Smith-Magenis Syndrome

This summary presents the common behavioural characteristics seen in individuals with Smith-Magenis syndrome. Every individual with SMS may not have all of these features. All children have differences in their personalities; individuals with SMS also show this variability in their personalities and have many endearing qualities, as well as some of the characteristics summarised below.

1) Intellectual Disability (ID)

Most people with Smith-Magenis syndrome (SMS) have an intellectual disability, typically in the moderate ID range [(1)]. Communication impairments are reported with speech delay greater than motor delay and weaknesses in expressive language compared to receptive language [(1,2,3,4)].

2) Sleep

- Sleep difficulties begin in childhood and carry on into adulthood and may change over time.
    i. ***Infancy:*** reduced sleep is reported despite parental beliefs that their children are good sleepers [(5,6).]
    ii. ***Childhood****:* Night-time and early morning waking, together with shortened sleep cycles are noted. Daytime fatigue is noted.
    iii. ***Older children, adolescents and adults:*** activity appears to be elevated early in the night suggesting it is difficult for children to get to sleep [(7)]. Increased age has been linked to reduced night-time sleep but increased shorter naps.
- Daytime sleepiness may account for some of the difficult behaviours shown by people with SMS [(7)]. Sleep difficulties are associated with an inverted melatonin release pattern (too much melatonin being released during the day and not enough at night) which disrupts circadian rhythm [(6)].
- Medication regimes which provide melatonin in the evening and suppress daytime release using beta blockers have been linked to improvements in sleeping of children with genetic disorders generally [(8,9)].

3) Repetitive behaviours

- Repetitive and stereotyped behaviours such as hand flapping, spinning/twirling objects or body rocking are very commonly reported SMS. The most common repetitive behaviours in SMS are teeth grinding and inserting hands and objects in their mouths [(10,11,12)] .

4) Attachment to favoured person

- 'Attachment' to particular people (shown by continually asking to see, speak to or contact a particular favourite person) is very common in SMS compared to other genetic syndromes [(13)]. Strong preference for adult attention (over that of peers) is also reported [(14)].

5) Challenging behaviour

- Challenging behaviour including self-injury and aggression (hitting, punching, biting, property destruction) is very common in SMS (with a prevalence rate of up to 70-96% for self injury and 70-87.5% for aggression) [(15,16)]. Self injurious behaviours commonly displayed by individuals with SMS are self hitting, hand biting and head banging. Behaviours such as skin picking, pulling out finger and toe

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nails (onychotillomania) and insertion of objects into bodily orifices (polyembolokoilamania) have also been reported [10, 12, 15 16]. These behaviours can serve multiple communicative functions [17].

6) Causes of challenging behaviour

- Challenging behaviour is more likely to be motivated by access to social attention than in other syndromes. Given the increased motivation for interaction with adults, negative behavioural responses may be shown if attention is withdrawn or directed toward others.

- Those with SMS are also more likely to display behaviours related to physical discomfort and pain [15] - *see health conditions section below*.

7) Impulsivity

- Impulsivity is more common in SMS than in others with ID without this syndrome and when compared to a range of other syndromes [18]. Caregiver accounts suggest that impulsive behaviour may be related to problems waiting for things (e.g. something the person wants or waiting their turn) (Clarke & Boer, 1998). Dykens and colleagues [13,19] found that impulsivity was reported by caregivers in 80-86% of individuals with SMS.

8) Emotional and behavioural dysregulation

- Research suggests that impulsivity may be related to difficulties delaying gratification (i.e. waiting for rewards) and that waiting in this situation can be emotionally aversive [20, 21]. Emotional control may be impaired in SMS (suggested by reports of emotional lability [10]), thus there may be problems controlling emotional responses to delay resulting in temper outbursts.

- Sloneem and colleagues [22] found that impulsivity was strongly associated with the severity of challenging behaviour (aggression). This supports the possible association between reports of 'explosive' temper outbursts in SMS and the impulsivity that is shown [22].

9) Strengths

- A number of positive features have also been attributed to those with SMS, including an "endearing" and affectionate personality and a strong sense of humour [23,24].

- Individuals with SMS have been found to have strengths in long-term memory, computer skills, perceptual skills and socialisation [15, 12].

10) Health conditions

- There are a number of health conditions, including vision and hearing problems that are associated with SMS. Information about these can be found on the following websites:

  i. Smith Magenis Foundation: http://www.smith-magenis.co.uk/index.php/health

  ii. Parents and Researchers Interested in Smith-Magenis Syndrome: http://www.prisms.org/us/living-with-sms/medical-management-guidelines

  iii. National Center for Biotechnology Information: http://www.ncbi.nlm.nih.gov/books/NBK1310/

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