

# Smith-Magenis Syndrome

Information and Advice for  
Caregivers & Professionals



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Acknowledgements: We are grateful to the Smith-Magenis Syndrome Foundation UK, who funded this updated booklet. We are also very grateful to the families who have generously shared photos of children and adults with SMS for us to include here and to the many families who participated in the research on which the information presented here is based. Finally we would like to thank the members of the Smith-Magenis Syndrome Scientific and Clinical Advisory Group who reviewed the content of this booklet.

# Welcome



This booklet is intended for families and caregivers of children and adults with Smith-Magenis syndrome (SMS), and professionals working with these individuals. We aim to guide you through the common features of the condition and explain how these may affect the development and daily life of someone with

this syndrome. We will also suggest ways of managing issues which might arise for the person with SMS in 'What might help?' sections.

Information is themed around specific topics that are relevant to caring for, or working with, someone with SMS. These are outlined in the index.



While the issues outlined here are more likely to affect people with SMS, it is important to remember that every person is an individual. Each person with SMS will have his or her own life experiences, likes and dislikes and strengths and weaknesses. Every family's experience of raising a child with SMS will be different. Not even

any issue outlined here will be relevant to every person with the condition, and it is important to be guided by an understanding of the person, in addition to the syndrome.

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# Newly diagnosed



If you are reading this guide as the caregiver of a child or adult newly diagnosed with SMS you may be experiencing many different emotions, some of which may be very difficult.

Being told that your child is different and finding out that he or she is likely to face challenges that other people may not face can be hard. Some families who already had concerns about their child's development find receiving a diagnosis a relief, as it explains some of their worries. But following this, there may be concern about what this diagnosis means for the child and his or her future. This is particularly the case where information available online or via the medical profession focuses on the difficulties associated with the syndrome.

When reading this guide, it is important to bear in mind that every child and adult is unique. Not every person will show all of the characteristics described here. A person with SMS may have more in common with his/her brother or sister than with another person with SMS; they may look

like them, enjoy the same games and films as them and hate the same food as them!



However, knowing about the common features of SMS means that it may be possible to introduce strategies early to intervene and prevent difficulties from arising or getting worse

The following description of a 9 year old boy with SMS was included in the first UK guide for caregivers and professionals; it

captures a common experience of raising a child with SMS:

*“My child is the most loving, most loved, joyful, enthusiastic, empathetic, predictable and rewarding child I know. He can also be the most entrenched, frustrating, unintuitive, quirky and unpredictable. He is not a simple child.*

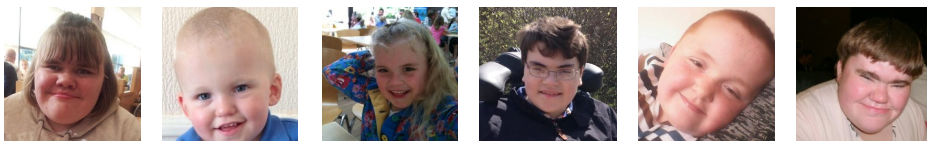
*There is a wide spectrum of characteristics associated with Smith-Magenis syndrome (SMS) and we as a family, have encountered many of these. It's been very challenging. I've learnt a lot. About creativity, about patience, about other people and their attitudes. About local education authorities. And it's got better. As each issue arose, we found a way around it. The grief lessened as the child grew. We realised the importance of working with his school and therapists. Our lives have changed immeasurably, yet we believe that we are no less happy as a family because of these challenges.*

*We found that the label of SMS is a description of his symptoms, not of him. Learn about your child first - he will be gifted in many ways. And take one day at a time.”*

# Syndrome Overview

Smith-Magenis syndrome (SMS) is a genetically determined condition, associated with intellectual disability, developmental delay and a pattern of distinctive behaviours. It was first described in the early 1980s in the United States by Anne Smith, a genetic counsellor & Ellen Magenis, a paediatrician and genetics expert.

It is believed that SMS affects at least 1 in 25,000 people, but this figure may be closer to 1 in 15,000 people due to under diagnosis.



## Inheritance

SMS is usually caused by a deletion on one copy of chromosome 17. The missing region contains several genes, but the loss of one particular gene, *RAI1*, is thought to be responsible for most of the characteristic features of this condition. A small percentage of people with SMS have a mutation in the *RAI1* gene rather than a chromosome deletion. Most cases of SMS are sporadic, or a one-off, within the family. However, very rarely, an apparently unaffected parent may carry the deletion or mutation in some, but not all, of their

germ cells (eggs or sperm). This is not something that can be tested for in the parent. Non-affected siblings of a child with SMS are not at increased risk of hav-

## What might help?

*If a couple were concerned about the possibility of having a second child with SMS - In this situation prenatal diagnosis could be undertaken in a second pregnancy. This would only be available if the diagnosis of SMS in an existing child had been confirmed by chromosome or gene testing.*

ing an affected child.

### Common features

People with SMS tend to have similar physical features and facial appearance, including a broad face and nasal bridge and a flattened mid-face. The upper lip characteristically has a Cupid's bow shape, and the corners of the mouth may be downturned. When children are young they have a 'baby-faced' appearance, described as 'cherubic', and facial features tend to become heavier as the children get older. Other physical features include short broad hands, inbent fingers, small toes and a hoarse deep voice.

Most individuals with SMS have some degree of intellectual (learning) disability, ranging from mild to severe. Most show moderate levels of intellectual disability. Speech development is markedly delayed, in particular their ability to express themselves (when compared with their ability to understand speech).

People with SMS are frequently described as loving and caring, with a good sense of humour and an eagerness to please. They tend to be friendly and outgoing, and to like adult attention and enjoy interacting with adults. Some people with SMS show a characteristic spasmodic upper-body squeeze, or "self-hug", when excited or happy.

Sleep problems are very common in SMS, for example difficulties falling asleep, frequent and prolonged night waking, early waking and excessive daytime sleepiness.

Self-injurious behaviours (e.g. head banging, hand biting) are more common in people with SMS than people without this syndrome and can become severe in some people. Other potentially difficult behaviours associated with SMS include aggression and temper outbursts, repetitive behaviours, impulsivity/inattention/hyperactivity and attention-seeking behaviours. Some people fulfil the diagnostic criteria for autism spectrum disorder (ASD).

In the following pages we describe the key features and behaviours characteristic of the syndrome in greater detail, and in the 'What might help?' sections we present suggestions for addressing many of the difficulties the children and adults present with.



# Abilities

## Cognitive Abilities

Levels of cognitive ability in SMS can vary quite a lot, but nearly everyone with the syndrome will have an intellectual disability, which is most often in the 'moderate' range.



There are reports of a small number of people with SMS whose cognitive abilities fall in the 'low-

average' range or within the 'borderline' range. With improved testing, less severely affected individuals with SMS may increasingly be identified and so the range of cognitive abilities may increase. Abilities do not appear to decline with age in SMS<sup>1</sup>.

In an early study of adults with SMS, reading skills were found to reach a 6-7 year

old level<sup>1</sup>. Caregiver report, however, indicates that some adults with SMS fall well below this range, whilst others far exceed it.

Cognitive abilities in SMS can be uneven, with relative strengths in some areas and

Strengths	Weaknesses
Long term memory	Short term/ working memory
Perceptual skills	Visual attention
Computer skills	Sequential processing

## What might help?

*Consider the person's profile of strengths/weaknesses* - These may inform the planning of educational programs for individuals with SMS to harness strengths. Encourage use of visuospatial cues, gestures or signing, and picture symbols to support verbal instructions. Encourage access to

<sup>1</sup>Udwin, Webber & Horn (2001)

computers/tablets and suitable programs and applications (apps). This can teach skills, be a relaxing 'chill out' activity and act as a shared interest with mainstream peers. Time with a computer/tablet can also be used as a reward for desired behaviour.

*Tailor approaches to reading* - Approaches which rely on visual memory of how a word looks (such as the 'Look Say' approach) are likely to be more effective than approaches which rely on auditory skills, which emphasise a phonetic approach. It is important to show the person that printed words are meaningful; they tell a story and impart information. Where people are preoccupied with particular objects or topics, introducing reading materials on these topics may enhance their interest and motivation to read.

weaknesses in others, as shown in the table below.

## Communication

Communication difficulties are common in SMS, with speech delay being greater than motor delay. Weaknesses in expressive language when compared to receptive language are also common.

Verbal comprehension has been found to be a relative strength in individuals tested formally using IQ tests. Children and adults with SMS may therefore un-



derstand more of what is being said but find it hard to produce verbal responses. This may result in frustration when individuals are unable to make their thoughts and feelings known, which may, in turn, impact on behaviour.

It is thought that difficulties with speech production might be related to oral-motor dysfunction, which is common in people with SMS. Hearing problems and ear infections may also affect the development of speech (see the 'Health' section).

## What might help?

*Check hearing* - Early identification of hearing problems may reduce their negative impact on communication.

*Use communication aids*

*Signing* - Used alongside speech, signing improves language development

and helps reduce frustration. Family members should be encouraged to learn Makaton or other simple signing systems.

*Pictures* - The Picture Exchange Communication System (PECS) approach, which involves using pictures to request things, may be useful, particularly for those with an additional diagnosis of autism spectrum disorder (ASD).

*Technology* - There are various apps available for mobile phones and tablet computers which aim to aid communication. They can enable users to customise how they communicate using pictures, photos, symbols and audio.

*Use a communication passport* - These are person-centered booklets containing information about the individual, including how best to communicate with them, and their preferences and views, which may aid communication for individuals with verbal difficulties.

*Seek professional input* - If there are specific concerns about communication, then it is important to pursue a referral to speech and language therapy services via the child's paediatrician and/or school.

## Adaptive Abilities

Adaptive abilities are the skills needed to get along in everyday life, from dressing and toileting to living independently and managing money. In SMS these types of abilities seem to be less well developed than may be expected based on either age or cognitive ability.



Older individuals are described as being quite dependent on the people around them and unable to complete tasks

such as dressing, cooking or travelling independently, which inevitably has an impact on their ability to live and work independently<sup>2</sup>. This picture may change, as many individuals who have been diagnosed more recently are less severely affected and thus may grow up to lead more independent lives.

Some areas of adaptive functioning are stronger than others; for example, socialisation (the ability to get along with others) has been found to be a relative strength. Interestingly, while aspects of socialisation and adaptive communication skills are reported to increase with cognitive ability, daily living skills are not

strongly related to cognitive ability. This supports the suggestion that there may be a mismatch between cognitive ability and achievement in everyday life skills. It has been suggested that this difference between cognitive ability and adaptive behaviour may result from some of the difficult behaviours associated with SMS such as impulsivity, temper outbursts and 'attention-seeking'.

### What might help?

*Use visual prompts, visual timetables or plans for everyday tasks* - These help to sequence activities and reinforce verbal instructions which are given. They serve as a memory aid and provide additional information that may be lost for those with poor hearing.

*Encourage independence* - Encourage the person with SMS to do things for themselves where possible, let them take part in activities around the house such as cooking, tidying up and planning activities.

*Plan for developing future skills* - Explore options for courses or placements for older adolescents and adults that may enable the development of independent living skills.

*Seek professional input* - Where a per-

son's self-care abilities are limited, Social Services may be able to offer support with personal care (e.g. washing, feeding). They can also provide access to equipment and adaptations to increase accessibility in the home which may provide more opportunities for independent living, where assessments carried out by occupational therapists suggest these would be beneficial.

<sup>2</sup>Udwin, Webber & Horn (2001)

# Schooling

Children with SMS have an unusual pattern of abilities (see the 'Cognitive Abilities' section) and this, together with behavioural problems (see the 'Behaviours that Challenge' section), can result in difficulties finding an appropriate school. There is no one type of school that is ideally suited to the needs of a child with SMS.

Education authorities differ greatly in the way they allocate resources for children with special educational needs (SEN); finding the most appropriate school will depend on the individual child's level of ability, their profile of strengths and weaknesses, and on the provisions in the particular schools that are available locally.

Increasingly, there is an emphasis on inclusion for children with SEN, especially for children who are of primary school age. In essence, this means placing children in mainstream schools (with or without additional support) or in units attached to a mainstream school. Caregivers may or may not feel that this is the

best option for their child; it is important to consider the child's profile of abilities and needs to inform this decision (academic and social abilities and behavioural issues).

Many children with SMS need the slower pace and more individualised attention that is currently provided in special schools and units. In all cases a structured environment with predictable routines is



recommended.

In a study including school aged children with SMS<sup>3</sup>:

- 35% attended schools for children with severe intellectual disability.
- 21% attended schools for children with moderate intellectual disability.
- 21% were in residential/boarding schools for children with an intellectual disability.
- 10% attended schools for children with mixed levels of intellectual disability.
- 7% were in a mainstream school.
- 3% attended a special class in mainstream.
- 3% attended a language unit.

This study was carried out quite a long time ago (the data were published in 2001) and it is quite possible that things have now changed, given changes in legislation and increasingly frequent diagnosis of less severely affected individuals. It remains the case, however, that most people with SMS are likely to be in special schools or units, or at least to require considerable additional support in the school setting.

<sup>3</sup>Udwin, Webber & Horn (2001)

## What might help?

*Early support* - Portage is a home-visiting education service for young children with additional needs which may be available to pre-school children. Provision may depend on geographical location (see the 'Sources of Help and Useful Contacts' section). For early education outside of the home, it is important to ensure that a nursery or preschool placement is able to provide the type of additional support that a child with SMS will need, this may include extra support during sessions and access to additional services.

*Provide schools with information about SMS* - Giving this booklet to school staff and other professionals who work with children with SMS, together with any additional resources that have been useful, may provide valuable information about difficulties that may be experienced by someone with SMS and how to help. Furthermore, it will be useful for teachers to understand the possible causes of some of the behaviours shown (see the 'Behaviours that Challenge' section).

*Provide schools with information about the child with SMS* - It is important for teachers to be aware of the child's

strengths, as well as weaknesses. This can inform planning of educational programmes for children with SMS.

*Teaching approaches can be used which encourage the child's areas of strength -* Visuospatial cues, gesture or signing, and picture symbols can be used to support oral teaching methods.

*Adapting the classroom & school environment -* Schools should consider how they can adapt the school environment to best support the child. This might include:

- Keeping the environment and voices calm.
- Keeping background noise to a minimum to avoid distraction and reduce anxiety levels.
- Planning where the child will sit and who they are next to.
- Providing a safe area if behaviour becomes challenging (see the 'Behaviours that Challenge' section).
- Providing somewhere to rest (see the 'Sleep' section).
- Managing transitions to avoid distress (see 'Repetitive Behaviour and Restricted Interests',

'Preference for routine' subsection).

- Ensuring support is also provided outside the classroom. This includes enough support with toileting and during lunchtime and breaks. Playgrounds can be noisy and intimidating; there may be ways for schools to reduce this e.g. assigning a 'buddy' who will look out for the child.

*Monitor progress and seek additional support and assessment -* Progress against goals should be monitored by schools and colleges and also caregivers, to identify where support is effective, and where further support is needed. This requires effective communication between caregivers and schools/colleges. Plans should be dynamic and support should be flexible to accommodate changes as they develop. Additional support may come from a variety of services including educational psychology, occupational therapy, speech and language therapy, Social Services, a Child and Adolescent Mental Health Service or Children with Disabilities Team.

*Plan for the future -* Looking towards the future is important at each stage of education. This may involve consider-

ing the child's level of ability and skills, their interests and the wider context (geographical location, funding resources), to plan for what type of school/ post school placement would be best for them.

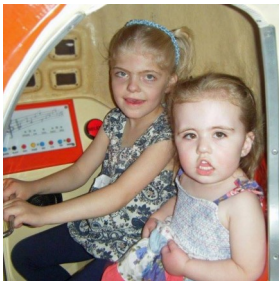


# Social Relationships and Autism Spectrum Disorder

## Social Relationships

People with SMS are often described as very friendly and outgoing, and most find social attention particularly rewarding. Many develop strong social relationships at home, school/college and in the wider community. This has benefits for the well-being and happiness of the person with SMS and can also be useful when planning programmes that aim to develop skills and behaviours, where social attention may be used as a reward.

In common with many people who have an intellectual disability, people with SMS can often lack understanding of the underlying, 'unwritten', rules governing social interactions



(e.g. social/personal space) and may not be aware of the social constraints that are apparent to others. As such, they may approach strangers in an over-friendly manner. Understandably, this can be a major concern for caregivers, who fear that their child is too trusting and may be taken advantage of if not supervised all the time.

People with SMS may also communicate with others by interrupting their conversations and by using repetitive questioning. They also have a tendency to talk about repetitive themes (see the 'Repetitive Behaviour and Restricted Interests' section).

Having an intellectual disability, and often an endearing personality too, may mean that adult friends and relations may unintentionally 'baby' or 'overindulge' people with SMS. It is important to explain to these adults how to behave appropriately towards the individual with SMS while being as tolerant and patient as possible in relation to any difficulties he/she may have. Being able to explain clearly what a person with SMS is capable of (and what

they might struggle with) can help with this.

Specific difficulties with social behaviour in SMS are found in two main areas; 'attention-seeking' behaviour and features of autism spectrum disorder (ASD).



### 'Attention-Seeking'

'Attention-seeking' behaviour is commonly described in people with SMS. This seems to be best characterised as a strong drive for the attention of particular people.

People with SMS are described as often wanting to contact or interact with particular 'favourite people'. This behaviour has been reported by caregivers and teachers, suggesting it is shown both at home and in other settings.

While 'attention-seeking' is often shown towards caregivers, it can also be siblings, teachers or even the bus driver who become the object of this behaviour. In children, this seems to be directed more to-

wards adults than towards peers of their own age.

It is likely that this attention-seeking behaviour is more striking in SMS because of some of the other behaviours that are commonly shown; for example there are often reports of temper outbursts, aggression or self-injurious behaviour (all of which are more common in SMS) being shown when attention is unavailable. This will make the behaviour stand out more and can make it more difficult to manage. For caregivers, being the focus of persistent demands for attention may become especially stressful.

### What might help?

*Encourage diverse social relationships* - Encouraging relationships with a range of different people may reduce the intense focus on getting attention from just one or two specific people and may provide many opportunities to practice and improve social skills. Social Services may be able to provide assistance to attend social activities such as youth clubs.

*Facilitate play and social skills* - Caregivers and teachers can encourage and teach social skills, including turn-taking and sharing. Some caregivers recom-

mend 'scripting', whereby the person is taught appropriate social phrases to use. 'Social stories' (made up stories about social situations, usually with pictures) are another useful set of techniques.

*Explain who he/she will be with during the day* - Communication tools, such as visual timetables, can be useful to let people know who they will be with throughout the day; they can also help with plans and expectations, and reduce negative responses to change (e.g. if a preferred member of staff is off ill). They may also help with repetitive questioning about what is happening and with whom, as the person can be directed to the timetable.

*Explore the availability of respite* - Usually accessed through Social Services, respite enables the person with SMS to spend time away with another family, or in a local residential unit, at regular intervals. Caregivers can have a well-earned break, as well as providing the person with SMS with the opportunity to meet new people and enjoy new experiences and new routines.

## Autism Spectrum Disorder

Autism or autism spectrum disorder (ASD) affects social interaction and communication and is associated with restricted and repetitive patterns of thought, interests and behaviours, as well as problems with imaginative and make-believe play and often multiple sensory sensitivities. A diagnosis of ASD is more common in SMS compared to other people with an intellectual disability, although it is not more common than in a number of other genetic syndromes.

Estimates of ASD in SMS vary, but quoted rates are usually over 50%. The source of the variability is probably due to how characteristics of ASD are measured. Results from questionnaires completed by caregivers are usually reported; less is known about rates of formal, clinical, diagnoses.

It has been suggested that some people with SMS may receive an ASD diagnosis mainly because of language abnormalities and repetitive behaviours rather than social impairments. As such, the profiles of behaviour may differ from ASD where there is not an identifiable underlying cause (which accounts of the majority of cases of ASD). Socialisation skills have

been described as a relative strength in SMS (compared to daily living skills for example), whereas in individuals with ASD where there is not an identifiable underlying cause, socialisation is a relative weakness.

While it is unclear exactly how common ASD is in SMS, there is good evidence that a number of issues relating to ASD are particularly prevalent in SMS. This includes repetitive behaviour and sensory issues (both of which are covered in separate sections in this booklet). Even for individuals with SMS who don't qualify for a diagnosis of ASD, there may be a range of developmental challenges relating to social skills and social relating; difficulties with language and communication, obsessional tendencies and difficulties with imaginary and make-believe play. All these issues need to be addressed, whether or not a formal diagnosis of ASD is given.

Given several case reports of individuals with SMS receiving formal diagnoses of ASD, it is clear that a diagnosis of SMS should not preclude the possibility that a person will fulfil the criteria for ASD. If a person struggles with issues relating to ASD symptomology, then receiving a diagnosis may help to identify strategies for altering the environment to make life easier for that person, and facilitate ac-

cess to services and funding. Having SMS describes the underlying *cause* of the individual's developmental and behavioural challenges, while ASD is a way of describing a pattern of behaviours that appear to occur together.

### What might help?

*Explore resources for ASD and see what works* - There are now many resources available which provide strategies for managing a range of issues associated with ASD; for example, repetitive behaviour, communication and social relating. The National Autistic Society website (see the 'Sources of Help and Useful Contacts' section) is a good place to start.

*Consider visual timetables* - These are commonly used ASD resources and can be very useful for people with an intellectual disability more generally too. They serve as a visual reminder of the plans for the day and activities where there are issues with remembering. They can also provide reassurance about what is coming next, which can reduce repetitive questioning around this topic.

*Communication facilitation is important* - There are a number of evidence-based

strategies including the Picture Exchange Communication System (PECS) and Makaton (a simplified sign language). See the 'Communication' section for further details.

*Seek professional input* - If there are concerns that a person with SMS has ASD, then their diagnosis of SMS should not preclude formal assessment for possible ASD and an assessment should be requested via the GP, Child Development Centre, Child and Adolescent Mental Health Service or Adult Learning Disabilities Service if the family is already in contact with one of these services. Where ASD is diagnosed, this diagnosis can provide valuable access to resources and support packages. It can also enhance understanding of the individual's social, language, play and other challenges, and facilitate appropriate educational support.

# Sensory Issues

People with SMS may have unusual responses to sensory input<sup>4</sup>. Sensory input can come from many different aspects of the environment including noises, smells, sounds, lights and images, tastes, textures and feelings. Sensory input is also involved in balance and coordination.

Difficulties processing sensory input may be described as 'sensory integration' problems. Such difficulties might be more common in people with SMS who have autism spectrum disorder (ASD), where sensory sensitivities are common, however these difficulties may also be shown by those who do not have ASD. Unusual sensory responses may include avoiding or seeking out sensory stimulation. Behaviours which provide sensory stimulation may be described as 'self-stimulatory'.

Some of the repetitive behaviours which are very common in SMS (e.g. rocking, spinning, fiddling and chewing) may provide sensory input for the individual engaging in them. If a person *seeks* sensory input then there are a wide range of sen-

sory toys and objects available that might provide the stimulation that they enjoy. Some types of sensory-seeking behaviours may not be appropriate or may put the person at risk (e.g. mouthing inappropriate objects) and effort should be made to reduce these.

Hyperacusis (extreme sensitivity to noise) is more common in SMS, so difficult behaviour when exposed to certain noises or avoidance of noisy environments may be an issue. Sensory sensitivities may also impact on feeding, and may result from, or be compounded by, oral sensorimotor



<sup>4</sup>Hildenbrand & Smith (2012)

## What might help?

*Provide sensory stimulation* - There are a huge range of sensory toys and objects available to provide input across the different senses. For example, for those who seek sensory stimulation through mouthing objects or hands, chewy rubber objects can be bought in the form of 'chewy tubes', wristbands, necklaces etc. Many schools have sensory rooms specifically designed to provide pleasant sensory input. It may be a process of trial and error to work out which things a person is most interested in. The children's charity Cerebra has a postal toy library where families can borrow toys to see if their child enjoys them (see the 'Sources of Help and Useful Contacts' section). Occupational therapists in Child Development Centres will also be able to suggest appropriate sensory toys.

*Adapt the environment* - Try to identify any specific stimuli which the individual finds aversive, and manage situations where these are present accordingly, (e.g. if noise is aversive then ear defenders could be used in noisy environments). Changing routines and environments to avoid aversive situations (e.g. whole class transitions) may also be effective. For stimuli that can-

not be avoided, systematic desensitisation programmes (involving gradual, systematic exposure to the specific aversive stimuli) may be useful (see 'Seek professional input').

*Access information resources* - There is a lot of information about managing sensory issues in relation to ASD, which may be of use when thinking about environments and activities, even if a person does not have an ASD diagnosis.

*Seek professional input* - Occupational therapy (OT) may be beneficial for people with SMS with sensory issues. Occupational therapists can carry out assessments of people in their everyday environment, looking at their skills, their environment and the activities that they do (e.g. everyday activities such as dressing and eating). An occupational therapist who is trained in Sensory Integration may be able to offer specialist advice on this issue. One approach suggested for managing sensory issues is a 'sensory diet'. This is an approach which aims to maintain the level of a person's stimulation during the day, using a planned programme of activities which is tailored to the individual. The occupational therapist may also be able to assess

and advise on risk management and interventions such as the use of technology may be recommended. A clinical psychologist may also be able to provide input, for example, around systematic desensitisation (see above).

If caregivers think there might be an issue with oral sensitivities and/or oral sensorimotor difficulties which are impacting on feeding then they should



# Behaviours that Challenge

People with SMS tend to show more behaviours which may be described as 'challenging', when compared with people with intellectual disabilities who do not have SMS. These behaviours include temper outbursts, aggression and self-injury.

'Behaviours that challenge' or 'challenging behaviours' are commonly used terms for such behaviours, however some people feel that this terminology suggests that the person showing the behaviour *intends* the behaviour to be difficult. Importantly, when these behaviours are shown, the person him/herself is not being challenging, rather it is the behaviour which may be distressing and may possibly cause harm. It is important to recognise that these behaviours may act as a form of communication, for example, as a means of expressing distress or discomfort where alternative means of communication are not available to the individual.

Furthermore although these behaviours are more common in people with SMS

than would be expected by chance, this does not mean that they are inevitable. Not all people with SMS will show these behaviours, and for those who do develop them there are strategies that can be effective in reducing the behaviours.

## Temper Outbursts

For some people with SMS, temper outbursts can be a problem. These can be difficult to contain and may go on for prolonged periods of time.

They are characterised as sudden outbursts of negative emotion and can involve a range of behaviours, such as crying, shouting, self-injury, aggression or destructive behaviour. It is important to remember that these outbursts are unlikely to be fully under the control of the person showing them, illustrated by the fact that many people with SMS express remorse after an outburst. Lack of control is also described in caregiver descriptions 'meltdowns' which they distinguish from tantrums. These meltdowns are described

as not being 'goal driven', and can occur in response to overexcitement as well as frustration.

## Aggression

Aggressive behaviour is often described in SMS, although the reported frequencies of these behaviours vary considerably (probably partly because of the different types of behaviours which might be described as 'aggression' e.g. verbal aggression, physical aggression).

When talking about aggression, it is important to be aware that it is not suggested that the person showing the behaviour *means* to cause harm.

Common estimates suggest that around 70 to 90% of people with SMS show aggressive behaviour. These rates are higher than other people with an intellectual disability. This suggests that aggressive behaviour is a specific issue associated with SMS that caregivers and professionals should be alert to.

The types of aggressive behaviour shown by people with SMS seem to be similar to those shown by other people with an intellectual disability and include hitting, kicking, grabbing, punching, and property

destruction. Other behaviours include biting (which, may in some cases be related to mouthing) and, anecdotally, 'strong hugging' and the poking of other people's eyes. Biting and hitting specifically seem to be more common in SMS than in other people with an intellectual disability.

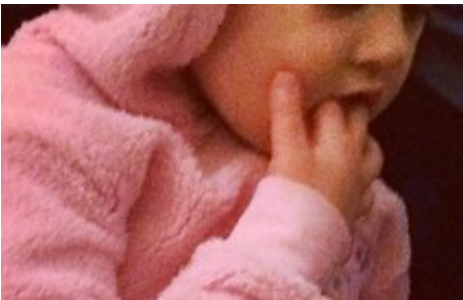
While aggression does seem to be shown more often by people with SMS, when it occurs it does not appear to be more severe than that shown by others with an intellectual disability. More severe aggression may be shown by those who are more hyperactive, by those who have more behaviours related to autism spectrum disorder (ASD), and particularly by those who are more impulsive, who may have difficulties 'putting the brakes' on a behaviour once it has started.

## Self-injury

Self-injury is non-accidental behaviour initiated by a person which causes them harm (e.g. head-banging, hand-biting, scratching). Up to around 90% of people with SMS will show behaviours which can be described as self-injurious, and these behaviours have been reported from as early as 18 months of age.

There is agreement that rates of this behaviour in SMS are much higher than in people with an intellectual disability who don't have SMS. The increased likelihood of these behaviours developing means that it is especially important to be aware of the types of behaviour shown, and what might influence these behaviours, in order to help with their management.

Different people may show different behaviours and the behaviours shown by the same person may vary over their lifetime. The types of self-injurious behaviour shown include those that are quite commonly exhibited by people with an intellectual disability e.g. self-hitting, and also rarer behaviours e.g. picking at finger and toe nails (onychotillomania) and inserting objects into bodily orifices (polyembolokoilomania). Self-hitting and self-biting (often hands) are among the most commonly shown self-injurious behaviours in SMS.



### What might help? General suggestions

General suggestions for managing behaviours that challenge are outlined below. The appropriateness and effectiveness of these techniques will depend on the function of the behaviour (i.e. what the behaviour 'achieves' for the person showing it). Although it is often difficult to establish what might be acting as a trigger for behaviours that challenge, it is worth observing the person carefully during any outbursts, to try to identify situations and events that may trigger them. It may be useful to keep a chart, noting down what happened immediately before the behaviour and what the behaviour was.

When dealing with difficult behaviour try to think about the situation from the point of view of the person showing the behaviour. Praise and reward positive behaviour and take as little notice as possible of the difficult behaviour. Ignoring can be an effective way of managing difficult behaviour, except when the person is being destructive or is injuring themselves.

*Manage temper outbursts* - If self-injury, aggression or other difficult behav-

aviours are shown during temper outbursts, effective early management of the temper outburst may reduce some of these accompanying behaviours (see 'Techniques for reducing severity of temper outbursts').

*Facilitate communication* – Use of tools for facilitating communication and, if necessary, seeking input from speech and language therapy services and working with schools or placements (see the 'Communication' section), may enable people to communicate their feelings and make requests more efficiently. This could potentially reduce self-injury or aggression which has become associated with communicating with caregivers and eliciting rewarding responses.

*Reinforce waiting* - If difficult behaviour occurs in situations where waiting is required, it may be helpful to develop waiting skills. Visual timers and graded exposure to increased time for waiting with rewards can build up these skills. It may also then be useful to teach a sign or use a picture card for 'wait' and use it frequently, rewarding 'good waiting'.

*Facilitate sleep* – See the 'Sleep' section for more information, but specifically here, allow people to take a daytime

nap both at home and school or an adult placement if necessary to avoid putting demands on a severely fatigued person, which may result in a temper outburst, self-injury or aggression.

*Identify and address pain* - There is good evidence that pain can increase the risk of showing self-injury and aggression. Ruling out painful conditions in those with SMS who are not able to effectively tell their caregiver that they are in pain, is therefore important. Tools have been developed which enable caregivers to assess the presence of pain by observing behaviour (see the 'Health' and 'Sources of Help and Useful Contacts' section). These may be useful when considering whether pain might be a potential cause of behaviours that challenge.

*Seek professional input* - If behaviours cannot be managed using strategies implemented by caregivers, it may be necessary to seek input from psychology services, which may initiate assessment of the behaviour (including identifying the function of the behaviour) and suggest management plans. One approach used by psychologists to reduce behaviours that challenge is differential reinforcement techniques.

Differential Reinforcement of Other behaviour (DRO) is where a reward (reinforcer) is provided when a specific problem behaviour is not shown. Differential Reinforcement of Incompatible behaviour (DRI) is another approach, where a reward is given when a behaviour is shown which is incompatible with the problem behaviour. These are just two approaches which have had some success in reducing rates of problem behaviours in people with an intellectual disability. There are a number of other approaches which may be effective in managing behaviour, and choice of approach will depend on a number of factors including the results of initial assessments.

When mood or behaviour disorder is very severe, impacting on well-being very significantly, and the person has not responded to psychological, educational and social interventions, then use of medications, prescribed and monitored carefully by a suitably qualified and experienced clinician, can assist when provided in combination with other non-pharmacological interventions. Medication must never be used as a substitute for other interventions being unavailable. This is a highly specialist area of clinical practice and it

is vital to ensure that the individual is being helped by the right clinician.

Social Services may be able to provide support for families where behaviour problems are having a severe impact on family life (e.g. providing respite care),

What might help temper outbursts?

*Techniques for reducing severity of temper outbursts* - The severity of behaviours shown during temper outbursts may be reduced using a variety of techniques. These include distraction (redirecting the person's attention towards something else) and de-escalation (using calming, verbal and non-verbal responses to reduce anger levels). Using humour (e.g. making a favourite funny action or joke) can be effective both as distraction during a temper outburst and can also de-escalate a situation. Teaching self-calming skills (things that a person can do themselves when they become angry that may calm them, e.g. moving to a quiet space) may also be effective.

What might help aggression?

Aggression often occurs during a temper outburst; therefore strategies described previously in the 'What might help temper outbursts' and 'What might help? General suggestions' section may also be effective in reducing aggression. The following are suggestions that may be useful in managing aggression specifically.

*Reduce mouthing* - If biting is thought to be related to mouthing (inserting hands or objects into the mouth) and the sensory input this provides, the suggestions in the 'Sensory Issues' section may be useful in reducing this.

*Wear clothing that protects against specific behaviours* - In the short-term, wearing clothing which protects specific areas of the body may reduce the impact of the behaviour. It may also enable those around the person to respond less to instances of aggression. This might reduce potentially rewarding responses which can be inadvertently provided to a person showing the behaviour e.g. moving away, stopping a task or telling them to stop. For example, if pinching is an issue, long sleeves worn by the caregivers might help to lessen the impact of this. Input from psychology services, Child and Adolescent Mental Health

Services or Adult Learning Disability services should be sought for severe aggression which poses a risk to safety and well-being.

### What might help self-injury?

In addition to the suggestions regarding managing challenging behaviours outlined previously in the 'What might help? General suggestions' section which may be applicable to managing self-injury, the following are suggestions that may be useful in managing specific types of self-injury. Additional detailed information about self-injurious behaviour can be found in the briefing on self-injurious created for the charity Cerebra (see the 'Sources of Help and Useful Contacts' section). Input from psychology services, Child and Adolescent Mental Health Services or Adult Learning Disability services should be sought for severe self-injury which poses a risk to health and well-being.

*Nail-biting*: Painting the nails with 'Stop n gro' (an unpleasant, non-toxic fluid marketed to stop nail biting) or making a fuss of painted nails may help to deter nail-biting.

*Self-biting*: Mouthing can be very com-

mon in SMS (see the ‘Sensory Issues’ section) and in some circumstances (but certainly not all) this mouthing may be ‘shaped’ into biting. For example, where mouthing results in a rewarding response (e.g. social attention or removal of a non-preferred task), this may make that behaviour more likely to reoccur. Providing an alternative item which is safe to mouth and provides sensory feedback, such as the rubber chewy objects available (which may be purchased in the form of ‘chewy tubes’, wristbands, necklaces etc.), may reduce mouthing.

# Impulsivity, Hyperactivity and Concentration

Impulsivity, poor concentration, distractibility and restlessness are often described in people with SMS.

This cluster of behaviours is commonly thought about in relation to Attention Deficit Hyperactivity Disorder (ADHD) where they form part of the diagnostic criteria.

There are reports of ADHD occurring quite often in individuals with SMS, supporting the suggestion that this cluster of behaviours may be a common characteristic of people with SMS. However, relatively little is known currently about just how common this association is. Impulsivity, poor concentration, inattentiveness, impulsiveness, distractibility, fidgetiness and restlessness may overlap, with some people having difficulties relating more to some of these issues than others. When these challenges are prominent, they can have a severe impact on functioning at home and school or adult placements.

## Impulsivity

Impulsivity is described by caregivers as being particularly problematic and also very common in SMS. Impulsive behaviour can be hard to manage both at home and within an educational setting. This type of behaviour can include difficulty waiting one's turn, interrupting conversations or games, being easily distracted and needing demands to be met immediately. Impulsive individuals tend to "act first and think later" – if at all. The levels of impulsivity seem to be greater than would be expected for the individual's level of intellectual disability, and are greater than in most other people with an intellectual disability. High rates of impulsivity are found in both children and adults with SMS and are likely to be related to other behavioural issues, in particular aggression. Thus, impulsivity is a specific issue in SMS that should be considered in education plans for school-aged children.

## Hyperactivity

Descriptions of individuals with SMS of-



ten include hyperactivity, and there are reports of raised rates of ADHD diagnoses in affected individuals. However, there has been limited formal research on hyperactivity in SMS and so the extent to which it is a particularly common or highly problematic behaviour is not clear. It has been suggested that hyperactivity may decrease with age in SMS to a greater extent than impulsivity.

### Concentration

It can be difficult to assess attention and concentration span in people with an intellectual disability, but descriptions of people with SMS by caregivers and teachers suggest that they may find it hard to attend to information for any length of time. This may affect their ability to acquire new skills, for example in a school setting, as individuals struggle to focus on learning activities.

#### What might help?

Inattentiveness, impulsivity, hyperactivity and poor concentration often co-occur and it may be hard to isolate which of these are affecting a person's behaviour and learning. The following strategies may be helpful in addressing this cluster of behaviours, providing structure to reduce the attentional

demand of tasks and increase motivation.

*Get the setting right* - People with SMS tend to function better in smaller, calmer, less distracting and more focused settings. Sometimes one-to-one assistance can help. Short periods of work interspersed with frequent breaks may be essential.

*Structure tasks clearly* - Give clear and simple instructions with short and simple steps. Cues (pictures or words) can indicate what needs to be completed at each stage. Regular prompting and reminders will encourage staying on task and remind the person what they have to do.

*Use visual cues* - Because of likely strengths in perceptual abilities and weaknesses in short term memory, use of pictures or visual cues/reminders to illustrate daily activities, schedules and other activities are likely to be more effective than verbal prompts.

*Utilise favourite topics/subjects and preferred activities* - If the person has a strong interest in particular topics, then books or games associated with these themes can be used to encourage concentration.

*Use specific rewards and timers* - Items

such as stickers on a chart or an activity the person enjoys (such as the computer) can be used as a reward that can be enjoyed after a set period of time has been spent engaged in more structured activities or work. A timer can be used to help judge how much time there is for each activity and this can be increased as waiting improves.

*Encourage self-prompting* - For older, more able children and adults, encouraging the individual to say instructions out loud can also aid concentration. This can help them to remember to keep working on the task, initially by thinking out loud and then hopefully progressing to saying the instructions to themselves.

*Seek professional input* - If inattentiveness, impulsivity, hyperactivity and poor concentration are of significant concern and affecting function across a range of settings, then it may be appropriate to seek input from a Child Development Centre, a paediatrician or an Adult Learning Disability service. As with other behaviour disorders described previously (see the 'Behaviours that Challenge' section), when features of ADHD are severe, and do not respond well enough to other interventions available, then the use of

medication, prescribed and monitored carefully by a suitably qualified and experienced clinician, can assist when given in addition to the above.

# Repetitive Behaviour and Restricted Interests

Nearly every person with SMS will show one or more different kinds of repetitive or stereotyped behaviours.

**Common repetitive behaviours:** The most common behaviours seen are teeth grinding and inserting hands or objects into the mouth (mouthing), which are also common in other people with an intellectual disability. Other frequently described behaviours include rocking, spinning, hand flapping and fiddling. In some people with SMS these behaviours are self-stimulatory, providing sensory stimulation for the individual (see the 'Sensory Issues' section).

**'Unique' behaviours:** Other behaviours that have been described in people with SMS are found less often in other individuals with intellectual disabilities. These include an unusual 'self-hugging' behaviour which often occurs when the person seems happy or excited. It can appear as the person either crossing their arms tightly across their chests and tensing their upper bodies or clasping their hands together in front and squeezing their

arms to their sides. A second unusual behaviour is repetitive page-turning ('lick and flip' behaviour), where people lick their hand and then use it to rapidly flip over pages.

**Preference for routine:** People with SMS may have difficulties making transitions from one activity to another and strongly desire consistency in their daily routine. Unexpected change can make some people with SMS anxious and may result in a temper outburst.

**Repetitive questioning and restricted conversation:** In individuals with sufficient expressive language, repetitive questioning or conversation is also de-



scribed. This may involve talking excessively about favourite themes or topics, which may be inappropriate or not well integrated into the conversation. Where preference for routine is an issue, repetitive questions may concern future plans and routines. This behaviour could also be related to short-term memory difficulties. One type of repetitive questioning which is particularly common in SMS is repetitively asking to see, speak to or contact particular favourite people (see the 'Social Relationships and Autism Spectrum Disorder' section).

### What might help?

**Repetitive behaviours** - If a repetitive behaviour seems to be related to a need for more sensory input (see the 'Sensory Issues' section), then having appropriate alternative activities or sensory items available that provide a similar sensory input to the behaviour, and which can be accessed independently by the person, may increase available stimulation and reduce the need for him/her to engage in self-stimulatory behaviour. For example providing an appropriate object to chew on may reduce mouthing if this occurs because it is providing oral sensory input.

**Preference for routine** - Where a child has not yet established routines, it may be useful to 'mix up' routines to teach flexibility. If routines are already established then, where possible, try to keep schedules as predictable as possible. If a routine has to change or if a person is about to experience something new, it is important to explain this well ahead of time, and on a number of occasions, if possible. It might be helpful to use 'Social Stories' as an aid. These are made up stories about social situations, usually with pictures which help to prepare the individual for an event or activity that lies ahead. Visual timetables, where the order of activities can be changed (e.g. where pictures are attached with Velcro), may also be useful.

**Repetitive questioning/conversation** - Try to limit responses to repetitive questions. It might be advisable to respond verbally only once when answering the question, and then to provide a visual cue to refer to. Be sure to make it clear that this is what you intend to do every time you are faced with repetitive questions.

# Sleep

Sleep problems are highly characteristic of people with SMS, more so than of other people with an intellectual disability (where sleep problems are also common).



There is now quite a lot of research into sleep difficulties in SMS and it is clear that sleep is a significant problem for nearly all people with the syndrome (75-100% depending on what measures of sleep are used), and that it is likely to have a biological cause in most people. In one group of children and adults where sleep patterns were examined, the average time of waking was 5.30 in the morning, with an average of two waking episodes during the night which lasted on average for around half an hour. The average total amount of sleep was just over 7.5 hours (with younger children sleeping for longer than older individuals)<sup>5</sup>. Despite our increasing knowledge, cur-

<sup>5</sup>Smith, Dykens & Greenberg (1998)

rently there remain many aspects of sleep in SMS which are not well understood.

## Patterns of sleep over the lifespan

### Babies

Infants with SMS are described by their caregivers as good sleepers, often having to be woken for feeds, yet even at this young age there is reduced overall sleep.

### Children

Significant sleep problems emerge in childhood which can impact on the child with SMS and those around them. Problems falling asleep are reported in some, but this is less problematic than frequent night-time waking and early waking, resulting in shortened sleep cycles. Out of sync waking can be particularly problematic in SMS as night-time behaviour





can be very disturbing to other family members and can sometimes be dangerous for the child. Bed-wetting and snoring are also reported to be problematic. Daytime sleepiness is noted.

### Older children, adolescents and adults

Activity appears to be elevated early in the night, suggesting difficulty initiating sleep at this age. As people get older, there appears to be reduced night-time sleep and increased but shorter daytime naps. Despite this apparent reduction in sleep quality with age, caregivers report decreases in sleep problems with age. This is likely to be due to reduced disruptive night-time behaviours with age rather than improved sleep, since older individuals are more likely to be able to keep themselves occupied when they wake up in the night or early morning without disturbing others.

### Daytime sleepiness

In addition to problems sleeping at night, daytime sleepiness is common - naps in individuals with SMS have been described as 'sleep attacks', alluding to the sudden and irresistible nature of the urge for sleep. Daytime sleepiness may account for some disruptive behaviours shown by those with SMS, with fatigue leading to increased likelihood of disruptive behaviour.

### Impact on the family

Unsurprisingly, sleep-related difficulties have been found to be related to increased stress in the family.

### Possible causes of sleep disturbance

Sleep disorder in SMS has been primarily attributed to an inverted release pattern of a hormone called melatonin, which disrupts circadian rhythm (the rhythm that tells our bodies when it is day and night in terms of sleeping patterns). This leads to elevated daytime levels of melatonin release and reduced night-time melatonin release, the opposite pattern to most people, resulting in daytime sleepiness and night-time waking. However, not all people with SMS who have sleep disturbance have this inverted melatonin release, suggesting that it is not the only cause of the sleep difficulties;

therefore, it is also likely that environment and routine have a significant impact on sleep for people with SMS.

### Environmental influences on sleep (night-time routines)

While there may be a biological factor affecting poor sleep in most people with SMS, is it still very important to ensure good sleep hygiene. This includes regular times for going to bed, routines (e.g. bath and a story) before bed and reducing stimulation (activity, TV, computer use) before bedtime.

There is extensive, high quality, information available for caregivers about sleep hygiene and this cannot be covered in detail here (see the 'Sources of Help and Useful Contacts' section for information specifically about sleep routines for children with disabilities). It might be hard to repeatedly return a person with SMS to their bedroom if they seek out their caregiver, as is often suggested by guidelines, as this can go on for a very long time when the person is simply not as sleepy as other people would be. However, given suggestions that people with SMS find adult attention very rewarding, it is likely to be particularly important not to 'reward' waking with attention. This may be through returning the person to his/her room with no eye contact or con-

versation, but if this doesn't work after sustained effort then it could be beneficial to distract a non-sleepy person with suitable activities (see 'What might help?').

It is likely that caregivers will need to include some flexibility when following sleep hygiene guidelines to accommodate specific features of SMS; for example, allowing daytime naps or using distractions at night to occupy those who are unable to go back to sleep.

### Medication

If inverted melatonin release is the primary cause of sleep disorder in SMS, then treatment providing evening melatonin or suppressing its daytime release (e.g. using beta-blockers) might be expected to be effective.

However, the evidence for the effectiveness of melatonin is mixed. Use of melatonin has certainly been reported as being effective in some people, including in combination with a second drug which blocks the effect of daytime melatonin release (a beta-blocker). Using this combination, there are reports of improvement in sleep behaviours (increased total sleep time, reduced early morning waking) and reduced hyperactivity and improved cognitive performance. However, melatonin is not universally effective for

improving sleep. For example, it can enable people to go to sleep earlier (yet settling is not reported to be the primary sleep problem in SMS) but then they may wake earlier, thus their total sleep is the same. In some people, melatonin is described as having no effect at all.

There are different types of melatonin which can be used (including time release melatonin), and other medications have also been used with varying effectiveness. It is important that the prescription and use of medication to treat sleep difficulties is monitored closely by a clinician experienced in this area and with experience of working with people with an intellectual disability.

#### What might help?

*Establish good sleep hygiene* - As described previously it is important to create an environment which promotes sleep (for example black-out blinds to create total darkness in the person's bedroom) and to establish good routines around bedtime including reducing stimulation.

*Help the person know when it is ok to get up* - Clocks which can be set to show when it is ok to get up in the morning with a picture of the sun (and moon and stars at night-time when they

should stay in bed), such as 'Gro clocks', may help to explain this aspect of time. This may not increase the amount of time a person sleeps, but can reduce the disruption caused by night-time and early waking.

*Allow daytime naps but consider their timing* - Preventing a tired person with SMS from napping in the day is unlikely to improve their night-time sleep and is likely to make them more tired and moody. Allowing naps will improve daytime behaviour. Naps during the middle of the day (12-3) are better, as later naps might reduce ability to sleep at night.

*Investigate medical issues* - There are health issues in SMS which may affect sleep quality. Gastro-oesophageal reflux (where stomach acid escapes into the oesophagus causing discomfort) can affect sleep, as lying down can make it easier for the acid to escape (see the 'Health' section). If signs of reflux are being shown, a GP or paediatrician may be able to carry out further investigations. In older people with SMS, sleep disordered breathing (e.g. loud snoring, sleep apnoea) may be an issue due to risk factors, including weight gain, hypotonia and ear, nose and throat abnormalities, and this can



affect sleep. If a person is snoring or gasping/snorting when asleep, it may be appropriate to seek medical advice.

*Seek professional input* - If sleep problems become an issue, caregivers may want to approach their health visitor, GP or paediatrician for advice, or ask to be referred to a sleep clinic or the local psychology service. These services may offer behavioural interventions for sleep difficulties which often focus on providing caregivers with techniques for changing the behaviour that individuals show when they are unable to sleep (as opposed to changing the actual amount of sleep they get). Commonly the aim of such interventions is to increase self-settling abilities to reduce the frequency that individuals call on caregivers when they are awake. Behavioural interventions will often include explanations of the behavioural approach to understanding behaviour problems, including sleep and an explanation of good sleep hygiene principles. They will also provide guidance on strategies for managing settling and waking problems and the effective use of rewards. Caregivers may be asked to record sleep in a sleep diary to help to determine exactly what the sleep problem is

and to monitor how well any treatment is working.

Where sleep problems cannot be addressed using behavioural interventions, careful use of medication, may help some people with SMS to sleep better. However, not everyone will be helped by medication. Furthermore, the effects of medication vary between different people and can change over time in the same person.

Social Services may be able to provide support for families where sleep problems are having a severe impact on family life (e.g. providing respite care).

*Ensuring the person's safety* - Minimising the disruption caused by night-time and early morning waking and ensuring the safety of the person with SMS may help to reduce the problems caused by sleep disturbance. Some ideas that other caregivers have found to be helpful include

- \* Create a room where the person cannot cause harm to themselves (e.g. removing large/heavy objects or using padding on hard surfaces).
- \* Use safety gates or secure room systems to prevent the person from injuring them-

selves while walking through the house.

- \* Provide safe activities to engage the person during periods of wakefulness. While sleep is best promoted in a dark environment, it may be a pragmatic approach for people who simply cannot go back to sleep after following all sleep hygiene guidance, to allow them to read books, look at magazines, do puzzles, drawings etc. Note: We advise that screens are removed at an agreed time each evening because of the understandable difficulty children and adults have of 'switching off' from these.
- \* Attach a small alarm bell on doors to alert caregivers that their door has been opened.
- \* Peep holes or 'stable' door designs can allow caregivers to check on the person without having to enter the room.
- \* Use of an enclosed bed can provide security and comfort (pictured right). These can be costly and funding can be hard to obtain, however.



# Feeding and Eating

There are several areas where difficulties with feeding may occur in people with SMS, including poor feeding in infancy, gastro-oesophageal reflux, textural aversion and weight gain.

**Poor feeding in infancy:** This is common in SMS and can lead to failure to thrive. It is often caused by oral motor dysfunction, with problems sucking and swallowing.

**Gastro-oesophageal reflux:** This is where stomach acid leaks up into the oesophagus which can cause discomfort after eating, pain, and difficulty with swallowing. It can also interrupt sleep, and given that sleep problems are associated with SMS, it is particularly important to identify and treat reflux (see the 'Health' section).

**Textural aversion:** Dislike of specific food textures is described, and may be an issue for people with oral motor difficulties or sensory sensitivities (see the 'Sensory Issues' section).

**Weight gain:** Weight gain may become an issue in older individuals. It is not clear whether this is the result of over-eating (although there are reports of increased

interest in food in some people, which emerges with age) or the effects of having SMS. Weight gain may also be the result of medications used; it has been suggested that medications including valproic acid, risperidone, and recent mood-stabilizing agents may not be the first choice of medication for individuals with SMS because of problems with weight gain.

Problems with eating/appetite and weight appear to be more commonly reported in females with SMS than in males and are more common in people whose SMS is caused by an RAI1 mutation than in those whose SMS is caused by the more frequent deletion.

## What might help?

*Poor feeding in infancy* - Speech and language therapy and occupational therapy evaluations should be pursued early to assess feeding difficulties, optimise oral motor abilities and develop intervention strategies.

*Textural aversion* - This may be related to oral motor difficulties and requires intervention from speech and language therapy. Where it is primarily a sensory issue, caregivers could try to give smooth foods before gradually introducing thicker foods, then introduce a few small pieces of food, and then move on to easily chewed foods.

*Gastro-oesophageal reflux* - Be vigilant for signs of reflux; check NHS guidance (see the 'Sources of Help and Useful Contacts' section) for signs, which include frequent projectile vomiting, wheezing, bad breath and pulling legs up to the stomach after feeding. Seek input from medical professionals if concerned.

*Weight gain* - Encouraging a balanced diet and exercise from an early age is strongly advised.



# Toileting

Delay in achieving continence in the day and persistent night-time wetting are particular issues in SMS (reported in up to 80% of children).

Delayed toilet training is common in children with an intellectual disability; however with consistent implementation of a toileting programme, most children can become toilet trained and this is also the case for most children with SMS.

Many of the approaches that can be used are the same as those which are used with other children; including rewarding children for using a potty/toilet, using a 'fun' potty decorated in a favourite colour or character, having regular trips to the potty which become more spaced out as children become dry, and not telling children off if there is an accident.

There is also a lot of guidance available specifically for caregivers of children with an intellectual disability, which may be useful for caregivers of children with SMS. ERIC is a UK charity which provides advice and support on continence problems, and the charity Cerebra also has a

guide on toilet training (see 'Sources of Help and Useful Contacts' section).

There may be specific health and behavioural issues associated with SMS that might affect toileting. These include difficulties communicating the need to go to the toilet, being easily distracted, delayed adaptive behaviours and constipation/urinary tract infections that children may not show clear signs of or be able to report themselves. Caregivers should be mindful of these difficulties when toilet-training a child with SMS.

## What might help?

*Check physical health problems* - Urinary tract infections can cause episodes of wetting in a person who has been toilet trained. Constipation may be a complicating factor in soiling. It may be necessary for a GP or paediatrician to examine the person to check whether they are constipated; this would need to be resolved before bowel training can commence. It is important to ensure that the person eats a balanced diet

including plenty of fibre, fruit, vegetables and liquids.

*Seek input from professionals* - If problems persist, seek input from professionals e.g. health visitor, GP, psychologist or paediatrician, who may be able to refer on to a specialist for advice.

*Use a bedwetting alarm* - Alarms can be borrowed from Enuresis clinics via referral from GPs or Child Development Centres. If the person begins to wet the pad the alarm will go off, which will then cause them to wake up and stop urinating. The caregiver then takes the child to the toilet and resets the alarm. Rewards may be used in conjunction with the alarm. Alarms are used in conjunction with a toileting programme that is being implemented by a psychologist, health visitor or nurse in partnership with a caregiver and the individual with SMS.

# Dressing

Dressing and undressing requires muscle co-ordination, fine motor skills and planning. Individuals with SMS may take longer to learn these skills than their peers.

A person with SMS might need help with putting on clothes and doing up buttons and shoelaces etc. As there is usually little time in the morning, caregivers may find it more convenient to dress a child with SMS themselves, to save time. However, children need to be encouraged to do these things for themselves, with caregivers slowly encouraging them to do more of the dressing independently. Some adults with SMS may still need help with dressing; in a survey of adults with SMS undertaken some years ago, around half were described as needing some help with this activity.

Some caregivers also report issues with inappropriate removal of clothing in public places. This might occur because the person finds clothing uncomfortable. This could be due to sensory sensitivities, for example a label, seam or other feature of

clothing might be irritating. Alternatively clothing may be too hot. It is also possible that this behaviour may be shown because it has previously been associated with a rewarding response. For example the person might have been taken away from a non-preferred activity when they previously removed their clothes (in order to re-dress them), or they may have been given attention (for example being told to put their clothes back on and perhaps helped to do this).

## What might help?

### Teaching dressing skills

*Practice skills at convenient times* - Learning to dress can be practised at other times of the day, not just during the hectic morning period.

*Practice skills in easier situations* - Teaching a child to do up buttons can sometimes be helped by encouraging him/her to practice on buttons on items of clothing that are not being worn at the time. In the same way, tying shoelaces

could first be practiced on loose shoes.

*Break down tasks into smaller steps and teach one step at a time* - For example, to teach putting on a pair of socks, you might first put on each sock up to the ankle and teach the child to p

ull the sock up from the ankle. Once this step has been mastered, move on to the next stage - put the sock on his/her foot half way over the heel and teach him/her to pull the sock over the rest of the heel, then onto the instep, then just over the toes - and ask the child to pull the sock up over more of the foot at each step. Eventually you should be able to hand him/her the socks, and then leave them on the bed to be put on independently.

*Use visual prompts for each step* - Pictures displaying each step of a getting dressed process e.g. tying shoelaces may also be effective.

*Provide verbal cues* - For example when teaching someone how to do up shoelaces, a person can be helped to talk through the routine, for example by saying out loud 'cross the laces over, pull one through, pull tight' as they perform each step. In this way they learn the words and can then prompt themselves during the task.

*Buy items with easy to use fastenings* - Footwear and clothing with Velcro are a good substitute for shoelaces, buttons and zips. Such items will be much easier for the individual to put on and take off with adult help.

*Managing removal of clothing*

*Ensure clothes are comfortable* - Check that the person is not too warm and dress them in light, cool clothing if necessary. Check for irritation from labels or dislike of certain types of clothes (e.g. tights). Soft jersey fabrics and loose clothing may be more comfortable. There are also companies which specialise in clothing for individuals with sensory issues.

*Respond to clothing removal in a 'low key' manner* - Limit the interaction provided (no eye contact, minimal conversation) and try to help the person to dress and return to their previous activity as quickly as possible to reduce any potentially rewarding attention.



# Health

There are a range of health problems which are more common in SMS than in other syndromes. While estimated rates vary widely, a review by experts in the United States<sup>6</sup> has grouped health problems according to how common they are, and report the following:

How many affected	Health Problems
Over 75% of people with SMS	<ul style="list-style-type: none"> <li>• Dental anomalies (Dental decay found in older adolescents might be attributed to poor dental hygiene)</li> <li>• Middle ear and throat anomalies</li> <li>• Oral sensorimotor dysfunction (early childhood)</li> </ul>
50-75% of people	<ul style="list-style-type: none"> <li>• Hearing loss (related to frequent ear infections)</li> <li>• Scoliosis (curvature of the spine)</li> <li>• Mild ventriculomegaly of brain (enlarged ventricles, the fluid filled structures, in the brain)</li> <li>• Hyperacusis (sensitivity to noise)</li> <li>• Tracheobronchial problems (problems affecting airway and lungs)</li> <li>• Velopharyngeal insufficiency (improper closing of the soft palate muscle in the mouth – can cause problems with speech, eating and sleeping)</li> <li>• Eye abnormalities (iris anomalies; microcornea)</li> </ul>

How many affected	Health Problems
50-75% of people	<ul style="list-style-type: none"> <li>• REM sleep abnormalities</li> <li>• Hypercholesterolemia/hypertriglyceridemia (increased cholesterol or levels of a specific type of fat in the blood)</li> <li>• Constipation</li> <li>• Abnormal EEG without obvious seizures</li> </ul>
20-25% of people	<ul style="list-style-type: none"> <li>• Heart defects</li> <li>• Thyroid function abnormalities</li> <li>• Seizures (estimates vary)</li> </ul>
Fewer than 25% of people	<ul style="list-style-type: none"> <li>• Kidney/urinary tract abnormalities</li> <li>• Seizures</li> <li>• Forearm abnormalities</li> <li>• Cleft lip/palate</li> <li>• Retinal detachment</li> </ul>

It is important for caregivers and professionals to be vigilant for health issues which are more common in SMS, as well as for everyday illnesses. Given the expressive language difficulties of some people with SMS, it may be hard to determine if someone is in pain or discomfort or is feeling unwell. Additional detailed information about pain in individuals with an intellectual disability can be found in a

guide for parents produced for the charity Cerebra (see the 'Sources of Help and Useful Contacts' section).

Specific common issues to be vigilant for include ear infections, constipation, and gastro-oesophageal reflux. It is unclear how common reflux may be. If a person with SMS is showing signs of reflux, then it is worth pursuing this with a medical professional as it can also affect sleep and

<sup>6</sup>Smith, Boyd, Elsea et al. (2001)

eating. As ear infections are so common and can impact on both well-being and development, a specific subsection on this is included at the end of this section.

Decreased sensitivity to pain and peripheral sensory neuropathy have also been reported in people with SMS. This is commonly described as people not feeling the effects of a painful event as clearly as others might - e.g. they can fall over and bang their head and then not cry or have an injury such as a broken bone and not complain of pain. When considering this, however, it does not mean that people with SMS do not feel pain - painful events and health conditions are still likely to result in distress and discomfort.

Observational tools, such as the Non-Communicating Children's Pain Checklist -Revised or the FLACC (which stands for Face, Legs, Activity, Cry, Consolability), may help to identify if someone with limited communication is in pain (see the 'Sources of Help and Useful Contacts' section). It is also important to note that there is an association between pain and discomfort, and behaviours such as self-injury and aggression. These behaviours are more likely to be shown when a person is in pain and a sudden increase in these behaviours may indicate pain or discomfort in some (but not all) people. Change in activity levels, mood, sleeping

and eating may also be related to experiencing pain. A change in behaviour and habits should alert caregivers to the possibility of health problems.

Below is a table showing the health-related investigations and assessments recommended in Guidelines for the management of children and adults with Smith-Magenis syndrome by the UK SMS Scientific and Clinical Advisory Group<sup>7</sup> that should be carried out when a child/adult is first diagnosed with SMS:

<sup>7</sup>Keen, Gringras, Male & Udwin (2009)

Clinical area	Investigation/assessment
Growth, feeding & nutrition	Height and weight centiles. Feeding evaluation – examination of palate, assessment of swallowing, oral motor skills, gastro-oesophageal reflux, caloric intake. Referral to specialist feeding team, if indicated.
Cardiac	Cardiac examination including echocardiogram.
Renal	Renal ultrasound.
Immune system	Assessment of immune function if presenting with frequent infections.
Hearing	Assessment for conductive and or sensorineural hearing loss. Grommets or hearing aid may be indicated.
Ear, nose and throat	Otolaryngologic evaluation to assess ear, nose and throat problems, with specific attention to ear physiology and palatal abnormalities.
Eyes	Ophthalmologic examination with attention to evidence of strabismus, microcornea, refractive error, retinal detachment.
Spine	Assessment for scoliosis.
Genetics	Individuals with larger deletions screen for adrenal function assessment of nerve conduction velocity if the gene PMP22 is involved.

Below are assessments recommended to be carried out annually:

- Monitor for scoliosis
- Thyroid Function
- Consider ophthalmologic evaluation
- Audiologic evaluation at regular intervals or as clinically indicated to monitor for conductive or sensorineural hearing loss
- Periodic assessment of presence of challenging behaviours and referral for treatment as needed

### What might help?

*Assess and monitor health* - Pursue the health checks identified above.

*Monitor behaviour for signs of pain* - Monitor any changes in general behaviour and if pain is suspected use observational tools to provide information about pain related behaviours in people with limited communication (see above and the 'Sources of Help and Useful Contacts' section).

### Ear Infections

Middle ear infections (also called 'otitis media') are a particularly common health issue in SMS and can affect hearing, as well as cause pain and discomfort. In childhood, three or more episodes a year are common.

They are caused by bacterial or viral infections causing mucus to accumulate in the middle ear, which can then become infected. This affects children more than adults because children's Eustachian tubes (which help air into, and drains fluid from, the middle ear) are smaller than adults'. Flattening of the midface contributes to this being a particular problem in SMS. If the middle ear becomes filled with

fluid for a long period, causing problems with hearing, this is called 'Glue Ear'. This can be caused by several things, including previous ear infections.

Presence of an ear infection might be indicated by a change in behaviour (irritability, increased temper outbursts, self-injury or aggression), change in eating or sleeping habits, a high temperature or the individual repeatedly touching the affected ear (rubbing, pulling).

The high levels of hearing loss in SMS (estimated to affect around 60% of individuals overall) is often associated with chronic ear infections.

### What might help?

*Vigilance for infection* - Awareness of behaviours that might indicate an ear infection, noted above, may help to identify when a person is experiencing an ear infection.

*Regular monitoring* - It is recommended that regular ear, nose and throat examinations are conducted (see table in main 'Health' section)

*Seek medical treatment* - If there are concerns about an ear infection (e.g. pain, length of infection) then seek treatment from a GP, and also seek their advice regarding any concerns

around a person's hearing.

*Surgical intervention* - Where indicated by a medical professional, for example, because of chronic issues (lasting more than 3 months), or where it affects hearing, speech or language development, a surgical procedure to insert small tubes which drain away the fluid, called pressure equalisation (PE) tubes, or grommets, may be appropriate.

*Address hearing loss* - Where hearing is impaired and if indicated by a medical professional, use of a hearing aid may help to reduce the impact of this on speech and language development.

# Growing Up With SMS



At the time of writing this guide, we know relatively little about what happens to people with SMS as they become adults.

This partly reflects

the fact that SMS was only identified relatively recently, and so those who were diagnosed as infants early in the syndrome's history have only recently become adults, thus there are not large numbers of people to tell us how things change.

Here, we summarise what is known, revisiting some of the topics considered previously with specific focus on adulthood. For more general information about transitioning to adulthood for families of people with an intellectual disability, see the 'Sources of Help and Useful Contacts' section.

## Ability

There is no evidence to suggest that gen-

eral cognitive ability declines in adults with SMS. Furthermore, the profile of strengths and weaknesses demonstrated in the syndrome seems to be fairly consistent across the lifespan, with adults showing similar strengths in verbal comprehension compared to working memory to those shown in children. Thus, the strategies that play to the strengths of younger individuals, such as visual prompts and timetables, may also be effective with older individuals, if tailored to be age-appropriate.



## Mental capacity

When people with SMS are children, caregivers can make decisions on their behalf, however, when they become adults this may change. As individuals with SMS become older, issues might arise around their ability to make decisions, from whether to go to the shops to whether to

have an invasive medical procedure; this is usually referred to as 'mental capacity'.

There is legislation which governs how this ability is assessed and what steps should be taken if it is felt that an individual does not have capacity to make such decisions. This can have important implications for the lives of individuals with SMS who have an intellectual disability. Where an individual is deemed to have capacity they will be able to make these decisions themselves. If a person is deemed, after assessment, not to have capacity, then a caregiver or professional (e.g. a support worker) may make decisions on their behalf, which must be in their best interests. In addition to medical issues and day-to-day life, capacity may affect issues such as planning for the future, e.g. financial decisions and decisions about living arrangements.

The implications of mental capacity will differ depending on where families are located, with different countries having different approaches to this issue.

#### What might help?

*Engage with legislation* - Caregivers or support workers should familiarise themselves with the relevant legislation regarding capacity in their location.

*Assess capacity* - Caregivers or support workers should engage in an assessment of capacity where this is needed.

*Plan for the future* - Caregivers or support workers should put in place any legal arrangements needed to plan for the future (e.g. wills and trusts).

## Health and Physical Appearance

While we lack the research to know with certainty, it has been suggested that life expectancy for people with SMS is likely to be in line with the wider population of people with an intellectual disability (provided the person does not have major organ involvement as part of the syndrome). The oldest known person with SMS lived to be 88 years of age.

Facial appearance may change with age, becoming 'heavier'. By the teenage years, weight may become an issue; with reports of a large percentage of people over the age of 14 with SMS being overweight or obese.

#### What might help?

*Monitor health* - Continue to follow guidelines for health checks (see the 'Health' section).



*Monitor weight* - Keep an eye on weight and also changes in medication that may be related to weight gain. Encourage healthy eating and engagement in

## Behaviour

There are variable accounts of how self-injury, aggression and temper outbursts may change over time; in some cases these become more problematic, in other cases less problematic, and in still other cases they may remain the same as individuals get older. This is quite a mixed picture, reflecting the variability of people with SMS. There are suggestions that, as individuals with SMS get older, the types of self-injury they show may increase, i.e. they may show a greater variety of behaviours, but not necessarily higher rates of self-injurious behaviour overall. Further help can be obtained from the local Adult Learning Disabilities Services.

Impulsivity appears to remain elevated in adulthood, whereas hyperactivity may reduce more noticeably. Where self-injury, aggression and temper outbursts are prominent in adults they may become more problematic by virtue of increased size and strength, making management more difficult for caregivers. In addition

to input from psychological services, caregivers may need to consider use of medication under the direction of a psychiatrist (if not already considered) – see the ‘Behaviours that Challenge’ section.

Accounts of the effectiveness of medications for behavioural issues are variable; a number of different doses or types may be trialled before an acceptable outcome is reached. In some cases, where behaviour becomes very hard to manage at home, a placement outside of the home which specialises in managing behaviours that challenge may be needed.

## Sleep

Sleep issues persist in adulthood but tactics for managing the sleep disruption are likely to be more effective, with people being able to occupy themselves with books, television or computers/tablets if they wake early. While the person is not actually getting any more sleep, this does greatly reduce the disruption caused to those sleeping nearby.

Objective measurements of waking suggest that adolescents have greater problems with settling to sleep than younger children.

## Puberty

Puberty in SMS usually follows the same path as most teenagers, although individual cases of early puberty and of delayed puberty have been described. Just like teenagers without SMS, hormonal changes during puberty affect those with SMS, for example, they may have increased mood swings.

There is no research yet on fertility of people with SMS but there are accounts of a very small number of adults with SMS having children. This suggests that some people with SMS, at least, are able to have children. If they do have children, there is a 50% chance of the child also having SMS.

Having an intellectual disability does not take away a person's right to getting information about sex and relationships and having the opportunity to seek out romantic or sexual relationships. Whether young people with SMS are interested in being in a sexual relationship or not, it is likely that they will want to explore and express their sexual feelings. These feelings may be confusing for them and it may feel difficult for caregivers to discuss sexuality and relationships. They may feel embarrassed or not know how best to approach the issue. However, it is important to give young people opportunities for open and honest discussion about

sexual matters, to advise on what is and isn't appropriate behaviour and allow them to express their sexuality in a private and safe place. Discussions about this might be with a caregiver or another trusted adult.

Caregivers will of course be worried about the risk of unplanned pregnancy, sexually transmitted infections and especially the young person's vulnerability to abuse. They will have to find a balance between safeguarding and allowing the young person choices and opportunities to become as independent as possible. Remember that ignorance does not protect them from abuse, it may make them more vulnerable because they won't know what is and what isn't abusive.

### What might help?

*Discuss puberty and sexuality* - Introduce these topics with the person at an appropriate age and at an appropriate level. They may also receive sex education at school or college, caregivers can find out from their teacher what they have been taught and how best to support them at home.

*Access advice and support* - Caregivers may want to get advice and support from the nurses or psychologists of

their local Adult Learning Disabilities Services who should have useful resources and can discuss the best ways in which to meet the young person's needs. Information and advice is also available from sexual health charities like the Family Planning Association and Brook (for people aged under 25).

*Find accessible resources* - There are a number of resources designed specifically for adolescents and adults with an intellectual disability to help discuss puberty and sexual health. For example, the Family Planning Association has books and interactive resources available and the British Institute of Learning Disability (BILD) website signposts to a number of different useful resources (see the 'Sources of Help and Useful Contacts' section).

## Independence

As described previously, independent living skills in individuals with SMS tend to be lower than would be expected given their level of cognitive ability. Adults have been described as being quite dependent on their caregivers and requiring a high level of support. This seems out of line with the level of intellectual disability in this syndrome, which is typically moder-

ate. This dependence may be attributable to characteristics such as impulsivity, attention-seeking, temper outbursts, aggression and self-injury, which are common in individuals with SMS.

The most complete information we currently have about independence in adults comes from a description of a group of 21 adults<sup>8</sup>. This information, published 2001, is summarised below, and provides a snapshot of people with SMS, to illustrate the range of adult outcomes in a relatively small group.

Living arrangements	Number of adults
Living at home with parents	11
Living in group homes or residential communities	8
Living in residential boarding schools	2

Duration adults could be left alone	Number of adults
Not more than one hour	18
A matter of minutes	12

<sup>8</sup>Udwin, Webber & Horn (2001)

Daily Living Tasks completed independently	Number of adults
Independent personal care (E.g. teeth Brushing)	3
Tidying/cleaning on own initiative	2
Cook meal on own	2
Dress independently	6
Travel around local area independently	21
Travel alone for greater distance (E.g. another town)	0

Employment/Further Education	Number of adults
Employed	1
Attending day centre	8
Attending day centre (with supported employment)	6
College course for people with special needs	6
Number achieving a formal qualification e.g. GCSEs	0

It is clear that there is a lot of variability in adults' independent living skills and their situations. We do not know why this is the case, but it can be seen that, while many people with SMS struggle with daily living tasks and depend significantly on their caregivers, a number of individual with SMS are more independent and have gained many key life skills.

### What might help?

Suggestions for increasing independence and daily living skills can be found in the 'Adaptive Abilities' section.



# Family

## Siblings

People with SMS often have very strong bonds with their siblings, which carry on into adult life.

When a group of siblings of people with SMS were asked as part of a research study<sup>9</sup> about how positively or negatively they felt towards their sibling, it was found that positivity increased, and negativity decreased, as the siblings got older.



Interestingly, the siblings who were able to identify benefits in being the sibling of a person with SMS also felt more positively towards their brother/sister.

While having a brother or sister who has

SMS can be fun and rewarding, at times it may also be confusing and stressful. The needs of individuals with SMS can, at times, be overwhelming, and can take up a lot of family time, energy and attention. As such, brothers and sisters may get less attention from parents, grandparents and other relatives, and at times they might understandably feel resentful or angry.



Siblings might feel embarrassed or resentful when, for example, they have to explain to their friends about their brother's/sister's difficult behaviour, or have to keep an eye on them. At other times they might feel guilty about the difficulties their brother or sister is experiencing

Often, they



<sup>9</sup>Moshier, York, Silberg & Elsea (2012)

also feel a great sense of responsibility towards their brother or sister who has SMS, and as they become adults they may worry about the future and who will look after their sibling once their parents are no longer able to do so.

### What might help?

*Give attention* - Sometimes easier said than done, but if at all possible parents should try and ensure that they give their other children as much individual time and attention as they possibly can. If siblings are finding it hard having a brother/sister with SMS, remember that negative feelings towards siblings are likely to decrease with age; it is unlikely to always be this way.

*Talk and share feelings* - Enabling siblings to talk about their feelings towards their brother or sister with SMS, and about their worries and anxieties, is extremely important.

*Provide information* - Many siblings are poorly informed about SMS, and, as such, may have unnecessary worries about the possibility that they may be at risk of developing the condition themselves or of having a child with SMS. It is vital for parents to talk with their unaffected children about

SMS, and to give them information about the cause of the condition, the negligible risk of recurrence in families, and the physical and behavioural characteristics associated with the syndrome.

Older siblings might find it useful to talk with their sibling's paediatrician or Clinical Geneticist if at all possible. Siblings can have as much need for information as parents, and accurate information will help to allay many of their worries and fears.

*Plan for the future* - Realistic planning for the future can help to alleviate many of sibling's justifiable concerns about how their brother/sister will be looked after in years to come, when parents are no longer able to do so.

### Caregivers

Caring for any person with a disability presents challenges which can, at times, become stressful. Dealing with a range of health, social and educational professionals, managing behaviour and concerns about the future, may all contribute to this.

As SMS is a rare syndrome, caregivers may also feel that they are responsible for





explaining the syndrome, and their child's difficulties, to professionals who are not familiar with

the condition. Given the possible range of health and behavioural issues that can affect a person with SMS, it is perhaps unsurprising that compared to caregivers of those with an intellectual disability but without a diagnosis of SMS, caregivers report higher stress levels.

It has been suggested that this increased stress is most likely related to the higher rates of behaviours such as self-injury,



aggression, sleep difficulties and temper outbursts associated with SMS. In addition to this, caregiver well-being has been found to be related to their concerns about their child's health.

It is not inevitable that caregivers will become stressed however, and being stressed, anxious or depressed is not something a caregiver should simply 'put up with'. We would urge caregivers to consider the importance of thinking about their own well-being and looking after themselves so that they can both care effectively for their child and also enjoy family life, work and other everyday activities and opportunities.

### What might help?

#### *Seek support from a health professional*

- If caregivers are concerned that they may be stressed, anxious or depressed they should contact their GP and discuss the possibility of referral on for counselling or psychological support.

#### *Access caregiver support groups*

- These groups can provide informal support, either locally or through social media. There are syndrome specific support groups focusing on SMS and more general groups for caregivers of children and adults with disa-



bilities. The national UK SMS support group is the Smith-Magenis Foundation. There are also several international SMS support groups. See 'Sources of Help and Useful Contacts' for more details about family support groups.

*Access services for the person with SMS -* If caregivers are experiencing high levels of stress due to specific issues, such as health or the behaviour difficulties in their child, then seeking input from an appropriate service may be a step towards addressing this. The local Child Development Centre, Child and Adolescent Mental Health Service (CAMHS), Adult Learning Disabilities Service or Children with Disabilities Team (Social Services) may be able to help, or national organisations such as Mencap, Cerebra or Contact a Family may be able to signpost and support caregivers (see the 'Sources of Help and Useful Contacts' section).

*Use of respite services -* Some of the difficulties in SMS make the syndrome much more difficult to manage than other syndromes or disabilities. Severe sleep problems that affect many of the children and adults can make it very hard for caregivers to get a break from caring, and in-

crease the impact of everyday worries. Respite care for a person with SMS may be a valuable resource and help caregivers in this situation. Access to respite is usually available through Social Services.

# Sources of Help and Useful Contacts

- a) Local Child Development Centres are organised differently depending on the area, but generally they have multi-disciplinary teams consisting of paediatricians, psychologists, community nurses, speech therapists and physiotherapists. Many teams also now have input from paediatric occupational therapists. One or more members of the team would assess and review the child's development at regular intervals. They can be a valuable source of help and advice about the child's development and any behavioural difficulties that may arise, and if necessary can refer the child to other professionals, with caregivers' permission.
- b) Child Learning Disabilities Teams are also available in some areas. Some of these teams provide access primarily to learning disability nurses who can offer advice about managing behavioural difficulties. Other teams involve the types of multi-disciplinary teams describe above.
- c) Adult Learning Disabilities Teams are available in most areas. They tend to be multi-disciplinary teams consisting of psychologists, occupational therapists, speech therapists, psychiatrists and community workers. These professionals can provide valuable advice concerning abilities and behavioural and emotional difficulties in adults with SMS.
- d) Social Service Departments can offer advice on benefit entitlements. They may also be able to organise respite care for children and adults for a few days or weeks (especially during holiday times or when there is a crisis or emergency). Respite care can be helpful not only for giving parents much needed breaks, but also for giving individuals the chance to experience different environments, meet different people and gain independence skills.

- e) Child/Adult Clinical Psychologists (employed within the National Health Service) help with behavioural and emotional difficulties and Educational Psychologists (employed by Education Authorities) are available to help with educational concerns.
- f) Portage schemes and other Early Years programmes are available in many areas, providing trained workers who work with parents in order to teach their young children new skills and to help them develop in such areas as language and motor development, co-ordination, self-help skills and socialisation. Information about such schemes, and also toy libraries, opportunity groups and other facilities in the local area can be obtained from the health visitor, Child Development Centre or Local Education Authority. Alternatively, the National Portage Association ([www.portage.org.uk](http://www.portage.org.uk)) allows caregivers to search for the nearest Portage service.
- g) Information about benefits which families and their child are entitled to claim can be obtained from the local Social Services Department or by contacting Disability Rights UK, Ground Floor, CAN Mezzanine, 49-51 East Road, London, N1 6AH. The Disability Rights UK website lists several Helplines within Disability Rights UK that provide information on a variety of issues: [www.disabilityrightsuk.org](http://www.disabilityrightsuk.org).
- h) The Smith-Magenis Syndrome Foundation (Parent support group in UK). BCM Smith-Magenis Syndrome Foundation, Registered Charity 1072573, London, WC1N 3XX, Email: [info@smith-magenis.co.uk](mailto:info@smith-magenis.co.uk), Tel: 020 7419 5007.
- i) Contact a Family National free-phone helpline: 0808 808 3555, email: [helpline@cafamily.org.uk](mailto:helpline@cafamily.org.uk). Website: [www.cafamily.org.uk](http://www.cafamily.org.uk). A confidential helpline specifically for parents and carers of children with disabilities, rare disorders, special needs.
- j) Carers UK operate an information and advice line for carers - free-phone 080 808 7777, email: [info@carersuk.org](mailto:info@carersuk.org); [www.carersuk.org](http://www.carersuk.org).
- k) Carers Trust work to improve support, services and recognition for anyone living with the chal-

lenges of caring, unpaid, for a family member or friend who is ill, frail, disabled or has mental health or addiction problems. - Tel. 0844 800 4361, Email: [Info@carers.org](mailto:Info@carers.org); [www.carers.org](http://www.carers.org).

- l) IPSEA (Independent Parental Special Education Advice) runs an advice line: 0800 018 4016 which provides free advice on Special Educational Needs. [www.ipsea.org.uk](http://www.ipsea.org.uk)

## Links to further resources

### Syndrome overview

GeneReviews description of Smith-Magenis syndrome: <http://www.ncbi.nlm.nih.gov/books/NBK1310/>

### Sensory Issues

National Autistic Society information about sensory difficulties <http://www.autism.org.uk/sensory>

Cerebra toy library: <http://w3.cerebra.org.uk/help-and-information/library/>

### Behaviours that Challenge

Cerebra briefing on self-injurious behaviour in children with intellectual disability: <http://w3.cerebra.org.uk/research/>

[research-papers/self-injurious-behaviour-in-children-with-intellectual-disability/](http://w3.cerebra.org.uk/research/research-papers/self-injurious-behaviour-in-children-with-intellectual-disability/)

### Sleep

Article published in Paediatrics about Sleep Hygiene for Children With Neurodevelopmental Disabilities <http://pediatrics.aappublications.org/content/122/6/1343.full.pdf>

### Toilet training

ERIC: <http://www.eric.org.uk/>

Cerebra parent guide on toilet training: <http://w3.cerebra.org.uk/research/guides-for-parents/toilet-training/>

### Health

NHS Information about gastro-oesophageal reflux: <http://www.nhs.uk/conditions/Gastroesophageal-reflux-disease/Pages/Introduction.aspx>

FLACC pain scale infographic: <http://w3.cerebra.org.uk/research/research-papers/self-injurious-behaviour-in-children-with-intellectual-disability/>

Non-communicating Children's Pain Checklist: [http://www.aboutkidshealth.ca/En/Documents/AKH\\_Breau\\_everyday.pdf](http://www.aboutkidshealth.ca/En/Documents/AKH_Breau_everyday.pdf)

Cerebra parent guide on pain: <http://w3.cerebra.org.uk/help-and-information/guides-for-parents/pain-in-children-with->

severe-intellectual-disability-a-guide-for-parents/

### Growing Up with SMS

Cerebra parent guide on transitioning to adulthood: <http://w3.cerebra.org.uk/research/guides-for-parents/transition-to-adulthood-a-guide-for-parents-2/>

British Institute of Learning Disabilities (BILD) information about sexuality and relationships: <http://www.bild.org.uk/our-services/books/practical-support-for-better-lives/exploring-sexual-and-social-understanding/>

<http://www.bild.org.uk/information/relationships/general-to-consent/#General>

Brook information about sexual health for under 25s: <http://www.brook.org.uk/>

Family planning association information about sexual health: <http://www.fpa.org.uk/>

### International Smith-Magenis organisations

#### UK

Smith-Magenis Syndrome Foundation UK  
[www.smith-magenis.co.uk](http://www.smith-magenis.co.uk)

#### America

PRISMS  
[www.prisms.org](http://www.prisms.org)

Smith-Magenis Research Foundation  
<http://www.smsresearchfoundation.org/>

Taylor Bug Kisses Foundation  
<http://www.taylorbugkisses.com/>

#### France

Association Smith Magenis - ASM17  
France  
[www.smithmagenis.com](http://www.smithmagenis.com)

Pas à Pas avec Alexia  
[www.pasapasavecalexia.fr](http://www.pasapasavecalexia.fr)

#### Germany

<http://www.smith-magenis.de/>

#### Denmark

[www.smithmagenis.dk](http://www.smithmagenis.dk)