Defecation Disorders in the Neurologically Impaired Child
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The neurologically impaired child with defecation problems, whether constipation, encopresis, or both, presents a therapeutic challenge. Standard treatment regimens may be insufficient to manage defecation disorders in these children. The degree of neurologic involvement will guide the clinician in developing a treatment plan, as well as provide some insight into the potential for normal defecation. It is unlikely that the profoundly mentally retarded, nonambulatory patient will ever be continent of stool, whereas the child with low-level myelomeningocele may obtain continence with appropriate therapy.

In this article, we discuss the normal anatomy and physiology of defecation, the neurologic diseases complicated by constipation, and proposed treatment schemes for the mentally retarded child and the child with myelomeningocele. When faced with defecation disorders in the neurologically handicapped child, the clinician should determine the underlying reason for the stool dysfunction, ascertain the concerns of the parents, offer a realistic goal for therapy, and develop a treatment plan that is both beneficial for the patient and attainable by the parents.

Anorectal Anatomy and the Physiology of Defecation
Defecation occurs as a result of a combination of reflex actions stimulated by the passage of stool from the sigmoid colon into the rectum. Continence, however, requires intact neurologic input, normal anatomy, and the integration of higher cortical function. To better understand the treatment of defecation disorders in children with neuromuscular disorders, an understanding of the normal anatomy, innervation, and the physiology of defecation and continence is important.

The pelvic complex controls normal defecation. It is composed of the levator ani, the puborectalis, the internal anal sphincter (IAS), and the external anal sphincter (EAS) muscles. The levator ani and the puborectalis muscles form a barrier between the pelvis and the perineum, playing an important part in defecation. The levator ani ensures that pelvic contents are not prolapsed or herniated through the anus during defecation. The puborectalis muscle is situated inferior to the levator ani; the inferior aspect is intimately associated with the superior portion of the EAS. Originating from the posterior aspect of the symphysis pubis, the puborectalis muscle forms a sling around the rectum. Innervation of the levator ani and the puborectalis is from the branches of the ventral nerve roots of S2 to S4.
The internal and external sphincter muscles encircle the anal canal. The IAS consists of involuntary smooth muscle, which ends proximal to the end of the EAS. The IAS receives excitatory sympathetic innervation (L5) and inhibitory parasympathetic innervation (S2–S4). The EAS is divided into three sections, extending from the puborectalis to the anal verge. The most superior portion is the deep loop, which represents confluence of the EAS with the puborectalis muscle. The middle portion is the superficial loop, which encircles the anal canal, including the IAS (Figure). The most distal portion of the EAS is the subcutaneous loop; it encircles the distal anal canal, extending inferior to the lower aspect of the IAS. The EAS is composed of voluntary skeletal muscle, innervated by the pudendal nerve (S2–S4). Afferent branches of the pudendal nerve transmit sensory impulses from the anal canal and perianal skin.

At rest, the puborectalis is contracted, creating the acute anorectal angle between the levator ani and the sphincters. The IAS and the EAS are contracted, generating pressure to close the anal canal into a slit-like conformation. The IAS contributes 85% of the resting tone of the anal canal; the remaining pressure is provided by the EAS.24 Voluntary contraction of the EAS can increase the anal canal pressure to twice the normal resting pressure.5

Passage of stool or gas into the rectum leads to distention and a reflex relaxation of the IAS and a reflex contraction of the EAS. The rectal contents come into contact with the anal canal, which can usually discriminate between solids, liquids, and gas. As the rectum becomes progressively more distended, the EAS relaxes and there is an urge to defecate. If the timing, situation, or both are not conducive to defecation, the EAS and the gluteal muscles are voluntarily contracted, stemming the flow of rectal contents. Also, the puborectalis is contracted, increasing the anorectal angle and further deferring defecation. Voluntary EAS contraction fatigues in 1 to 3 minutes25; however, this provides sufficient time for the rectum to accommodate the increased volume. In time, the urge to defecate passes. If defecation is desirable, the person assumes a sitting or squatting position, straightening the anorectal angle. The EAS and the puborectalis are relaxed and the intra-abdominal pressure is increased by Valsava's maneuver, thus expelling the rectal contents.

NEUROMUSCULAR DISORDERS WITH ASSOCIATED CONSTIPATION

The neuromuscular causes of constipation vary extensively. They range from common to rare and from benign to severe. The "level" of involvement can be anywhere from the cerebral cortex to the muscles of the anal sphincters (Table). Often, constipation in neuromuscular disorders is complicated by dietary factors such as inadequate fiber and fluid intake. The following is a differential diagnosis of defecation disorders based on the level of neuromuscular involvement.

The cerebral cortex is important for fecal continence. As outlined above, the urge to defecate can be suppressed by higher cortical function. The power of this control is well recognized in children with stool-withholding behavior. Conversely, the child with developmental delay or mental retardation may not be able to exercise "control," despite normal rectal and anal sensation; in these children defecation remains a "reflex" action, similar to that in infants. The child with static encephalopathy (cerebral palsy) often has defecation difficulties. Constipation may result from a combination of motor abnormalities (hypotonia or hypertonia), mental retardation, and nutritional factors. Encopresis may complicate constipation secondary to "overflow" incontinence or may occur as an isolated problem.
TABLE

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<th>Neuromuscular Disorders That Can Lead to Constipation, Encopresis, or Both</th>
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<td>Cerebral cortex</td>
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*GBS = Guillain–Barre syndrome; IDDM = insulin-dependent diabetes mellitus.*

Spinal cord injury of any etiology (trauma, stroke, tumor, or transverse myelitis) is associated with defective defecation. Approximately 15,000 individuals suffer spinal cord injuries each year, and often these individuals are children and adolescents. Following spinal cord transection, there is permanent loss of voluntary motor and of sensory function, and a temporary loss of reflex functions below the level of the injury. This temporary loss of distal reflex function is part of the “spinal shock” syndrome. With resolution of spinal shock, somatic and visceral reflex functions recover. However, visceral sensation is lost below the level of cord injury.

Constipation is a common complication of spinal cord injury, often presenting during the first month after injury. Patients with high injury (cervical or thoracic) tend to have “proximal” impaction of stool in the transverse colon, whereas those with low injury (lumbosacral) tend to have rectal impaction. Patients with spinal cord injury have an increased transit time and decreased colonic compliance. The presence of a gastrocolic reflex is variable. Rectal findings include loss of sensation but normal rectal compliance and reflex evacuation. Rectal distention leads to normal relaxation of the IAS, but the EAS does not contract. The loss of anorectal sensation and failure of conscious control of the EAS makes fecal continence difficult. So defecation returns to a “reflex” action.

Even more ominous than constipation is the loss of important symptoms and signs of intra-abdominal pathology, namely, abdominal pain, abdominal distention (already present from constipation), localized tenderness, guarding, and rebound tenderness. Undiagnosed abdominal emergencies account for approximately 10% of fatalities in patients with spinal cord injury.

Myelomeningocele is a relatively common birth defect, with an incidence of approximately 1 per 1,000 live births. This anomaly disturbs the structure of the lumbosacral nerves, resulting in dysfunction of the pelvic floor musculature and the EAS, as well as absent or decreased sensation of the anorectal mucosa. Myelomeningocele affects the dorsum of the spinal cord more extensively than the ventral portion. Thus, motor function tends to be less severely involved than sensation. Rectal compliance and accommodation remain normal, as does the relaxation threshold for the IAS. The resting tone of the IAS may be normal or decreased. The major factors contributing to fecal incontinence in myelomeningocele include diminished or absent sensations of the perianal skin and anorectal mucosa, decreased sensation to rectal distention, loss of or diminished voluntary contraction of the EAS, and decreased or absent contraction of the puborectalis and the levator ani.

The probability of bowel and bladder incontinence is high with defects affecting the lumbosacral spine. However, the extent of bowel dys-
function varies with the severity of the defect. Some children may overcome EAS dysfunction by learning to contract gluteal muscles and thereby achieve some degree of continence. In general, children who retain some sensation tend to be more apt to achieve continence.

Spina bifida occulta (SBO) is a common and usually asymptomatic defect occurring in 10% of children. This anomaly represents the benign end of the continuum of the dysraphic disorders, resulting from failure of the posterior vertebral arches of L5 and S1 to fully close. There is absence of herniation of neural tissue through the spinal defect and the skin is completely closed. Thus, as the term implies, spina bifida is “hidden.” Clues to the underlying defect include midline cutaneous findings such as hair tufts, dimples, dermal sinuses, hemangiomas, and lipoid tumors. Some have questioned whether SBO can be a cause of constipation. However, the constipated patient with cutaneous features suggestive of SBO should be evaluated with spine radiographs and, perhaps, magnetic resonance imaging to determine whether there is an underlying spinal cord abnormality.

Multiple sclerosis (MS) is a common neurologic disease with a prevalence of approximately one-quarter million in the United States. The pathophysiology of MS is focal axonal demyelination and resulting plaque formation in the white matter of the brain and spinal cord. Constipation, fecal incontinence, or both are common (43% to 68%) complications of MS. The occurrence of defecation disorders correlates with the severity of MS, with a prevalence of 56% in severely affected persons and 24% in those with mild disease. Factors contributing to constipation and fecal incontinence include prolonged colonic transit time, absent gastrocolic reflex, decreased rectal compliance, decreased resting anal pressure, weakened puborectalis and EAS muscles, and decreased perception of rectal sensation. However, MS is not usually symptomatic until the third to fourth decades of life, making it an unlikely cause of constipation in children.

Two disorders of the anterior horn cells that may have difficult defecation as a complication are poliomyelitis and spinal muscular atrophy. Overall, poliomyelitis has become rare as a result of immunizations. As a result, the majority of cases now follow the administration of live vaccine in the United States. To avoid postimmunization polio, immunization practices are being changed. The use of inactivated, intramuscular polio vaccine for at least the first dose of vaccine will reduce future oral vaccine-induced polio. Spinal muscular atrophy is a disease of infancy or early childhood that is manifest by widespread muscular atrophy. The clinical picture is predominated by severe hypotonia. Constipation occurs as a secondary problem.

Examples of peripheral neuropathy that are complicated by constipation include Guillain–Barré syndrome and neuropathy following longstanding insulin-dependent diabetes mellitus. Guillain–Barré syndrome is an ascending paralysis, probably autoimmune in nature, that follows acute infections. Bowel dysfunction generally improves with resolution of the underlying neuritis. Insulin-dependent diabetes mellitus can result in autonomic neuropathy that may lead to constipation. However, this problem is not restricted to those with neuropathic symptoms. Constipation is found in one-third of patients without neuropathic features.

Two classic neuromuscular junction disorders, myasthenia gravis and botulism, may have associated constipation. Myasthenia gravis is rare in children, but can be seen in the newborns of affected mothers. Infantile botulism is caused by the toxin of Clostridium botulinum and is uncommon in the United States. One of its first signs is constipation, followed by lethargy, poor feeding, progressive weakness, and cranial nerve dysfunction.

The muscular dystrophies are complicated by constipation for a number of reasons: sedentary lifestyle, weak abdominal musculature, and associated visceral smooth muscle myopathy. Any condition that affects the functional integrity of the abdominal musculature (eg, prune-belly syndrome or gastrochisis) can lead to constipation as a result of inadequate production of intra-abdominal pressure.

Diseases of the enteric nervous system and visceral smooth muscles may also produce defecation disorders. In brief, any condition that leads
to hypotonia can result in problems of defecation, including Down's syndrome, hypothyroidism, and central hypotonia.

**TREATMENT STRATEGIES**

The developmentally delayed child is at increased risk for having constipation, encopresis, or both. Contributing factors include inadequate dietary fiber, poor personal hygiene, lack of attention to the urge to defecate, and poor muscle tone. In some of these children, the lack of exercise or the previous prolonged use of laxatives, enemas, or both further increases this problem.\(^25\)

Treatment of the mildly retarded child with constipation should not be significantly different from that of the child with normal development. A regimen that includes a "clean out" phase followed by medical and dietary therapy in combination with behavior modification is often successful. Behavior modification should be based on a positive reward system; negative reinforcement (punishment) should be avoided, as it is counterproductive. The developmental level of the child should be taken into account when planning an appropriate reward system. A 6-year-old child with a developmental quotient of 0.6 has a developmental age of younger than 4 years and will not respond to the same rewards as a 6-year-old child with normal development. Timed "sit downs" on the toilet, particularly after meals (to obtain benefit from the gastrocolic reflex), are helpful in attaining fecal continence. Following successful defecation in the toilet, the child should be praised and rewarded. If encopresis is present, frequent checks of the undergarments and rewarding the child when they are clean has been suggested.\(^24\)

A unique approach was used by Piazza et al. for two mentally retarded children who did not respond to aggressive medical and dietary management and standard behavior techniques. First, the frequency of bowel movements was increased. Then, irrespective of whether defecation was in the toilet or the undergarments, all bowel movements received positive reinforcement. Once the stool frequency had increased to normal, "discrimination training" was begun. Any bowel movement made in the toilet was praised. If the child had an "accident," he or she was brought to the bathroom to watch the stool being placed into the toilet. Next he or she was placed on the toilet for 30 seconds, after which he or she received praise.\(^25\) Although successful in these two children, this technique has not been studied in any scientific fashion, so further studies are in order before this can be recommended as an acceptable technique.

Treatment of constipation in profoundly retarded children, including those with cerebral palsy, is more problematic. Physical inactivity, medications, and defective intestinal innervation and motor activity are potential causative factors in these children. Children fed strictly by gastrostomy tubes may have poor fiber and inadequate fluid intakes, which exacerbate constipation. Continence is not a realistic possibility in profoundly handicapped persons. Rather, the goal of therapy is to make the stool soft so it is painless to pass. To this end, the use of nonstimulatory, osmotic laxatives is beneficial. Additionally, adding fiber to tube feedings improves daily stool weight and bowel function in profoundly retarded, nonambulatory patients.\(^26,27\) The use of fiber may allow the discontinuation of laxatives.\(^28\) Enemas should be reserved for the individual unresponsive to laxatives and fiber supplementation, and then used sparingly.

The therapy of defecation disorders in children with spinal cord injuries and those with myelomeningocele is discussed as a single approach here. The goal for both groups is to promote regular emptying of the bowel to prevent constipation, incontinence, skin breakdown, and the psychological problems associated with fecal incontinence. As in any child with constipation, encopresis, or both, a treatment regimen that includes a combination of dietary manipulation, medication, behavior modification, and positive reinforcement offers the best chance for a desired result. Care must be taken when using laxatives because they might "cure" constipation but worsen encopresis. This is because individuals with abnormal spinal cord function may not be able to control leakage. In these instances, stool softeners may be more appropriate to keep the stools soft and easy to pass.

Scheduled visits to the toilet in combination with positive reinforcement can habituate the
colon to empty at the same time each day. A good time for these trips is after breakfast; this allows
the child to benefit from the gastrocolic reflex
and, if successful, will keep the child clean during
the daytime. An adjunct that improves chances
for success is proper positioning. The child
should fit comfortably on the toilet seat and the
feet should be supported by a flat surface. The
child should then be instructed to bear down to
increase intra-abdominal pressure. Initially,
Attempts at defecation can be augmented with
rectal suppositories. A stimulant suppository
(bisacodyl) should be placed 15 to 20 minutes
before the time of desired bowel movement.
Alternatively, one can use a glycerin suppository
or digital stimulation to provoke "reflex defecation."

Biofeedback has been used with mixed results.
The goal of treatment is to obtain continence by
teaching the child how to increase EAS pressure,
contract the gluteal muscles, or both.29 For
biofeedback to work, some degree of rectal sensa-
tion is required.30 Manometric studies have
revealed that the mean threshold of rectal sensa-
tion is significantly higher in children with
myelomeningocele compared with normal chil-
dren. However, the threshold of the rectosphincteric
reflex is similar in both groups. This combi-
nation promotes encopresis. Other findings that
hinder continence are a decreased anal resting
tone and decreased maximal "squeeze" pres-
sure.31

Initial studies of biofeedback for the treatment
of constipation in children with myelomeningoce-
les demonstrated that continence was
improved in 50% to 75% of treated patients.30,32,33
Success was defined as either complete contin-
ence or at least a 75% decrease in incontinent
stools. However, these studies were uncontrolled.
Whitehead et al. showed that the addition of
biofeedback to a program of behavior modifica-
tion did not significantly boost the overall success
rate. However, when the level of the spinal lesion
was taken into account, children with lesions
below L2 did respond better to a combination of
behavior modification and biofeedback.34

In a controlled study, biofeedback significantly
decreased the frequency of fecal soiling after 12
months, compared with such rates before thera-
py. However, the decrease in frequency of fecal
incontinence was not significantly different when
biofeedback was compared with conservative
treatment.35

In summary, biofeedback is beneficial in a sub-
group of children with myelomeningocele and
defecation disorders. Factors that increase success
include the presence of rectal sensation, sufficient
abdominal musculature to generate increased
intra-abdominal pressure, sufficient strength and
coordination of lower back and gluteal muscles to
produce a "squeeze," a low spinal defect, minimal
global disability, and personal and familial
motivation.8,29

Many children with myelomeningocele contin-
uce to suffer from constipation and fecal inconti-
ence despite an aggressive medical and dietary
treatment regimen. To keep them "clean" during
the day, routine use of enemas may be adopted.
Standard retrograde enemas may not be accept-
able to patient or parents. The patient cannot hold
the enema in the rectum for a sufficient amount of
time to get the full benefit and leakage makes this
an aesthetically unpleasant procedure.

Shandling and Gilmour developed an enema
continence catheter, based on the catheter for giv-
ing barium enemas, to improve the acceptance
and benefit of retrograde enemas. The catheter
consists of a tube with an inflatable balloon at the
end. This is inflated in the rectum and an external
baffle locks the catheter in place. Large volumes
of saline solution can be given through the
catheter without leakage. When ready for defeca-
tion, the patient is placed on the toilet, the
catheter balloon is deflated, the catheter is
removed, and the patient's abdomen is massaged
in an attempt to move the fecal material around
the colon and out of the rectum.35 The ability to
attain continence using this catheter is high and
enemas may be required only every other day to
maintain continence.36,36

Despite the combination of bowel training,
dietary manipulation, laxatives, and retrograde
enemas, some patients continue to have fecal
incontinence. In the past, the only options were
continued incontinence or colostomy. In 1990,
Malone et al. developed a surgical procedure that
constructed an appendicocecostomy through
which antegrade enemas could be given. This
procedure has been termed the antegrade continence enema (ACE) procedure. The original technique brought the appendiceal tip out on the abdominal wall as a stoma; the proximal end was reimplanted in a nonrefluxing manner into the cecum. The stoma is intubated so enemas can be given, with the goal being to keep the colon clean of fecal material.

In the original report, all five patients attained fecal continence when enemas were administered every 48 to 72 hours. Further studies have supported the efficacy of the ACE. The percentage of patients who are completely “clean” or have only mild post-enema leakage has ranged from 71% to 85%. Variations on the original procedure have included the formation of a continent valve by intussuscepting the appendix into the cecum, and the formation of a cecal tube in those without an appendix. Further variations of ACE include the placement of a gastrostomy button in the stoma. This ensures continence and provides a means of keeping the stoma patent. Most recently, a method has been developed for the percutaneous placement of a cecostomy tube for administration of ACE; the results have been good.

Although the success rate for the ACE procedure is high, there is a real potential for complications, including stomal stenosis, stomal “breakdown,” bowel obstruction, cecal torsion, and wound infection. Initially, phosphate solutions were used as the agent for ACE. However, this was associated with phosphate poisoning in some cases. The agent most commonly used today is polyethylene glycol/electrolyte solution. Continence can be maintained with enemas given every 2 to 3 days.

More dramatic than ACE is the use of neuroprosthetic devices to maintain continence. In individuals with myelomeningoceles, neuroprosthetic devices have been implanted to give continuous stimulation of the pudendal nerve and thus contraction of the EAS. When defecation is desired, the stimulation is removed. This modality remains highly experimental.

A stepwise approach should be used when developing a treatment regimen for the child with myelomeningocele and defecation disorders. The following is an example.

First, previous treatments should be reviewed with the parents or guardians. Adequate intake of fluids and fiber should be assessed and corrected as needed. If there is evidence for stool impaction, the child should have a bowel clean out with laxatives, enemas, or both. Once the colon has been emptied, treatment can be initiated. Timed “sit downs” on the toilet after meals should be started in combination with positive reinforcement. If the patient and the parents feel that it will be too difficult to make three trips to the restroom in a day, settle on a single time with the patient and the parents. To give the best chance of daytime cleanliness, breakfast time is a good choice. The morning “sit down” can be supplemented with an enema. As improvement is seen, the morning enema can be spaced out to every other day. If continence is maintained, a glycerin suppository can be used in place of the enema. If continence continues with every-other-day suppositories, the frequency can be decreased as long as continence is maintained. If incontinence returns, go back to the last step in the treatment plan that attained continence. If retrograde enemas are needed to maintain continence, an ACE procedure should be entertained.

In summary, there are a number of neuromuscular disorders that can lead to defecation disorders. An understanding of normal defecation physiology and the pathophysiology of the underlying neuromuscular disease will guide the clinician in the development of a treatment strategy. A stepwise approach to treatment should be followed to ensure adequate treatment. The degree of the child’s impairment should be assessed to guide certain aspects of therapy, including type of behavior modification and positive reinforcement. Also, knowing the degree of neurologic impairment will aid the clinician in counseling the parents as to the potential for continence.

REFERENCES