



PRISMS Clinic
and Research
Consortium

CONSTIPATION IN INDIVIDUALS WITH SMITH-MAGENIS SYNDROME

*Recognition and Recommendations
for Treatment*

PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

Gail Reiner,^{1,2} DNP, FNP-C, Myra Woolery,³ PhD, RN, APRN-CNS, CPON, and Ann C.M. Smith,^{1,4,5} MA, DSc (Hon.), CGC

Authors' affiliations:

¹ PRISMS Clinic and Research Consortium (PCRC)

² Nurse Practitioner III, UCSD Neurosciences, Pediatric Neurology, Rady Children's Hospital

³ Pediatric Clinical Nurse Specialist, Neurology, Behavioral Health and Pediatrics Nursing Service, Nursing Department, Clinical Center, National Institutes of Health, Bethesda, MD

⁴ PRISMS Professional Advisory Board (Chair Emeritus)

⁵ Sr. Genetic Counselor & Special Volunteer; Office of the Clinical director, National Human Genome Research Institute, NHGRI/NIH, Bethesda, MD.

The treatment and intervention recommendations for SMS are written by professionals, with input from parents, and with the understanding that they will be published online by PRISMS, read by caregivers, parents, and other family members, and shared by those parents and caregivers with their child's own healthcare professionals. **Do not initiate any medications or, dietary changes without first consulting a licensed healthcare provider.**

The documents are recommendations only and are not formal medical guidelines or scientific papers. They are based on the consensus of clinicians with expertise working in the SMS field. The documents are not referenced in the way articles published in medical journals would be referenced. This was purposefully done to make them more understandable for non-medical individuals.

The PRISMS Clinic & Research Consortium founded in 2020 exists to improve the delivery of clinical services to families impacted by Smith-Magenis Syndrome and to develop a research infrastructure for advancing the development and implementation of new and improved treatments. Please contact **PRISMS** for more information (www.prisms.org).



BACKGROUND

Constipation is a common concern reported by caregivers of individuals with special health care needs, such as Smith-Magenis Syndrome (SMS). Smith and colleagues identified constipation as a frequent medical symptom observed among 53% of study participants.¹ Further exploration under the natural history study of SMS at the National Institutes of Health (NIH protocol 01-HG-0109) suggested that symptoms of constipation are more common than previously identified.² The modified Pediatric Constipation Assessment Scale (PCAS), a tool originally developed and validated for a pediatric oncology population^{3,4,5} was used to assess the frequency of constipation in 43 individuals with SMS (20 from USA and 23 from Australia).² In this study of SMS, 79% were documented to have a history of constipation. Most individuals did not have a normal stool pattern, reporting significant evacuation symptoms such as hard stools, straining, and bloating that are common complaints in constipation. These descriptive symptoms were consistent with the stool form chosen by caregivers using a clinical assessment pictorial scale (see figure 1).^{2,4-8}

The Bristol Stool Form (BSFS)









<p>Slow Transit</p>  <p>Fast Transit</p>	Constipated Stool Consistency	Type 1		Separate hard lumps, like nuts
		Type 2		Sausage-like, but lumpy
	Borderline	Type 3		Like a sausage, but with cracks in the surface
	Normal Stool Consistency	Type 4		Like a sausage or snake, smooth and soft
		Type 5		Soft blobs with clear-cut edges
	Diarrhea Stool Consistency	Type 6		Fluffy pieces with ragged edges, a mushy stool
		Type 7		Watery, no solid pieces

Figure 1. Bristol Stool Form Scale. This pictorial scale illustrates the range of stool formation, from constipation to normal stool to diarrhea. Source: Adapted from Lewis and Heaton, 1997, see [Bristol stool chart](#).

The rate of constipation in SMS (53-79%)^{1,2} is higher than median rates found in neurotypical children (up to 30% worldwide),^{1,9,10} adults (up to 16%), and adults older than 60 years (33%).⁹ The frequency of constipation among individuals with intellectual disability (ID) varies depending on how the studies were conducted, with most in the range of 33% to 50%.¹¹ Even higher rates of constipation have been reported in non-ambulatory individuals with profound ID¹¹ and particularly those living in institutions.¹²

SMS-related Factors for Constipation

Several factors in SMS can contribute to the development of constipation. Not every individual has all these factors, but any one of these can contribute to constipation:

- Hypotonia (reduced muscle tone) can cause problems with coordination of voluntary and involuntary muscle function. Depending on its severity, hypotonia, along with other factors, can result in reflux and/or constipation. Please see additional information at [AACPDM guidelines](#).
- Tethered cord (spinal cord attachment to surrounding tissues), which may occur in SMS, can make both normal stooling and toilet training more difficult.¹³
- Excessive drooling (sialorrhea or hypersalivation) can cause fluid loss and dehydration that can contribute to constipation.
- Medications that have anti-cholinergic side effects (drying effect) may harden stool making it difficult to move through the bowel.
- Soft foods, which individuals with SMS may prefer due to chewing difficulties, are often lower in fiber, slowing the movement of digesting food through the gut toward elimination. Dietary examples of constipating foods include cheese and bananas.
- Inadequate fluid intake can lead to constipation. The individual's inability to express their thirst and/or sensory deficits leading to a lack of awareness of thirst may be contributing factors.
- Expressive language problems may limit the ability to verbally report whether it hurts or is difficult to bowel movement.
- Other medical problems like hypothyroidism, diabetes, and obesity can increase the rate of constipation.
- Disruption in the normal circadian rhythm can disrupt the bowel significantly, leading to issues such as constipation and irritable bowel syndrome.¹⁴ This is of interest in SMS given the well-documented circadian sleep disturbance and warrants further study.

[SMS Medical Management Guidelines](#) (2018) published by PRISMS acknowledge gastroesophageal reflux disorder (GERD) and chronic constipation as two gastrointestinal (GI) problems that are common in SMS. Overflow diarrhea can be a result of severe constipation and encopresis.

The longer stool is in the colon, the drier it gets. Sometimes constipation will trigger inflammation, or the stool may become impacted, resulting in diarrhea or loose stool.¹⁵ The amount of stool (loose or hard) is often small, while drier stool or impacted stool remains higher in the bowel (like a plug in a pipe).

People with routine bowel movements are typically better able to enjoy eating and moving through their day with less discomfort from an overly full bowel. Mood problems such as anxiety or depression can contribute to gastrointestinal (GI) problems, including constipation, and conversely, GI problems may also contribute to mood dysregulation. An important reason for constipation management and prevention is to help with mood and behavior regulation, including anxiety, moodiness, and increased frequency of meltdowns.¹⁶ If bowel movements become associated with discomfort or pain, individuals with such concerns may avoid stooling. For individuals who cannot express themselves through words or sign language what is wrong, an observation of behavioral changes/escalation may serve to communicate distress, discomfort, or dread of having a bowel movement.

The Goal

Caregivers are encouraged to discuss their concerns about constipation with the primary care provider and discuss whether a GI specialist evaluation would be helpful. **Do not initiate any medications or dietary changes without first consulting a licensed healthcare professional.**

Diagnosics and Referral

Referral to a GI specialist will usually depend on whether the problem is ongoing, involves more symptoms such as nausea, bloating, and/or vomiting or is severe and not responsive to typical management strategies. A number of more specialized tests may be ordered by a GI specialist if treatment using usual medications and strategies are not effective or if the severity of the problem is significant ([Mayo Guidelines](#)).

Sometimes when imaging studies such as an MRI or CT scan are done for another reason, the radiologist may notice that the bowel is full and backed up (impacted) with stool.

Sometimes a flat-plate abdominal X-ray can also show this problem and because it is quick, simple, and inexpensive to obtain, this is sometimes the first test that is ordered when stool impaction is suspected.

Common Management Strategies

Strategies reported to be helpful in over 40 individuals with SMS were increased intake of fluid, fruit, and fiber. Combined intake of fluid, fruit, and fiber was associated with softer and more frequent stools.² These findings suggest that changes in fluid intake and diet may be effective in treating constipation in SMS. Described below are suggested simple approaches to treatment of constipation. **Consult with the primary care provider before making any changes to diet or before adding any medications or supplements.**

Lifestyle Approach

Adequate fluid, fiber (bran and psyllium are two good examples that can boost fiber intake easily), prune juice, and Yakima paste (see **Appendix 1** for recipe) are natural nutrition-based stimulants that some families report can be effective if constipation is not chronic or severe. Sorbitol candies such as gummy bears can work as a very mild stimulant, as well as a positive potty-training association.

Exercise and massage of the lower back or abdomen can sometimes be helpful if constipation is mild. A warm, relaxing bath may be helpful in relaxing the person, thus stimulating stool passage that may be inhibited by anxiety.

Having a routine (consistent) time of day to stool, ideally after a meal and drinking water, which is unhurried and relaxed is key. Appropriate body positioning on the toilet e.g., stepstool or “squatty potty” where the legs are bent to the level of the hips may be helpful.¹¹ Giving individuals a simple activity, such as reading or looking at a picture book while they are on the toilet may help them relax.

Probiotics may reduce the need for enemas but have not been shown to be solely effective in treating constipation.¹⁷ Probiotics are available in many yogurts and liquid drinks that can be added to the diet.

Over the Counter and Prescription Medications

If medicine is recommended, the dosing should be determined by the provider caring for your child with SMS and guided based on their judgement of the cause of constipation. Remember, **do not initiate any changes without first consulting a licensed healthcare professional.**

Probiotics, such as Culturelle, may sometimes be prescribed and covered by insurance whether they are over the counter or prescription.

A **natural magnesium citrate supplement**, CALM, is available over the counter. It comes in a powder that can be dosed very easily starting with ¼ tsp/day in water and increased to benefit.

Osmotic laxatives are medicines that draw water into the bowel. Examples include lactulose, polyethylene glycol (Miralax), and sorbitol.

Bulk forming laxatives are medicines that bulk up stool by increasing the amount of water in the stool (not to be used if there is fecal impaction). Examples include methylcellulose powder, Fibercon, or Metamucil.

Stool softeners (best used as a preventive rather than to soften already-hard stool). Examples include docusate sodium (Colace).

Stimulant laxatives help the bowel to push the stool out. Examples include bisacodyl (Dulcolax) and Senna.

Prescription medications – only available as recommended by a provider with prescribing privileges, include linaclotide (Linzess) and lubiprostone (Amitiza).

Cod liver oil and mineral oil **should be avoided** due to the risk of lipid pneumonia.¹¹

Procedures to Help Stool Evacuation

Procedures to help with bowel emptying include suppositories, enemas, and removal of stool using a finger to perform what is called “manual disimpaction,” by which stool is removed from the rectum by the nurse’s finger. These procedures are more invasive and should only be considered when other methods fail and **only with the recommendation of a licensed healthcare provider**. In SMS, invasive methods should be avoided to prevent rectal injury. Individuals with SMS are prone to a behavioral challenge of inserting objects into their rectum. However, if stool has backed up and/or is very dense and cannot be expelled even with straining, one of these methods may be necessary to prevent a bowel obstruction (blockage of the intestines), which can become a surgical emergency. **None of these methods should be used without the recommendation and instructions of a licensed healthcare provider and sometimes, the supervision and care of a home health licensed nurse.**

APPENDIX 1

Yakima Paste Recipe

2.5 cups of water

4 oz Senna tea leaves

1 pound raisins

1 pound figs

1 pound prunes

1 cup brown sugar

1 cup lemon juice

Boil water, add Senna tea and let it steep for 5 minutes then strain to remove tea leaves. Pour 2 cups of tea water into a pot and add all the fruit and boil for 5 minutes. Remove from heat. Add brown sugar and lemon juice. Blender all ingredients with stick blender or food processor. Store in freezer—the paste will not freeze but should be kept cold.

Spread 1 Tbsp on toast or make a tea or eat it right off the spoon.

Adapted from [Family Cookbook Project](#).

See [Seattle Children's](#) for other recipes.

REFERENCES

1. Smith, A.C., Dykens, E. & Greenberg, F. Sleep disturbance in Smith-Magenis syndrome (del 17 p11.2). *Am J Med Genet* 81, 186-191 (1998).
2. Woolery, M., Duncan, F., Introne, W. & Smith, A. Constipation in Smith-Magenis Syndrome: What Goes In Must Come Out. in *American College of Medical Genetics* (Charlotte, NC, 2012).
3. Woolery, M., et al. A constipation assessment scale for use in pediatric oncology. *J Pediatr Oncol Nurs* 23, 65-74 (2006).
4. Woolery, M. & McGuire, D. Content validity and pre-testing of the pediatric constipation assessment scale. *Journal of Supportive Care in Cancer* 20, S-41 (2012).
5. Woolery, M. & McGuire, D. Content validity of the pediatric constipation assessment scale. *Oncology Nursing Forum* 39, 174-175 (2012).
6. Lewis, S.J. & Heaton, K.W. Stool form scale as a useful guide to intestinal transit time. *Scand J Gastroenterol* 32, 920-924 (1997).
7. Lane, M.M., Czyzewski, D.I., Chumpitazi, B.P. & Shulman, R.J. Reliability and validity of a modified Bristol Stool Form Scale for children. *J Pediatr* 159, 437-441 e431 (2011).
8. Chumpitazi, B.P., et al. Creation and initial evaluation of a Stool Form Scale for children. *J Pediatr* 157, 594-597 (2010).
9. Mugie, S.M., Di Lorenzo, C. & Benninga, M.A. Constipation in childhood. *Nat Rev Gastroenterol Hepatol* 8, 502-511 (2011).
10. Mugie, S.M., Benninga, M.A. & Di Lorenzo, C. Epidemiology of constipation in children and adults: a systematic review. *Best Pract Res Clin Gastroenterol* 25, 3-18 (2011).
11. Robertson, J., Baines, S., Emerson, E. & Hatton, C. Constipation management in people with intellectual disability: A systematic review. *J Appl Res Intellect Disabil* 31, 709-724 (2018).
12. Bohmer, C.J., Taminiu, J.A., Klinkenberg-Knol, E.C. & Meuwissen, S.G. The prevalence of constipation in institutionalized people with intellectual disability. *J Intellect Disabil Res* 45, 212-218 (2001).
13. Ambartsumyan, L. & Rodriguez, L. Bowel management in children with spina bifida. *J Pediatr Rehabil Med* 11, 293-301 (2018).
14. Duboc, H., Coffin, B. & Siproudhis, L. Disruption of Circadian Rhythms and Gut Motility: An Overview of Underlying Mechanisms and Associated Pathologies. *J Clin Gastroenterol* 54, 405-414 (2020).
15. Nurko, S. & Zimmerman, L.A. Evaluation and treatment of constipation in children and adolescents. *Am Fam Physician* 90, 82-90 (2014).
16. Hosseinzadeh, S.T., Poorsaadati, S., Radkani, B. & Forootan, M. Psychological disorders in patients with chronic constipation. *Gastroenterol Hepatol Bed Bench* 4, 159-163 (2011).
17. Jin, L., et al. Systematic review and meta-analysis of the effect of probiotic supplementation on functional constipation in children. *Medicine (Baltimore)* 97, e12174 (2018).



EDUCATION | AWARENESS | RESEARCH
prisms.org