases (61%) as severe. As can be seen in Table 11, difficulties in settling,

night waking and early morning waking were most frequently reported, followed by difficulties in night waking and early morning waking .

Table 11. Type and frequency of sleep problem

	% Total group (n= 29)	
	n	(%)
No sleeping difficulties	0	(0)
Settling only	1	(3)
Waking in the night only	1	(3)
Waking early only	4	(14)
Settling and waking in the night	2	(7)
Waking in the night and waking early	9	(31)
Settling, waking in the night and waking early	12	(41)

Table 12 summarizes the sleep cycle for the sample of 29 children with SMS. The mean wake-up time was 5.00am (range 3.00am-6.30am), with 28% waking up earlier than 5.00am, and mean bedtime (time falling asleep) 8.76pm (range 7.00-10.00pm). Participants woke up twice a night on average, and took an average of 64 minutes to fall back asleep. Twenty-four children took between one and two naps during the day, lasting for an average of 34 minutes each (range 5-60 minutes).

Table 12. Sleep Cycle

	Group total mean n=29	Range
Wake-up time	5.00am	3.00am-6.30am
Bedtime	8.76pm	7.00pm-10.00pm
Number of naps per day (n=24)	1.1	1-2
Length of naps (minutes) (n=24)	34.4	5-60
Number of times up at night	2.32	0-5
Time to fall back to sleep (minutes)	64.6	5-180

Sleep problems tended to be of a long standing nature, starting early in childhood (at a mean age of 15 months, range 0 – 54 months).

(i) ***Night Waking***

- ***Frequency***

All participants were reported to wake at least once during the night, and 27 (93%) woke at least once on a daily basis. Eighteen individual's (62%) woke at least once or twice, but 10 (34%) woke at least 3-4 times a night, if not more. Twenty-seven (93%) of participants were classed as 'sleeping soundly until waking'.

- ***Impact on family members***

Parents were asked about the impact of night waking on the family. Twelve (41%) reported that they did not feel that they had sufficient sleep, and 15 (52%) reported that other members of their family suffered loss of sleep. Despite all children waking during the night, 13 (44%) were able to resettle within between a matter of minutes and half an hour. Eight children (28%) took at least an hour to resettle, and 8 (28%) took 2-3 hours, if not more, to resettle.

(ii) ***Early Waking***

Of those who woke early in the morning (5.00am or earlier), over half did this on a daily basis or every few days, and the remainder did this only occasionally (see Table 13).

Table 13. Frequency of Early Waking

Frequency of early waking	n	(%)
Never	4	(14)
Less than once a month	1	(3)
About once a month	6	(21)
2-4 times a month	1	(3)
Several times a week	4	(14)
Daily	13	(45)

(iii) ***Sleep during the day***

Despite the poor quality sleep exhibited by these children, the majority, 26 individuals (90%) are reported to wake up looking well rested in the morning, with 20 children (69%) usually waking in a 'good' mood. One child was reported as usually waking in a 'bad' mood, and 8 individuals (28%) were described as sometimes waking in a 'bad' mood. About half (48%) of the children sleep during the day, with an additional 8 children (28%) occasionally having a sleep during the day. Parents reported that a further 4 (14%) of children would sleep during the day if they were allowed to. Most commonly parents reported that their child fell asleep in the car/bus returning from school. However, there were reports of children falling asleep whilst eating and in class, especially during the afternoon. Teachers reported that 18 children (62%) seemed tired during the day (especially during the afternoon).

Interestingly, several parents reported that their child tended to fall into a deep sleep immediately after a tantrum. The majority of parents who reported that their child sleeps during the day also stated that if they wake their child up from a nap they tend

to be irritable, whereas if they leave them to wake up themselves they are in a far better mood.

(iv) ***Common sleep problems***

The frequency of other sleep related difficulties are summarized in Table 14.

Table 14. Additional Sleep Related Difficulties

	Frequency	(%)
Wets bed	23	(79)
Snores loudly	13	(45)
Insists on sleeping with someone	12	(41)
Makes quick movements of arms and legs	10	(34)
Talks in sleep	10	(34)
Grinds teeth	9	(31)
Rolls from side to side to get off to sleep	9	(31)
Moves around a lot	9	(31)
Stops breathing	7	(24)
Is frightened to go to bed	5	(17)
Nightmares	5	(17)
Gags, chokes or snorts loudly	4	(14)
Wakes in night screaming in terror	4	(14)
Bangs head	3	(10)
Walks in sleep	2	(7)
Upon waking feels paralysed	1	(3)
Bites tongue	0	(0)

Of the sleep problems presented in Table 14 the most frequently reported problems were bed-wetting (enuresis) and snoring. Other fairly frequent problems, occurring in just under a third of cases, were talking during sleep, grinding teeth, rolling from side to side to get to sleep, moving around a lot during sleep, and making rapid movements with arms and legs. There was a significant association between age and

frequent bedwetting (daily /several times a week); increased age was significantly associated with less bedwetting (Chi-square = 4.39, p=0.04).

(v) ***Natural History of Sleep Problems in Childhood***

According to their parents, 10 participants (34%) reported that their child's sleeping habits had improved between 8-10 years of age. Most felt that this was due to the children being able to entertain themselves better when they did wake up, rather than an actual increase in the amount of time asleep. Three parents felt that the actual time their child slept had increased a little as they got older.

In order to determine if there was any association between age, IQ (sum of scaled scores) and the sleep behaviours, correlations were carried out. Increased age was associated with less severe sleep problems (Spearman rho = -0.33, p = 0.04), and fewer awakenings during the night (Spearman rho = -0.34, p = 0.04). On the other hand, there was no association between IQ (Spearman rho = -0.25,p = 0.18; Spearman rho = -0.14, p = 0.47) and gender (Chi-square = 0.74, p = 0.39; Chi-square = 0.06, p = 0.80) , and severity of sleep problems and number of awakenings at night.

Furthermore, there was no association between age (Pearson r = 0.16,p = 0.40; Pearson r = -0.07,p = 0.74; Pearson r = 0.12,p = 0.53), and IQ (Pearson r = 0.003,p =0.99; Pearson r = -0.01,p = 0.97; Pearson r = 0.11,p = 0.58) on sleep duration, time spent to resettle during the night, and time of waking. In addition, there was no association between gender and sleep duration and time of waking (Chi-square =

0.14, p = 0.71; Chi-square = 0.05, p = 0.82). However, there was an association for gender and time to resettle (Chi-square = 4.89, p = 0.03), with boys taking less time to resettle after waking in the night.

Some of the other sleep behaviours were found to relate to each other. Ratings of increased severity of sleep problems were related to parents feeling as though they were not getting enough sleep (Spearman rho = -0.52, p = 0.004).

3.3.2 Treatment of Sleep Difficulties

Parents were asked if they felt they would currently benefit from professional advice to address their children's sleep problems. Despite the severity of sleep problems, only 11 (38%) responded in the affirmative. Twenty-two (76%) had received professional advice in the past, and of these only 9 (31%) had felt that the interventions were effective (see Table 15).

Table 15. Effectiveness of intervention (n = 29)

Rating	n	(%)
No intervention	7	(24)
Not effective	13	(45)
Somewhat effective	5	(17)
Effective	4	(14)

Parents were asked to give details of professional advice given, its effectiveness, and any additional methods they had found to be effective. As can be seen in Table 16, the most effective interventions used were locking the bedroom and other doors in the house (although several parents had not found this technique useful), allowing the

child to sleep with a parent(s), and to watch television or listen to music, ignoring the child and implementing a firm and consistent approach to returning the child to their bed (although in other cases this was reported not to be effective). Certain strategies were effective for some parents and not for others. Parents also commented that several of the techniques were not consistently effective. Where intervention strategies were successful, it was often the case that the amount of sleep had not increased, rather that the child had become less disruptive during the night and in the early morning, and as such parents and other family members were able to sleep more.

Table 16. Type and efficacy of intervention for sleep difficulties (n=29)

Problem and intervention*	Intervention n	Effective n	Not effective n
Settling and night time waking			
Medication	24	5	19
Cranial osteopathy	2	1	1
Lock bedroom and other doors (including cutting top half of the bedroom door off, high handle, spy hole and alarming house)	15	11	4
Reduce daytime naps and keep awake as long as possible	8	6	2
Reduce noise and light in the bedroom (including the use of black-out blinds)	6	4	2
Remove toys, books, computer from the bedroom	6	4	2
Provide toys, books, computer for the bedroom	4	4	- -
Child sleeps with parent(s)	11	11	- -
Encourage routine (sometimes a routine instilled by the child)	5	4	1
Ignore	10	7	3
Provide less disruptive distractions (i.e. soft toys, magazines)	3	3	- -
Firmly and consistently instruct child to return to bed	9	6	3
Provide an alarm clock	1	- -	1
Positive reinforcement (star charts, rewards etc.)	2	- -	2
Relaxation (tape, scented candles)	4	4	- -
Child falls asleep in lounge	3	3	- -
Watches TV, listens to music	8	8	- -
Child is kept warm	2	2	- -
Read to the child	5	5	- -

* Most children received more than one intervention

Twenty-four of the 29 children participating in the study had been given medication at some time for sleep difficulties; they showed a mixed response (see Table 17). The majority (19) found medication ineffective, while 5 had found it effective. Eight children had tried at least two types of medication to facilitate sleep. Table 17 presents the types of medication taken, its efficacy and side-effects, as reported by the parents.

Table 17. Type, efficacy and side-effects of medication to facilitate sleep

Medication	Effective	Not effective	Side-effects
Melatonin	0	2	
Tegretol	0	2	Headaches
Vallergan	2	8	Very drowsy, made sleep worse
Fenegan	0	7	Sleep worse
Melleril	2	0	Increased appetite
Welldorm	0	2	
Sedative (name unknown)	0	3	Sleep worse
temazepam	0	1	
Imipramine	1	0	

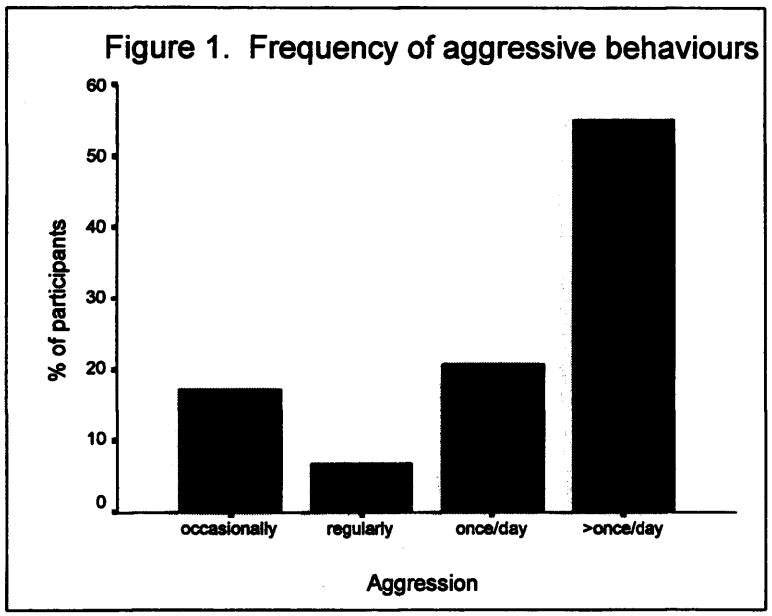
As can be seen, three types of medication were reported to be effective for only 5 children. At the same time, Vallergan, although effective for 2 children, was not effective for 8 children. It must be stressed that in several cases parents took their child off the medication after a very short period of time (often 1-3 days) because their child had either become worse, or was very drowsy the following day. Thus, this data cannot be considered to be based on systematically administered trials of medication. Imipramine was prescribed for night-time incontinence for one child, and concomitantly improved night waking.

3.4 AGGRESSIVE BEHAVIOUR

3.4.1 Description of Aggressive Behaviours

- ***Prevalence***

Parents were asked whether their child exhibited each of four main categories of aggressive behaviour (verbal and physical aggression, aggression against property and self-injury). As can be seen in Figure 1, the overall frequency of aggressive behaviours is high.

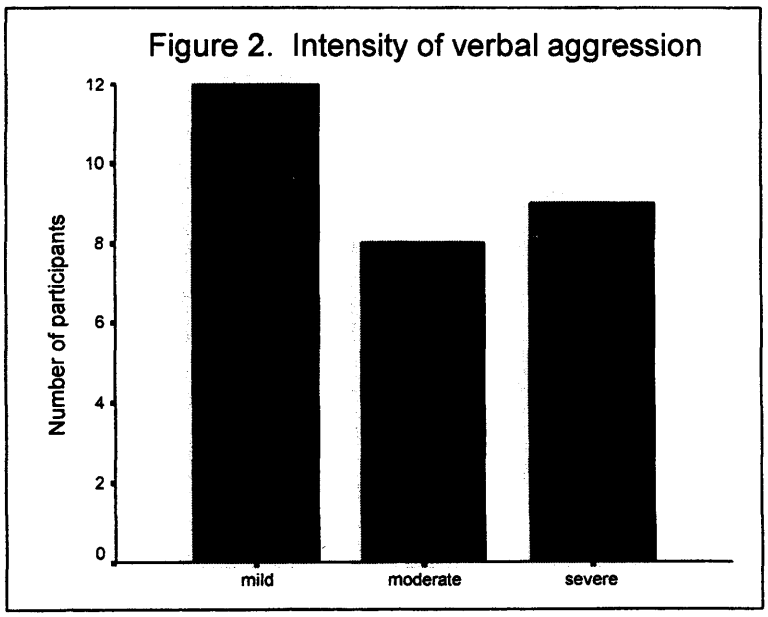


In 22 cases (76%) the aggressive behaviours were displayed once or more a day. There were very high levels of all four types of aggressive behaviours (see Table 18). In 28 cases (97%), aggressive behaviours were of long duration with onset at over a year prior to the interview.

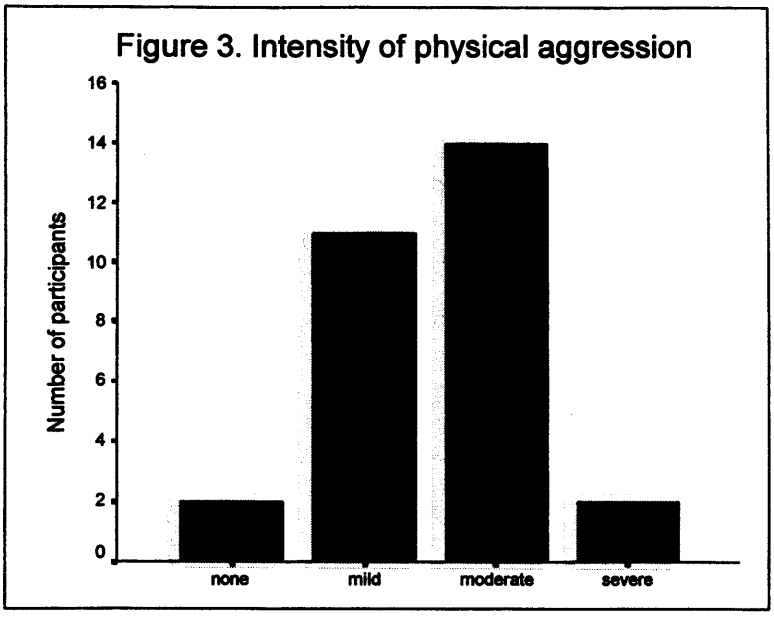
Table 18. Parents reports of the prevalence of aggressive behaviours

Type of aggression	n	(%)
Verbal	29	(100)
Physical (towards others)	27	(93)
Against property	29	(100)
Self-injury	28	(97)

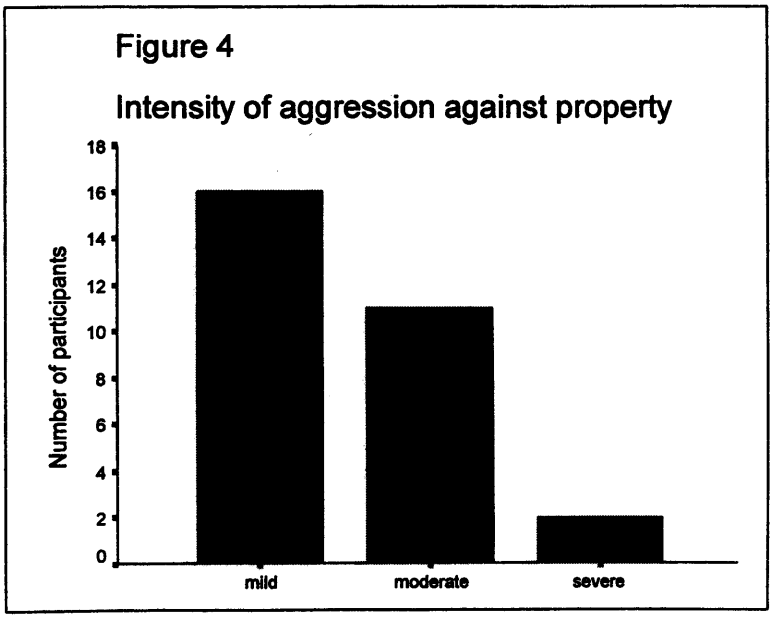
Considering the intensity of different types of aggressive behaviours, verbal aggression was rated as mild (shouting angrily, cursing mildly, or making personal insults) in 12 cases (42%), as moderate (cursing viciously) in 8 cases (28%), and as severe (threats of violence) in 9 cases (31%) (see figure 2).



Physical aggression was overall quite severe in nature with 11 (41%) grabbing clothes, striking, kicking, pushing, scratching, and pulling hair of others (mild), and 14 cases (52%) attacking others causing bruises, sprains and welts (moderate). Two (7%) were reported to have caused serious injury to others (severe) (see Figure 3).



Aggression to property was also intense with 16 (55%) reported to kick furniture and break things (mild), and 11 (38%) having broken objects/smashed windows (moderate). Two children (7%) were described as putting themselves or others at risk through violence to property (severe) (Figure 4).



- ***Triggers***

Parents were asked to describe the triggers or reasons for their children's aggressive behaviour (see Table 19). Over half the parents were unable to identify consistent triggers and described their child's behaviour as unpredictable depending on the child's mood that day.

Table 19. Triggers for aggressive behaviour (n = 29)

Triggers	n *	(%)
Unpredictable, no consistent trigger identified, depends on overall mood	15	(52)
Tiredness	8	(28)
Unable to get own way	11	(38)
Certain people (including parents, siblings, grandparents, doctors)	9	(31)
Crowds of people (i.e. shopping centers)	5	(17)
Frustration (i.e. if can't express self, dress self)	5	(17)
To avoid doing something	5	(17)
Attention seeking	11	(38)
Change in routine	5	(17)
When told off	2	(7)
Fizzy drinks, chocolate, sweats, biscuits	2	(7)

*N.B. For some children more than one trigger was reported

The most commonly reported reasons/triggers were to gain attention (11 cases), and in situations when the children were unable to get their own way (11 cases). Other triggers included particular people (parents, siblings, grandparents, doctors)(9 cases), and tiredness (8 cases). In those children whose aggressive behaviours were triggered by the desire for attention, several became aggressive if someone significant to them even talked to another person. Six parents stated that they knew when their child was about to become angry because they would tense their body, and would stare 'in a menacing way'. Less common triggers included being in a crowd of people, frustration (at being unable to express themselves, or dress themselves) , to avoid

doing something , and a change in routine, each reported in 5 cases. These anecdotal reports are supported by ratings made by parents as to the degree of sensitivity of children, in terms of how easily they became annoyed. None of the children were rated as showing 'appropriate levels of sensitivity'; 11 (38%) were rated as showing a 'tendency to get upset more easily than normal', 8 (28%) were rated as 'markedly oversensitive, with much lower threshold than normal', and 10 (35%) rated as showing 'a degree of hypersensitivity such that life has to be organized specifically to avoid upset'.

Teachers were also asked to describe the children's aggressive behaviours at school. Eighteen (62%) of the children were reported to have used physical force against another person, with only 3 (10%) reported as showing appropriate levels of control. Nineteen (66%) displayed aggression on a daily basis.

Associations were examined between the severity of different types of aggressive behaviours and IQ, gender, age and severity of sleep problem. There was no association between severity of verbal aggression, physical aggression and aggression against property and IQ (sum of scaled scores) gender and age. Both severity of physical aggression (Spearman rho = 0.45, p = 0.01) and severity of aggression against property (Spearman's rho = 0.42, p = 0.02) were found to be associated with severity of sleep problems.

To test whether IQ, age, gender and severity of sleep problems were associated with frequency of aggressive behaviour correlations were performed. There were no associations between IQ, age and gender and frequency of aggressive behaviour. However, frequency of aggression was associated with severity of sleep problems (Spearman rho = 0.39, p = 0.04).

- ***Self-injury***

Twenty-seven children (98%) displayed some type of self-injurious behaviour. Parents reported the most common self-injurious behaviours as pulling at nails and picking skin around nails (69% picked at dry/loose skin on their hands and feet, as well as biting and tearing nails off, 21% just picked at the skin and did not actually tear the nails off), biting the hand and/or fingers, self-pinching/scratching/picking at sores and hitting the head/body with the hand. These behaviours occurred in over 69% of the children (see Table 20).

Table 20. Types of self-injurious behaviours displayed by the sample

Self injurious behaviours	N	(%)
Pulling at nails/picking skin around nails	27	(93)
Hand/finger biting	25	(86)
Pinching, scratching, picking/rubbing skin and sores generally on body (not around nails)	21	(72)
Hitting the head/body with the hand	20	(69)
Hitting the head on a hard surface	13	(45)
Hitting objects to the head	10	(34)
Hair pulling	7	(24)
Eye pressing	4	(14)
Inserting objects into bodily orifices (ears and nose)	2	(7)

- ***Frequency of self-injurious behaviours***

Parents were asked about the frequency of the self-injurious behaviours displayed by their children (see Table 21).

Pulling at nails/picking skin around nails and hand/finger biting were reported by parents to occur frequently throughout the day for the majority of children displaying these behaviours. Self-pinching/scratching/picking at sores and hitting the head/body with the hand were reported to occur daily for the majority of the children in the sample displaying the self-injurious behaviours.

Parents were asked if their child displayed any other self-injurious behaviours: Three children had pulled teeth out; two children injured their feet; one by repeatedly and vigorously rubbing his feet together hard and biting his feet, and the other by banging his feet together hard. One child had reportedly bitten his tongue in anger, and another had licked a hot radiator and oven causing burns.

Table 21. Frequency of self-injurious behaviours

	Never	Less than once a week	1-2 times a week	Daily	Several times a day	Constantly in bouts throughout the day	Once/twice in lifetime
	n (%)	n (%)	n(%)	n (%)	n (%)	n (%)	n (%)
Hitting head on hard surface	19 (66)	3 (10)	3(10)	2 (7)	2 (7)	0 (0)	0 (0)
Eye pressing	26 (90)	1 (3)	1 (3)	0 (0)	1 (3)	0 (0)	0 (0)
Hitting object to head	19 (66)	3 (10)	5(17)	1 (3)	1 (3)	0 (0)	0 (0)
Hitting head/body with hand	9 (31)	5 (17)	5(17)	7 (24)	2 (7)	1 (3)	0 (0)
Pinching, scratching, picking/rubbing skin and sores generally on body (not around nails)	8 (28)	3 (10)	6(21)	6 (21)	5 (17)	1 (3)	0 (0)
Hair pulling	22 (76)	2 (7)	3(10)	2 (7)	0 (0)	0 (0)	0 (0)
Hand/finger biting	5 (17)	5 (17)	4(14)	3 (10)	6 (21)	6 (21)	0 (0)
Pulling out nails/picking skin around nails	2 (7)	7 (24)	3(10)	1 (3)	11 (38)	5 (17)	0 (0)
Inserting objects into bodily orifices	20 (69)	2 (7)	0 (0)	0 (0)	0 (0)	0 (0)	7 (24)

Associations were examined between age, IQ (sum of scaled scores), gender and sleep severity and the frequency of the most commonly shown forms of self-injury (pulling at nails/picking skin around nails, hand biting, self-pinching/scratching / picking at sores, and hitting the head/body with the hand). There was no association between age, IQ and gender and any of the most commonly reported self-injurious behaviours. However, frequency of hand biting, self-pinching /scratching /picking at

sores, and hitting the head/body with the hand were found to be significantly associated with severity of sleep problems (Spearman rho = 0.41, p = 0.03; Spearman's rho = 0.45, p = 0.02; Spearman rho = 0.45, p = 0.02).

A number of self-injurious behaviours were found to be significantly associated with each other: frequency of hand biting was associated with frequency of self-pinching/scratching/picking at sores (Spearman rho = 0.58, p = 0.002), frequency of hitting the head/body with the hand (Spearman rho = 0.52, p = 0.008), and frequency of pulling at nail/picking skin around nails (Spearman rho = 0.43, p = 0.03).

To test whether there was an association between the most common forms of self-injury and verbal and physical aggression and aggression towards property, Spearman rank correlations were performed. Results showed significant associations between: verbal aggression and hand biting (Spearman rho = 0.37, p = 0.04); physical aggression and hand biting (Spearman rho = 0.62, p = 0.001), self-pinching/scratching/picking at sores (Spearman rho = 0.50, p = 0.009) and hitting the head/body with the hand (Spearman rho = 0.43, p = 0.03); and aggression against property and self-pinching /scratching /picking at sores (Spearman rho = 0.40, p = 0.05).

- ***Causes of self-injury***

Parents identified triggers for the various self-injurious behaviours described (see Table 22). Most of the self-injurious behaviours were reported to occur in conditions where the individual is frustrated or angry. The main exception to this is for pulling at nails/picking skin around nails which was reported to occur most often when the child is bored or when the behaviour has become a habit.

Table 22. Triggers for self-injury

	Anger / Frustration		**Attention seeking**		**Fear/ Anxiety**		**Excitement**		**Boredom/ Habit**		**Unknown**	
	n	**(%)**	**n**	**(%)**	**n**	**(%)**	**n**	**(%)**	**n**	**(%)**	**n**	**(%)**
Hitting head on hard surface (N=13)	8	(62)	5	(38)	0	(0)	0	(0)	0	(0)	0	(0)
Eye pressing (N=4)	2	(50)	2	(50)	0	(0)	0	(0)	0	(0)	0	(0)
Hitting object to head (N=10)	8	(80)	0	(0)	1	(10)	1	(10)	0	(0)	0	(0)
Hitting head/body with hand (N=20)	18	(90)	2	(10)	0	(0)	0	(0)	0	(0)	0	(0)
Pinching, scratching, picking/rubbing skin and sores on body (N=21)	10	(48)	1	(5)	1	(5)	0	(0)	8	(38)	1	(5)
Hair pulling (N=7)	6	(86)	1	(14)	0	(0)	0	(0)	0	(0)	0	(0)
Hand biting (N=25)	22	(88)	3	(12)	0	(0)	0	(0)	0	(0)	0	(0)
Pulling out nails/picking skin around nails (N=27)	0	(0)	2	(7)	0	(0)	0	(0)	23	(85)	2	(7)

A spasmodic upper-body squeeze was described in 27 children (93% of cases); this was reported to occur at least daily (if not several times a day) in half of the children. The main trigger for this behaviour was reported to be excitement, for 25 cases

(96%). Other unusual behaviours included taking clothes off (including stripping or removing shoes and socks) when angry, in 4 cases(14%), and one child was reported to make a clicking noise with her mouth when excited.

- ***Treatment of self-injury***

Parents were asked to describe effective methods of intervention used to reduce self-injury. These are summarized in Table 23. The most commonly used means of reducing self-injury were 'time-out', using firm, clear commands, and ignoring the self-injurious behaviour. Medication was used in 9 cases (31%): Ritalin was tried and found to be helpful in 5 cases, although the parent of one of the children reported side-effects of low mood and loss of appetite; Lithium had been tried in one case and was found to be very effective, Tegratol had been tried by 2 children and was not effective, and Carbemazepine was tried by one child and was also ineffective.

Table 23. Effective interventions for self-injurious behaviour

Intervention*	**n**	**(%)**
Firm, clear commands	12	(41)
Time-out	14	(48)
Ignore	12	(41)
Reward system	1	(3)
Avoid difficult situations likely to cause self-injury	3	(10)
Medication	4	(14)
Cranial osteopath	1	(3)

*N.B. For some children more than one intervention was reported

3.4.2. Other Maladaptive Behaviours

A further set of maladaptive behaviours listed in the Schedule of Handicaps, Behaviour and Skills (HBS) (Wing, 1980) was presented to parents and they were

asked to rate each as being either 'no problem', a 'mild' or a 'marked' problem. Some of these behaviours overlap to some extent with areas covered in the semi-structured interview reported on above. As can be seen in Table 24, behaviours most commonly rated as 'marked' were 'embarrassing behaviour in public places', approaching strangers, destructiveness and difficult or objectionable personal habits.

Table 24. Prevalence and degree of maladaptive behaviours (n=29)

Maladaptive behaviours	'no problem'		'minor'		'marked'	
	n	(%)	n	(%)	n	(%)
Wandering	1	(3)	17	(59)	11	(38)
Destructiveness	3	(10)	14	(48)	12	(41)
Noisiness	7	(24)	11	(38)	11	(38)
Aggressive behaviour	4	(14)	14	(48)	11	(38)
Embarrassing behaviour in public places	3	(10)	7	(24)	19	(66)
Crying and moaning	10	(35)	8	(28)	11	(38)
Approaching strangers	16	(55)	0	(0)	13	(45)
Inappropriate response to others' emotions	14	(48)	12	(41)	3	(10)
Difficult or objectionable personal habits	6	(21)	11	(38)	12	(41)
Lack of cooperation	5	(17)	13	(45)	11	(38)
Needs constant supervision	20	(69)	5	(17)	4	(14)
Demand's carers' attention	3	(10)	23	(80)	3	(10)
Difficulties with other people	5	(17)	13	(45)	11	(38)

None of the children were presently on the Child Protection register; however one child had been on the register in the past. This occurred as a result of reports of repeated head injuries and worse behaviour at home compared to the nursery school. As such, a Social Services case conference was held and the child's name was placed on the 'at risk' register for 'emotional abuse'. An attempt to remove the child from

her home was successfully opposed by her parents. The girl was then assessed at a tertiary referral clinic, at the request of her GP and family, and was diagnosed with SMS. The girl's name was subsequently removed from the 'at risk' register since the aggressive behaviours were considered to form part of the SMS phenotype.

The Developmental Behaviour Checklist (Einfeld & Tonge, 1989) was completed by 26 of the parents and 27 of the teachers in order to assess behavioural and emotional problems. Three 'parent' questionnaires and 2 'teacher' questionnaires were not returned. Table 25 presents the mean developmental behaviour checklist scores and percentiles for each subscale of the DBC, for the parent DBC and the teacher DBC.

Table 25. Mean Developmental Behaviour Checklist Scores and Percentiles (Parents and Teachers)

Subscale	Parents n=26		Teachers n=27	
	Score	Percentile	Score	Percentile
Disruptive	25.1	77.4	17.3	75.5
Self-absorbed	20.1	72.6	12.6	75.2
Communication disturbance	7.7	71.5	4.0	69.4
Anxiety	11.2	69.3	6.1	54.8
Autistic Relating	5.8	55.4	3.9	53.3
Antisocial	1.7	65.2	0.7	71.3
Total behaviour problem score	86.3	77.3	59.1	75.2

There was no correlation between the mean 'total behaviour problem' percentile scores rated by the parents and teachers (Pearson $r = 0.23$, $p = 0.22$), indicating that parents and teachers rated the same children's behaviour rather differently. A clinical cut-off can only be derived from the 'parent' questionnaire. Out of 26 children, 22 scored above the clinical cut-off, indicating 'major behavioural/emotional problems'.

The highest scores were for the 'Disruptive' and 'Self-absorbed' subscales, and the lowest on the 'Antisocial' and 'Autistic relating' subscales.

3.5 ATTENTION AND HYPERACTIVITY

Parents were asked selected questions from the 'Activity Level and Hyperactivity' section of the Parental Accounts of Children's Symptoms (PACS) (Taylor and Schachar, 1993). All items needed to diagnose Hyperkinetic Disorder, according to the International Classification of Disease (ICD-10) criteria, were included. None of the subjects fulfilled all three of the main features for a diagnosis of Hyperkinetic disorder (attention problems, impulsivity and restless overactivity/hyperactivity (see Table 26). The most frequently reported behaviour difficulties were distractibility, having difficulty waiting one's turn, and remaining seated during mealtimes .

Teachers were also asked about the child's ability to concentrate: 17 (59%) of children were reported as 'almost always being distractible'; 18 (62%) as 'invading other's personal space', and 13 (45%) as 'getting up and moving around the classroom frequently'.

Table 26. Prevalence of attention problems, impulsivity and restlessness according to parent responses from the PACS.

	n	(%)
Attention Problems:		
Inattentiveness whilst:		
Watching TV	2	(7)
Reading	9	(31)
Joint play	4	(14)
Distractibility	18	(62)
Seeming not to listen	6	(21)
Difficulties in organizing	3	(10)
Difficulties following instructions	11	(38)
Often loosing things	13	(45)
Being forgetful	7	(24)
Impulsivity:		
Difficulties in waiting for his/her turn	20	(69)
Blurting out answers before questions are completed	10	(35)
Butting into conversations or games	15	(52)
Restless Overactivity (Hyperactivity):		
Running about or climbing excessively	6	(21)
Always on the go	9	(31)
Fidgeting with hands and feet whilst:		
Watching TV	14	(49)
Reading	11	(38)
Unable to sit still whilst:		
Watching TV	6	(21)
Reading	13	(45)
Joint-play	8	(28)
Mealtimes	15	(52)

3.6 AUTISM AND AUTISTIC SPECTRUM DISORDERS

The Schedule of Handicaps Behaviour and Skills (HBS) (Wing, 1980) was administered in order to identify those children who fulfilled ICD-10 and DSM-III-R criteria for typical Autism. Out of the 29 children, 27 (93%) were rated as having ICD-10 and DSM-III-R typical autism on the basis of the HBS assessment interview.

This finding supports the hypothesis that individuals with SMS exhibit high rates of autistic spectrum behaviours.

The social age of the children was also calculated from the HBS. Social ages ranged from 2 years – 9 years 5 months, with a mean of 5 years 2 months (SD = 1.54). The association between social age and Full Scale and Verbal IQs (scaled scores) was examined using Pearson Product Moment correlations. There was no significant association between these measures (Pearson r = 0.21, p = 0.27; Pearson r = 0.10, p = 0.62).

The 'Autistic Relating' subscale on the Developmental Behaviour Checklist (Einfeld & Tonge, 1989) was also used as a screen for autistic behaviours. Table 27 presents the mean percentiles for this subscale. It is interesting to note that although 27 of the children were rated as having autism on the basis of the HBS, the mean scores on the Autistic Relating subscale of the DBC were low – at the 55th percentile of a sample of children and adolescents with learning disabilities on the Parent Scale and at the 53rd percentile on the Teacher Scale of the DBC.

Table 27. 'Autistic Relating' subscale percentiles on the Developmental Behaviour Checklist

Percentile groups	Parents (n=26)		Teachers (n=27)	
	n	(%)	n	(%)
0-50	8	(28)	9	(31)
51-60	3	(10)	3	(10)
61-70	5	(17)	7	(24)
71-80	5	(17)	4	(10)
81-90	3	(10)	3	(14)
91-100	2	(7)	1	(3)

Over the total sample there was no significant association between a diagnosis of autism and the child's age, gender or IQ (chi-square = 2.68, p = 0.10; chi-square = 0.31, p = 0.58; chi-square = 0.12, p = 0.73). The severity of Autism (taken from the HBS 'Autism' score) was not associated with IQ score, age, gender, severity of sleep problems, or the severity of aggressive behaviours (Pearson r = -0.08, p=0.68; Pearson r = 0.03, p = 0.86; chi-square = 0.07, p = 0.79; Spearman rho = 0.17, p = 0.38; Spearman rho = 0.32, p = 0.09).

- ***Specific behavioural items***

The prevalence of specific autistic behaviours in the sample of children with SMS is presented in Table 28. Overall, children were reported to display social and communication impairments, as well as stereotypic and repetitive behaviours. In terms of social and communication impairments, a large percentage were reported to prefer adult company; over half were reported to have inappropriate eye contact, limited variation in facial expression, and to talk about repetitive themes. Repetitive questioning was reported in just over half of the sample. An insistence on the maintenance of the same routine and repetitive destructive activities were also reported in a large percentage of children.

Table 28. Prevalence of specific autistic behaviours

Autistic behaviours	n	(%)
Social/communication impairments		
Repetitive questioning	14	(48.3)
Repetitive themes	17	(58.6)
Difficulties understanding gesture/mime	0	(0)
Difficulties understanding facial expression	1	(3.4)
Limited changes of facial expression	17	(58.6)
Looks to familiar person for reassurance	13	(44.8)
Inappropriate social use of eye contact	16	(55.2)
Limited social play	12	(41.4)
Impaired imaginative play	12	(41.4)
Difficult behaviour in public places	7	(24.1)
Overfriendliness	13	(44.8)
Prefers interaction with adults	23	(79.3)
Stereotypic behaviours		
Fascination with bright lights/shiny objects	5	(17.2)
Interest in watching things spin	9	(31.0)
Twisting& turning hands/objects	1	(3.4)
Repetitive destructive activities	20	(69.0)
Repetitive self-chosen activity	13	(44.8)
Dislike of change of routine	19	(65.5)
Routines invented by child	4	(13.8)
Unusual interests	9	(31.0)

Parents were asked questions about how their children responded to peers and whether they had friends. As can be seen in Table 29, the majority were able to interact in small groups, although none had particular friends. Teachers were also asked about the child's ability to form friendships; they confirmed these findings, reporting that over half had acquaintances, but did not have a special friend. The same number were reported to attempt social interactions, but these were often inappropriate.

Table 29. Response to peers/friendships (n=29)

	N	(%)
No interest in peers	1	3
Enjoys the presence of peers, but does not join in	1	3
Plays in parallel with peers	3	10
Interacts in small groups (no special friend)	8	28
Seems to prefer some people to others(no special friend)	10	35
Makes friendships	6	21

3.7 IMPACT ON THE FAMILY

Parents were asked to reflect on the effect that their child's behaviour difficulties had upon their family. Of the 24 parents who were married/living together, 10 (35%) reported that their child's special needs frequently caused arguments, and 11 (38%) stated that they occasionally argued because of extra pressures placed on them due to their child's special needs. Parents were also asked to comment on the relationship between their child with SMS and his/her sibling(s). Nineteen children (65%) were reported to get on well with their sibling(s). Thirteen (45%) of parents felt that their social life had been very disrupted, and 7 (24%) felt that there had been some disruption due to their child's special requirements.

Twenty-five parents returned completed General Health Questionnaires (GHQ)(Goldberg & Hillier 1979; Goldberg & Williams 1988), (all of them completed by mothers), used to screen for parental/maternal mental illness. Eight (32%) mothers scored above the clinical cut-off for a psychiatric disorder. The relationship of GHQ

scores with age, marital status, severity of behaviour difficulties (based on the total score on the Developmental Behaviour Checklist), and severity of sleep problems was examined statistically. A higher score on the GHQ was associated with more severe sleep problems (Spearman rho = 0.45, p = 0.02), and marital status (the mean rank GHQ score for the single mothers being higher than for the married mothers) (chi-square = 5.1, p = 0.02). However, there were no significant associations between the GHQ score and age (Pearson r = 0.14, p = 0.50), or severity of behaviour difficulties (0.26, p = 0.21).

Parents were asked if they had received any professional advice as regards their children's difficult behaviours, and if so they were asked about its nature and efficacy. Eighteen families (62%) were in contact with mental health professionals at the time they were interviewed, and 13 (45%) had had contact with mental health professional in the past. The mental health professionals seen were: Clinical Psychologist (7 families, 39%), Psychiatrist (5 families, 28%), Family therapist (1 family, 6%) other members of mental health teams (5 families, 28%). Of those receiving input, 50% saw the mental health professional(s) 2 or 3 times a month at most; 44% were seen 2-3 times a year and 6% were seen once a year. The most common reasons for referral were for assessment and advice regarding aggressive behaviours and sleep problems. Of those receiving help, 21 parents(72%) were mostly satisfied or very satisfied with the input they were receiving from mental health professionals, and the others were mostly dissatisfied or very dissatisfied. The main reasons given for dissatisfaction were infrequent contact, and inappropriate

advice concerning intervention strategies. Eighteen families (62%) had an allocated Social Worker, but only 8 (44%) had regular contact with their Social Worker; the remainder had only met with the Social Worker a few times.

4 DISCUSSION

The main findings of the current study serve to confirm and expand upon previous reports in the literature on Smith-Magenis syndrome, which indicate a distinctive behavioural phenotype for the syndrome. Prior to discussing this in more detail, it is important to consider a number of methodological issues.

4.1 METHODOLOGICAL ISSUES

A limitation of the current study concerns the representativeness of the sample. Participants were recruited through the Smith-Magenis syndrome Parent Support Group, and through Regional Genetics Centres. Children known to the Clinical Geneticists and to the Parent Support Group would have been referred by Pediatricians, Psychologists and Psychiatrists owing to the severity of their developmental difficulties and/or behavioural problems. As a result it is possible that the present sample represents the more severe end of a spectrum of physical and behavioural difficulties associated with the condition, and that there are other children with Smith-Magenis syndrome in the population who are less severely affected and show appropriate adaptive behaviour and as such have not come to the attention of health professionals. Only epidemiological studies of whole populations screened for SMS could establish how representative the present sample is of the syndrome in terms of cognitive and behavioural characteristics.

A further limitation of the study was the decision not to use one or more comparison groups. However, at this stage of our knowledge of SMS it was considered more valuable to report on internal consistencies in behaviours within the SMS group. A further step would be to use information to design controlled studies of specific behaviours to further delineate the cognitive and behavioural phenotype.

In view of the lack of comparison groups in this study, the present findings, where possible, will be compared with findings previously reported in studies of individuals drawn from either general learning disabilities populations, or samples of children with specific syndromes. However, it must be borne in mind that this approach does not allow for an exact match of participants in terms of age, level of learning disability and socio-economic status. Secondly, some of the instruments used in the current study are very different to those used in previously reported studies, making direct comparisons on frequencies of particular behaviours difficult.

An additional limitation with the study is the relatively small sample size; however, given the rarity of SMS, a sample of 29 affected children aged 6-16 is likely to yield useful findings. This sample is in fact one of the largest cohorts of SMS individuals studied to date. Nevertheless, the small sample size and the large number of variables examined suggest that the findings of the current study will require replication. A final point is that this study relied to a large extent on reports gathered from parents and teachers; as a result, the information collected is subject to inaccuracies and biases and informants' personal views.

4.2 INTELLECTUAL AND COGNITIVE ABILITIES

All children in the present study had learning disabilities, (IQ<70), and the majority (72%) fell within the Severe Learning Disability range (IQ of 50 or below). These findings are consistent with those of Greenberg et al. (1996), who reported that the intellectual abilities of individuals with SMS range from Moderate to Severe, with the majority of individuals falling within the IQ range of 40-54. In contrast, Horn et al. (1999, personal communication) found that the majority of her sample of adults with SMS had IQs within the Moderate Learning Disabilities range. This difference may be a result of a difference in the instrument used; the child and adult versions of the Wechsler Intelligence Scales. Different types of abilities may be assessed by the two instruments.

On the verbal scale of the WISC-III, children showed a relative strength in 'Information' a test assessing 'fund of information' or general knowledge. A relative weakness was identified in Arithmetic. In the non-verbal domain, better performance was shown for tests of visuo-spatial ability, and visual recognition. Relative weaknesses were seen in tasks requiring visuomotor coordination and visual sequencing. In 72% of the sample of children with SMS, long-term memory for events was rated by parents and teachers as being superior compared to children of the same age without learning disabilities. These findings are consistent with those of Dykens et al.(1997). Horn et al. (1999) found that the scores for the Information subtest were significantly lower than for the Comprehension and Vocabulary subtests in her sample of adults with SMS. A possible explanation for this might be that the

acquisition of general knowledge is influenced by school learning and richness of early environment, and the older adults in particular may not have had access to the type of education that the children in the present sample had. In support of the findings from the present study, Dykens et al. (1997) also identified weaknesses in Arithmetic and sequential processing, and suggested that individuals with SMS may have difficulties placing stimuli in serial and temporal order in auditory, visual and motric short term memory. Problems in learning and retaining mathematical information has been associated with deficits in sequential processing which would relate to the findings presented above (Kaufman, Kaufman, & Goldsmith, 1984).

Although distinctive, such profiles of strength and weakness may not be unique to Smith-Magenis syndrome. Many males with fragile X syndrome show deficits in sequential processing (Hodapp et al., 1992), and individual's with Prader-Willi syndrome have been reported to have strengths in long-term memory (Curfs & Fryns, 1992).

It should be noted that the WISC-III is a measure of global intellectual ability, and as such any conclusions regarding the specific strengths and weaknesses of children with SMS must be tentatively drawn. More detailed neuropsychological investigations will need to be undertaken in the future to address this issue.

Although Dykens et al. (1997) found relative strengths in reading/decoding, these findings were not replicated in the present study; only half the sample were able to

read at all, and the mean age for Reading Accuracy was 7 years, 2 months, at a chronological age of 9 years and 4 months.

- **Implications of cognitive abilities for educational approaches**

Larger scale studies are required to confirm the findings of the present study. If a specific profile of cognitive strengths and weaknesses can be identified this would inform the planning of educational programmes for individuals with Smith-Magenis syndrome. It could further enhance understanding of some of the behaviours exhibited by this group; for example, the repetitive questioning observed may be related to short-term auditory memory or sequencing difficulties proposed by Dykens et al. (1997). If a relative strength were to be confirmed in the visuo-spatial domain, then it would be advisable to use visuo spatial cues to supplement teaching methods. Visual reminders could be used to help process sequential information in an academic task or a daily schedule. Gestures, signing and picture boards could also be used to facilitate comprehension of spoken language and to support speech. In the present sample a good ability in computers was identified and as such could be used for educational instruction using computer-assisted technology.

4.2 SLEEPING DIFFICULTIES

Despite claims in the literature that sleep abnormalities are part of the behavioural phenotype of SMS, only a limited amount of information has been published to date with respect to actual daily and nocturnal sleep habits and behaviours of affected individuals.

The present study found that all of the children with Smith-Magenis syndrome were classed as having sleep problems, characterized primarily by difficulties with settling, night-time waking and early morning waking. These findings support earlier reports suggesting that sleep problems are a prominent feature of SMS; Greenberg et al. (1996) reported sleep problems in 75% of participants, while Smith et al. (1997) reported prominent sleep problems in 100% of individuals with SMS aged 1 to 32 years, and Horn (1999) reported sleep problems in 81% of adults with SMS. Dykens et al. (1997) and Horn (1999) found similar sleep problems to the current study; difficulties in falling asleep, night wakening and day-time sleeping. The mean wake-up time of 5.30am found by Smith is close to the present finding of a mean wake up time of 5.00am. The number of times participants woke up in the night was the same in all the studies (mean of twice a night). The children in the present study took longer to return to sleep compared to the children in Smith et al's (1997) study. Smith et al. (1997) found an effect of age on sleep duration, with duration decreasing with age. She also found that increased age was related to earlier wake up times and an increased number of wakenings in the night. In contrast, in the current study and in Horn's study (1999), increased age was related to less severe sleep problems, and fewer awakenings at night. It maybe that as the individuals get older their behaviour at night becomes less disruptive, and therefore parent/carers are less likely to be disturbed and may not be aware that the individual has actually woken up.

- **Comparison with sleep studies of individuals with general learning disabilities and individuals with other syndromes.**

Sleeping problems in young children are common. As many as 20% of 2 year olds and 14% of 3 year olds are reported to wake regularly during the night (Richman et al., 1975; Jenkins et al. 1980). Investigations of children with learning disabilities have shown sleeping problems to be even more widespread and persistent (Bartlett, Rooney, & Spedding, 1985; Quine, 1991; Wiggs and Stores, 1996a). Bartlett et al. (1985) in a study of 214 children with learning disabilities under 16 years of age, reported that 56% woke on average once a night, 53% were difficult to get to go to bed, and 56% were difficult to settle. These figures are lower than those found in the present sample of children with Smith-Magenis syndrome, lending support to the idea of sleep disturbance being part of the Smith-Magenis syndrome.

Stores et al. (1996) investigated the occurrence and nature of sleep problems and behaviours in a group of children with Down syndrome, compared with their siblings, children from the general population, and children with an intellectual disability other than Down syndrome. The sleep problems and behaviours they investigated can be compared with the current sample (refer to Table 30). The children with Smith-Magenis syndrome showed far higher rates of settling, waking, and early waking problems compared to all of the other groups; they were also far more likely to insist on sleeping with someone, and more reluctant to go to bed due to fears. There were higher rates of gagging/choking, and apnoeic episodes in the SMS sample compared to the other groups.

Children with Smith-Magenis syndrome also showed higher rates of teeth grinding and they were more likely to have nightmares and night terrors. Moreover, bedwetting occurred at almost twice the rate in children with Smith-Magenis syndrome compared to the group with mixed learning disabilities, and at almost three times the rate for children with Down syndrome. The high incidence of bedwetting in the children with Smith-Magenis maybe explained by their developmental level, and/or low muscle tone. Alternatively, there may be other physical problems related to the syndrome that need to be investigated further. Children with Smith-Magenis syndrome were far more likely to have daytime naps and to have excessive daytime sleepiness compared to the other groups.

Overall, the children in the present sample had a far higher rate of combined settling, night waking and early waking problems, compared to the findings of Wiggs & Stores (1996), for their sample of 486 children with severe learning disabilities, 41% versus 10% of the samples.

Table 30. Percentage of children showing frequent sleep problems and behaviours from Stores et al. 1996, and the present study.

Sleep problem/behaviour	Down syndrome	General population	Intellectual disability	Smith Magenis syndrome (current study)
Disorders of initiating and maintaining sleep				
Settling	20	2	29	45
Waking	32	10	44	97
Early waking	17	6	30	62
Insists on sleeping with someone	9	3	10	17
Reluctance to go to bed due to fears	3	4	0	38
Features associated with obstructive sleep apnoea				
Restlessness during sleep	60	26	52	31
Loud snoring	43	10	27	31
Gags/chokes	7	1	7	14
Apnoeic episodes	12	1	6	20
Behaviours occurring during sleep				
Sleep talking	19	8	15	14
Teeth grinding	17	8	10	24
Bedwetting	16	2	28	59
Nightmares	0	1	3	7
Sleep walking	3	1	0	0
Night terrors	0	0	2	3
Tongue biting	4	0	0	0
Head banging	7	3	13	3
Behaviours occurring during the day				
Daytime naps	9	1	14	82
Excessive daytime sleepiness	8	2	15	80

- **Implications for intervention**

A very high proportion of the sample (83%) had been prescribed some form of medication to facilitate sleep at one time or another. However, medication was generally found not to be effective in this group.

The underlying mechanism for sleep disturbance in SMS is not yet understood; Greenberg (1996) postulated that a gene found in the SMS critical region might be involved in regulating REM sleep. A 24-hour study of the urine of individuals with SMS found increased levels of the hormone melatonin (Potacki et al., 1997). This finding could be related to the abnormalities of sleep found in the syndrome, as melatonin is known to be involved with control of sleep patterns and circadian rhythms. There are anecdotal reports of melatonin being used with some success to treat sleep difficulties in several American children with SMS (Smith et al.1997). However, in the absence of randomized controlled trials examining the effectiveness of melatonin no recommendations can be made on its value.

The most effective methods of managing sleep problems reported by parents in this study involved minimizing the disruption caused by waking during the night or early in the morning. Parents reported that locking bedroom doors and other doors in the house was a useful strategy, as well as providing non-disruptive activities for the child during the night whilst awake. Encouraging a routine and implementing firm and consistent techniques was also helpful. From both the literature and parent reports, no effective methods have been identified thus far to prolong sleep duration or reduce early morning waking and night-time waking in SMS. Some behavioural methods such as ignoring the child and implementing a firm structure and routine with consistent commands were found to be helpful in some instances.

Bramble et al. (1997) carried out a small treatment study of sleep problems on 15 children with severe learning disabilities; one of the children in the study had Smith-Magenis syndrome. A behaviour modification programme was implemented using 'rapid extinction' for the severe, longstanding sleep problems presented. Although all of the children's sleep problems initially improved, the child with Smith-Magenis syndrome was the only one to develop settling difficulties again by the 4-month follow-up. This confirms the severity of the sleep problems in children with Smith-Magenis syndrome.

There is a need for controlled evaluation of the efficacy of different interventions for the sleep difficulties in this population, including behavioural interventions and medications.

- **Relationship between sleep, behavioural problems and parental stress**

In the present study, a higher score on the General Health Questionnaire (GHQ) (Goldberg & Hillier 1979; Goldberg & Williams, 1988), used to detect psychiatric disorders amongst the mothers, was associated with more severe sleep problems in their children with SMS. Bartlett & Beaumont (1998) in their sleep treatment study, also used the General Health Questionnaire to screen for parental psychiatric disorders. The mean score obtained for their study was 10.90, compared to a mean score of 16.0 in the present study, indicating higher levels of psychiatric disorder/psychological difficulties amongst parents of SMS children with sleep problems compared to other parents of children with learning disabilities with sleep

problems. This would suggest that the sleep problems in children with SMS are more severe and hence lead to more psychological problems for parents. These findings are also consistent with those by Quine and Wage (1991) who reported that having a child with a sleep problem has been found to have negative effects on parents, in terms of increased stress and irritability (Quine and Wage, 1991).

Severity of sleep problems was also found to be associated with frequency of aggressive behaviour, and severity of physical aggression and aggression against property, along with certain types of self-injurious behaviour, (hand/finger biting, self-pinching/scratching/picking at sores, and hitting the head/body with the hand). These findings are consistent with reports that sleep problems are associated with daytime behaviour difficulties (Richman 1981; Zuckerman et al. 1987; Quine 1991). There are a number of ways in which sleeping difficulties might affect a child's daytime behaviour. Firstly, reduced or disrupted sleep has been found to impair daytime functioning. More specifically, loss of slow wave sleep (stages 3 and 4 non-rapid eye movement, NREM) has been seen to have serious effects on behaviour and mood in other contexts (Guilleminault & Winkle 1981; Horne 1988), and rapid eye movement (REM) sleep, which is already often impoverished in children with learning disabilities (Stores 1992) is thought to be associated with the learning process. Secondly, the consequences of sleep problems include daytime fatigue and sleepiness in both parent and child. Sleep-deprived children have also been seen to manifest their lack of sleep in daytime over-activity rather than under-activity (Guilleminault & Winkle 1981). Daytime drowsiness or over-activity is not conducive to the parent teaching appropriate daytime behaviour, or to the child

learning new behaviours. Thirdly, hypnotic medication may itself contribute to daytime drowsiness and has been seen to further reduce REM sleep (Kales et al. 1983).

The high prevalence and persistence of sleep problems in children with SMS warrants urgent attention given that sleep problems tend to be associated with high levels of maternal stress (Quine 1991) and add a considerable burden to families already stressed in a variety of ways by having a handicapped child in the family (Glendinning 1983). Hence the need for appropriate intervention strategies to address the sleeping problems of children with Smith-Magenis syndrome should be viewed as a priority.

4.4 MALADAPTIVE BEHAVIOURS AND AGGRESSION

This study identified particular patterns of aggression, self-injury and maladaptive behaviours in the sample of children with Smith-Magenis syndrome, which were consistent across participants. Almost all the children showed high levels of verbal and physical aggression to others, aggression against property and self-injury. The aggressive behaviours were of long duration, and in 72% of cases aggressive behaviours occurred at least once a day. These findings are consistent with previous reports on children and adults with Smith-Magenis syndrome (Greenberg et al., 1991, de Rijk-van Andel et al., 1991; Dykens et al., 1997, Dykens et al., 1998). The 97% prevalence of self-injury found in the current sample is higher than rates previously reported in Smith-Magenis syndrome (Dykens et al., 1997; de Rijk-van et al., 1991),

but it is consistent with the findings of Dykens et al.(1998) and Horn (1999) who reported prevalence rates of 90-100% in their studies of children and adults with Smith-Magenis syndrome.

Verbal aggression and aggression against property were found to be the most frequent forms of aggression shown. Although physical aggression towards others was found to be slightly less frequent, it was overall more severe in nature, with almost half of the children reported to attack others causing moderate injury, and 2 were reported to have caused serious injury to others. There were reports of cruelty to animals, such as taking fish out of their bowls, squashing ladybirds, and kicking dogs.

Large scale studies have been conducted on the prevalence of aggression in the general population of individuals with learning disabilities and estimates range from 8.9 to 24% (Gardener and Cole, 1993; Jacobson, 1982). Prevalence estimates for self-injury in the population of individuals with learning disabilities range from about 6-22% (Read, 1998). The present finding of considerably higher rates of aggressive and self-injurious behaviours in the sample with SMS, suggests that these behaviours are part of the phenotype or behavioural profile of Smith-Magenis syndrome. These results indicate that children with Smith-Magenis syndrome are at greater risk for psychopathology than other individuals with comparable levels of learning disabilities, and that access to mental health services for this client group should be a priority.

In addition to examining the literature on children with learning disabilities in general, a further aim of the study was to compare findings with those previously reported in groups of children with learning disabilities of known etiology, so as to further delineate syndrome specific characteristics. Previous studies have shown high rates of physical violence (72%) and temper tantrums (80%) in individuals with Oculocerebrorenal syndrome of Lowe (mean IQ in the moderate range, mean age = 16.8 years) (Kenworthy et al., 1993), these figures are lower then the findings in the present study. Studies have reported maladaptive behaviour in 70-95% of children with Prader-Willi syndrome (Dykens & Kasari, 1997), again the figures are higher for the children with SMS in the present study. A study of the behavioural phenotype of Down syndrome (Collacott et al., 1998) found considerably lower rates of aggression (8.61%) and property destruction (9%) than those found in the current study. Fifteen percent of children and 20-38% of adolescents showed behavioural or psychiatric difficulties (Gath and Gumley, 1986; Meyers & Peuschel, 1991). Thus aggression appears to be more prevalent in SMS than in a number of genetically determined syndromes for whom comparative data are available, supporting the hypothesis that aggression is part of the SMS phenotype.

Aggressive behaviour was not found to be associated with either gender or level of intellectual functioning. This is at variance with previously reported findings ; a study of aggression in a general learning disability population found that aggressive behaviours were exhibited more commonly by males than females, and by those with more severe intellectual disabilities (Sigafoos et al., 1994).

The Developmental Behaviour Checklist (DBC) (Einfeld and Tonge, 1989), assesses a broad range of behavioural and emotional difficulties in children and adolescents with learning disabilities. Several studies have used the instrument to compare individuals with a syndrome and control individuals (see Table 31).

Table 31. Percentage scoring above the clinical cut-off on the Developmental Behaviour Checklist from various studies

	(Einfeld et al. 1994) (n=48)		(Einfeld et al. (1997) (n=70)		Hoare et al. (1998) (n=143)	(Present study) (n=29)
	Fragile X	Control (LD)	Williams	Control (LD)	Severe LD	Smith-Magenis syndrome
Clinical Cut–off	31.2%	40.7%	61.4%	40.7%	38%	85%

The figures from the studies presented are much lower in comparison to those for the current sample of children with Smith-Magenis syndrome, of whom 85% scored above the clinical cut-off.

Examining the individual subscale scores on the DBC of children with SMS and children with fragile X and with general learning disabilities (Einfeld et al., 1998), it can be seen that the children with Smith-Magenis syndrome scored far higher on all of the subscales of emotional and behavioural problems, especially the 'Disruptive' and 'Self-absorbed' subscales, in comparison to children with fragile X syndrome, Williams syndrome and children with learning disabilities (see Table 32). Additionally, Hunt et al., (1994) in their study of 40 children with Tuberous Sclerosis, reported mean total scores on the DBC of 54.72, which is much lower than the mean figure of 86.3 in the present sample.

Table 32. Comparison of Mean Developmental Behaviour Checklist Scores for children with Fragile X syndrome, Williams syndrome and Learning Disabilities

Subscales	FragileX syndrome (n= 48) Einfeld 1998	Control(LD) (n=454) Einfeld'97'98	Williams syndrome (n=70) Einfeld 1997	Smith-Magenis syndrome (n=26) Present study
Total behaviour problem	35.7	42.1	57	86.3
Disruptive	8.7	11.4	14.1	25.1
Self-absorbed	5.8	10.6	11.8	20.1
Communication disturbance	4.7	4.4	6.3	7.7
Anxiety	6.1	6.9	10.4	11.2
Autistic relating	4.9	5.4	5.0	5.8
Antisocial	0.6	1.1	1.1	1.7

Bax et al., (1995) studied the nature and prevalence of behaviour problems in 258 children with Mucopolysaccharide disorders. In Hurler's syndrome (mean age 6.5 years), aggressive/destructive behaviour was found in 8%, in Hunter's syndrome (mean age 8.6 years) aggressive/destructive behaviour was found in 42%, in Sanfilippo's syndrome such behaviour was found in 57%, and in Morquio's syndrome they were found in only 4%. Again, this highlights the high rates of such behaviour in the present study. In a study of 105 individuals with Williams syndrome (Gosch et al., 1997) (mean age = 77.1 months, (27-118)) 30% of Williams syndrome children were found to be disobedient, in comparison to 45% in the current sample of children with SMS. However, a higher rate of children with Down syndrome (65-77%) were described as stubborn and disobedient (Dykens and Kasari, 1997). Overall, emotional and behavioural problems appear to be more prevalent in SMS than in a number of genetically determined syndromes and samples of children with learning disabilities in general, for whom comparative data are available.

Very high rates of self-injurious behaviour were found in the present sample. Authors have suggested that the severity of self-injury in SMS may be associated with peripheral neuropathy in this group, Greenberg et al., (1996a). The gene abnormality associated with peripheral neuropathy is located near the SMS critical region on chromosome 17. The most common forms of self-injury reported in children in this study are presented in Table 33, where comparisons are made with a group of children and adults with learning disabilities (Oliver et al., 1987).

Table 33. A comparison of self-injurious behaviour in children with Smith-Magenis syndrome and individuals with Learning Disabilities

Self-injurious behaviour	Learning Disabilities n=616 (Oliver et al. 1987)	Smith-Magenis syndrome n=29 (Present study)
Pulling at nails/picking skin around nails	39%	93%
Hand/finger biting	38%	86%
Self-pinching/scratching/picking at sores	4%	72%
Hitting the head/body with the hand	36%	69%
Hitting the head on a hard surface	-	45%
Hitting objects to the head	28%	34%

While the rates in the present study are higher than those reported in children and adults with learning disabilities of unknown aetiology, some are equivalent to those previously reported in children with disorders of a biologically known cause. Self-injurious behaviour is reported in over 85% of individuals with Lesch-Nyhan syndrome (Christie et al., 1982; Anderson and Ernst, 1994), although this is still lower then the 97% prevalence of self-injury in the present study. Greenswag (1987) reported moderate to severe skin picking in 78% of adults with Prader-Willi

syndrome, with even higher rates (85%) reported by Clarke et al., (1989) in the same syndrome. The prevalence of skin picking has been estimated to be 52% in individuals with William's syndrome (Davies et al., 1998). These findings suggest that the rate of self-injury in SMS may not be so different to that observed in certain other biological syndromes. However, these findings do not exclude self-injury from the behavioural phenotype of SMS. In keeping with the criteria for the existence of a behavioural phenotype suggested in the first chapter, it is expected that there may be some overlap with other syndromes. Indeed, the finding of a 97% prevalence of self-injurious behaviour in children with Smith-Magenis syndrome shows sufficient internal consistency within the sample to be included as a phenotypic marker.

Previous studies have shown that the functions most commonly served by self-injurious behaviour in individuals with learning disabilities are escape from aversive situations and positive reinforcement (Iwata, Dorsey et al., 1994). The most common functions of self-injury reported in the current study were: to express anger and frustration, to gain attention, to express fear and anxiety, and to alleviate boredom. The range of triggers for self-injurious behaviour reported in children with SMS in the current study highlights the importance of carrying out a thorough functional analysis of self-injurious behaviour in order to implement appropriate and effective treatment programmes.

- **Implications and intervention strategies used by parents and teachers for the behavioural difficulties displayed by the sample**

Sixty-two percent of the present sample were found to be distractible, which has inevitable consequences in the classroom. Children with SMS would do better in smaller, calmer and more focused classroom settings. As the findings from the present study show, children with SMS are very adult-oriented, often requiring the teacher's attention, and when they do not receive this, aggression towards others, behavioural outbursts, tantrums and self-injurious behaviour are frequently the result. Transitioning from one activity to another and change in routine, at home or school, often precipitate behavioural outbursts. Consistency, structure and routine, and the use of visual cues would help greatly in this respect. Teachers identified various effective teaching approaches to manage the difficult behaviours displayed; giving rewards and praise to the child, a high child/teacher(support worker) ratio, and because children were reported to concentrate and to be less tired in the morning, to use this time of the day for teaching more difficult subjects. Other, less frequently reported strategies included: being firm and consistent, teaching in short blocks (a few minutes at a time, depending upon the children) and then having a break, repeating work and using computers to aid learning.

As with sleep difficulties, further work is urgently needed to develop effective intervention strategies to address the aggressive and self-injurious behaviours enlisted by children with SMS.

Strategies reported as effective by parents in terms of managing difficult behaviours included 'time-out', the implementation of firm, clear commands, and ignoring the difficult behaviours. Medication was only found to be helpful in 6 out of the 9 children who had tried it. The effective medications were Ritalin and Lithium. A more thorough analysis of the self-injurious behaviour and effective interventions are urgently required.

4.5 ATTENTION & HYPERACTIVITY

A study by the National Health and Medical Research Council, Canberra, in Australia (1997) reported rates of ADHD of between 2.3% and 6%, in the general population, depending on the diagnostic criteria used. Using DSM-IV, and limiting criteria to "hyperactive/impulsive' or "combined" types in the United States has given rates of 5-8%, in the general population (Levy, 1998). Hyperactivity in the general learning difficulties population was reported in 5.4% (Jacobson 1982).

In the present study, over 50% of the sample of children with SMS displayed: attention problems, in particular distractibility (62% of cases); impulsivity, especially in terms of difficulties waiting their turn (69% of cases); and hyperactivity, mainly in terms of being unable to remain seated during mealtimes (52% of cases). None of the children fulfilled the ICD-10 criteria for a diagnosis of Hyperkinetic disorder. These figures are much lower than previously reported findings of a 100% prevalence of hyperactivity in children with SMS (Smith et al., 1986, Stratton et al., 1984, deRijk-van Andel et al., 1991). However, the latter findings were based on small samples

sizes (maximum 6), and the authors did not assess the individuals themselves, but were reporting on previous health professional's findings. As such, the criteria on which the diagnosis was made are unclear. In addition, it should be remembered that criteria for diagnosing ADHD in this country tend to be much stricter than those employed in America. Hence the discrepancy between earlier findings and the present study in terms of rates of hyperactivity may be the result of stricter criteria and or more rigorous methodology used in the present study. The figures from the present study are higher than for children with Down syndrome, where only 15% were reported to display overactive behaviours (Gath & Gumley, 1986; Meyers & Pueschel, 1991). However, in other syndromes even higher rates are reported; for example hyperactivity or severe attention deficits have been reported in approximately 80% of males with fragile X syndrome (Baumgardner et al., 1995). Additionally, in a cohort of 146 children with Cri-du-chat syndrome, hyperactivity and distractibility were seen in 75-85% of the sample (Dykens and Clarke, 1997). Thus, although the rates of attention difficulties and hyperactivity were fairly high in the present sample, they were not as high as rates reported for various other syndromes.

4.6 AUTISM

The prevalence rate of autism in the general population has been estimated at around 5 per 10,000 (Lotter, 1996). On the Autism subscale of the DBC, Einfeld et al. (1998) reported the prevalence of autism to be 5.4% in a general population of children with learning disabilities. The figure for the children with fragile X in

Einfeld's sample was 4.9%, which is consistent with other studies on the prevalence of autism in fragile X syndrome (Paton et al., 1989; Piven et al., 1991; Bailey et al., 1993). Rates of autism in other genetic syndromes such as Down syndrome and Tuberous Sclerosis are considerably lower. In a study of individuals with Tuberous Sclerosis, Baker et al. (1998) estimated the prevalence to be 29%.

The present study found the prevalence of autism, in a sample of children with Smith-Magenis syndrome, to be 55% on the basis of the Developmental Behaviour Checklist, and 93% on the basis of the Schedule of Handicaps, Behaviour and Skills (HBS) assessment interview. The difference between prevalence on the two scales of autism used in the present study, might be explained by a concern regarding the HBS. A subjective element occurs in diagnostic ascertainment because of absence of guidance as to which questions in the HBS relate to which Autistic Disorders Diagnostic Checklist (ADDC) items. In some instances it is difficult to identify this relationship, and the presence or absence of an ADDC behavioural item has to be made on the basis of the experimenter's overall impression from the completed questionnaire. A revised version of the HBS, developed after this study was begun, addresses these issues. It may be that more individuals with SMS have been diagnosed as autistic in the present study than would be the case using a more conservative instrument such as the Autism Diagnostic Interview, (Le Coteur et al., 1989; Turk, personal communication, 1999), or clinical interviews (Baker et al., 1998). Clinical examination, in combination with a standardized assessment schedule would have improved the reliability and validity of the findings reported in the

current study, and again could well have resulted in lower prevalence rates for a diagnosis of autism. Nevertheless, the findings indicate high rates of autistic spectrum behaviours in children with SMS, when compared with the general population and with other genetic syndromes.

While accepting that the prevalence of autism may be an overestimate, the findings do provide helpful information on the characteristic behavioural profile of individuals with SMS in terms of their communication and social behaviours. One of the most commonly reported autistic type behaviours reported by parents and teachers of children in this sample was a desire to stick to routine. The social age from the HBS was 5.2 years. Unlike the case for 'typically autistic' persons, the children's communicative abilities were found to be less impaired than their social awareness and stereotypic behaviours. What particularly characterized their communication according to descriptions by their carers, was repetitive questioning and a tendency to talk about repetitive themes. If it is the case that individuals with SMS have difficulties in sequencing and short-term memory, as found by Dykens et al. (1997), then it may be that these particular autistic- like behaviours serve as a means for facilitating communication by supporting auditory short-term memory. This would be in contrast to the rather meaningless echolalia exhibited by individuals with 'typical' autism.

In the social domain, 62% of children with SMS were reported to lack appreciation of social requirements, and 45% were reported to approach strangers to introduce

themselves. Parents of 55% of the sample felt that their child made eye contact, but that at times the eye contact was inappropriate. However, 44% would look to a familiar person for reassurance. Interrupting conversations and a desire for attention has been reported in a large number of the sample. Deficits were seen in the ability to socialize with peers; the children were reported to prefer the company of either older more able individuals, or weaker children who can be dominated.

More rigorous research strategies and clinical observations should provide a more accurate estimate of the prevalence of autism in the population of children with SMS. Nevertheless, in terms of clinical practice, these findings suggest that once an individual has been diagnosed with SMS, s/he should receive a multi-disciplinary assessment for autism. This would facilitate a greater understanding of the individual's communication difficulties and needs by parents, teachers and health professionals, and would promote access to appropriate mental health and education services. The current study has also suggested a particular profile of autistic type behaviour for individuals with SMS. Further studies are required to shed light on this profile and the specific social and communication difficulties and needs of children with SMS, using comparison groups of autistic individuals from the general learning disability population and from groups of individuals with known biological disorders.

4.7 OTHER BEHAVIORUAL FEATURES

Previous authors have noted a characteristic self-hug, or upper-body squeeze in the majority of individuals with SMS (Finucane et al., 1994, Dykens et al., 1997, Dykens et al., 1998). The findings of the current study are consistent with those reported previously (a spasmodic upper body squeeze was reported by parents in 93% of children when pleased or excited), lending support to the suggestion of Finucane et al., (1994) that this behaviour is sufficiently characteristic of individuals with SMS to be used as a diagnostic marker.

4.8 IMPACT ON THE FAMILY

Families of children with disabilities are now acknowledged as undergoing increased stresses, but often as coping fairly well (Hodapp 1995). Family stress in other genetic disorders seems related to the child's maladaptive behaviours. In both Prader-Willi syndrome and Cri du chat syndrome - two disorders with extremely high levels of child maladaptive behaviour (Dykens and Clarke 1997; Dykens and Kasari 1997) - the child's behaviour problems are the single best predictor of familial stress (Hodapp et al. 1997a,b).

The General Health Questionnaire (GHQ) was administered to detect psychiatric disorders amongst the parents of the children with SMS in the present sample.

Twenty-eight percent of parents scored above the clinical cut-off, indicating a psychiatric disorder, and higher scores were associated with more severe sleep problems in the children. These rates are higher than those reported in other studies of families of children with Severe Learning Disabilities (Hoare et al., 1998) and are consistent with high levels of stress in parents of children with SMS, as a response to these children's severe behavioural and sleep difficulties.

Hodapp et al. (1998), in a study of 36 children with Smith-Magenis syndrome (mean age 8.35), found that the families of children with Smith-Magenis syndome showed high levels of both family stress and family support. Compared with two studies of young children with mixed aetiologies of disabilities (Dyson 1991, 1993), the Smith-Magenis group showed markedly higher levels of stress.

5. CONCLUSIONS

The findings of the present study show that rates of aggression, self-injury, sleep disorder, autism and certain maladaptive behaviours in children with Smith-Magenis syndrome are higher than the rate reported in the general population of individuals with learning disabilities of unknown etiology, and in other syndromes with a known biological cause. Although there is evidence of high rates of some of these behaviours in particular syndromes, the **combination** of difficulties and abilities identified in the present sample of children with SMS is indicative of a behavioural phenotype for the syndrome.

The combination of severe aggressive outbursts, self-injurious behaviour, extreme attention seeking behaviour and severe sleep problems, co-existing with high rates of autistic behaviour, make many children with SMS extremely difficult to manage. Hence an enormous amount of stress and burden is placed on the families of children with SMS, as combined by parental report and the results of a screening questionnaire for mental health difficulties. As such there is a great need for support and advice from Child and Adult Mental Health Professionals and Social Services for the families of affected children. There is also a need to develop and evaluate appropriate psychological and biological interventions to address the severe management problems posed by these children's behaviour difficulties. In particular, on the basis of the findings gleaned in this study, and as a next step, the present

investigator is planning to produce an advice booklet to inform parents/carers/teachers and health professionals about the cognitive and behavioural characteristics of the syndrome, and about intervention strategies that parents and teachers have found to be effective.

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APPENDIX 1

(i) Letter to the parents of the SMS Parent Support Group

Dear Parents,

I am a Clinical Psychologist working with children and families at the Mary Sheridan Child Development Centre in London. I am planning to carry out a study in order to obtain information about the abilities, educational needs and behaviour of children with Smith-Magenis syndrome. I hope to use the information gathered in this study to produce guidelines and advice booklets for families on appropriate ways of meeting your children's educational and behavioural needs.

I hope you will agree to participate in this study. It will involve my meeting with you at your home or other location that is convenient to find out about your child's progress and any aspects of behaviour or development that you may be concerned about. The interview will take approximately three hours. I would also ask to meet separately with your son or daughter for approximately 1 hour at home or at school, in order to undertake a number of assessments and activities to obtain information on his/her communication abilities and progress.

All information obtained will be treated with the strictest of confidence, and no individuals will be referred to by name.

I would be happy to answer any questions you may have about the study and can be contacted on the above telephone number. I would be grateful if you could return the enclosed slip in the prepaid envelope. I plan to undertake the interviews over the next 12 months and, with your agreement, will contact you to arrange a convenient appointment in the next few months.

I look forward to hearing from you.

Yours sincerely,

Carolyn Webber
Child Clinical Psychologist

(ii) Letter to parents via the Regional Clinical Genetics Centres

Dear Parent,

I am a Clinical Psychologist working with children and families at the Mary Sheridan Child Development Centre in London. I am planning to carry out a study in order to obtain information about the abilities, educational needs and behaviour of children with Smith-Magenis syndrome. I hope to use the information gathered in this study to produce guidelines and advice booklets for families on appropriate ways of meeting your children's educational and behavioural needs.

I have asked members of the Regional Clinical Genetics Centre who diagnosed your child as having Smith-Magenis syndrome to pass on this letter to any families they know of to see whether they would be agreeable to my contacting them to discuss the project further.

I hope you will be agreeable to meeting with me in order to complete some questions about your child's development and behaviour. I would also like your child's teacher to complete some questions about his/her behaviour and progress, and would also like to undertake some developmental and educational tests with your child. I would meet with you in your home or other location convenient to you for approximately three hours.

All information obtained will be treated with the strictest of confidence, and no individuals will be referred to by name.

I would be happy to answer any questions you may have about the study and can be contacted on the above telephone number. I would be grateful if you could return the enclosed slip in the prepaid envelope, and I will contact you to arrange a convenient appointment in the next few months.

I look forward to hearing from you.

Yours sincerely,

Carolyn Webber
Child Clinical Psychologist

(iii) Consent form

Name: (parent/guardian) _____

Name: (child) _____

Child's date of birth: _____

Address: _____

_____

_____

Home telephone number: _____

Work telephone number
(if agreeable to being
contacted there): _____

Convenient time(s) to be
phoned: _____

I agree / do not agree to _____ (child's name) being included in the study on abilities and behaviour in Smith-Magenis syndrome that is being conducted by Carolyn Webber.

I have had the nature of the study explained to my satisfaction. I understand that participation is entirely voluntary and that I am at liberty to withdraw from the study at any time without having to give a reason.

Signed: _____

Date: _____