

Guidelines on Scoliosis Monitoring and Treatment for Children with Prader-Willi Syndrome

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Background

Children with Prader-Willi Syndrome have about a 70% risk of developing scoliosis before the end of growth. Approximately 15% of children with Prader-Willi syndrome will develop severe or significant curves, requiring bracing or surgery. The earlier the curve is detected, the better the possibilities for treating the curve with casting or bracing.

There are two peak ages for scoliosis presentation in children with PWS. Under the age of four years, most of the curves begin as C-shaped, and are most likely related to the hypotonia. The second peak, centered around ten years of age, typically is the more common S-shaped curve that is seen in otherwise typically developing adolescents. Fifteen percent of curves diagnosed before four years of age will require surgical treatment, 41% of curves diagnosed after four years of age will require surgical correction, as per the 2007 PWSA-USA survey of membership. Spinal deformities in children with PWS, especially the young ones, are often picked up late, because the signs are extremely subtle on physical examination. Where most typically developing infants with even a small spine curve will have a noticeable back asymmetry, those with PWS do not. That asymmetry is caused by rotation of the spinal column, called vertebral rotation, causing one side of the rib cage to be more prominent in back when doing the forward bending test, usually the first sign of scoliosis. Children with PWS have less vertebral rotation than children without PWS with scoliosis curves of a similar size. Therefore, the child with PWS may have mild findings clinically, which are overlooked but a moderate curve radiographically. In the past, the delay in making a clinical diagnosis was blamed on obesity, but this does not appear to be the case. Fewer children with PWS now develop obesity, and often the curves are diagnosed prior to the onset of obesity. For that reason, routine screening for spine curves is important, and there should be a much lower threshold for working up clinical findings in children with PWS, compared to otherwise unaffected children.

Conditioning

Children with PWS should be encouraged to be as active as possible, particularly those activities that build up their core, strengthen their abdominals and the back muscles. In addition to sports and recreational activities, focused physical therapy and hippotherapy, emphasizing core muscle strengthening, may help improve a hypotonic

curve in a young patient with a flexible deformity. Kyphosis is the normal rounding of our upper back, which is often increased in persons with PWS. If increased kyphosis is noted to develop, the physical therapy regimen should include specific exercises for hyperextension stretching and strengthening of the upper spine. Some parents have found that their children responded well to Pilates.

Monitoring

Scoliosis in infants with PWS is unlikely to develop until the child is sitting and gravity acts across the spine. Therefore, monitoring of spine should begin when the child first sits independently, usually around the first birthday in PWS. Yearly screening radiographs of the seated or standing child should be used in addition to a clinic examination. If a curve greater than 10° is seen, spine x-rays should be taken more frequently so that any progression can be quickly appreciated.

Casting

Spinal casting has been shown to be effective if scoliosis is detected in the infant before 3 years of age, and even up until 5 or 6 years old. These body or “Mehta” casts can actually reduce the size of the curve. Curves over 20° or 25° should undergo casting, with a goal of decreasing the curves as much as possible. Many curves under 50° can be reduced to under 20° with casting, after which the child is braced for a year, with plans of being brace-free thereafter. The goal with larger curves is to control them and allow the child to grow several more years. Even curves over 90° can obtain some correction with casting, delaying surgery until after 5 years of age. The casts are changed every 2 months in children under the 2 years of age, every 3 months in children between 2 and 3 years, and every 4 months for children over 3 years of age. Casts are continued until the curve has been successfully reduced, or if correction plateaus over successive casting. The child is then braced to maintain the correction.

Bracing

Brace treatment should be considered for curves over 20° to 25° in growing children when casting is not an option. Spine braces come in different styles with different names but are all some version of a thoracic-lumbar-sacral orthosis or TLSO. For smaller, more flexible curves, a nighttime only brace is useful, trying to side-bend the spine as much as possible to straighten the curve; because of the unnatural position, these braces are not for daytime use. Larger curves, over 30°, need a daytime brace as well, countering the effects of gravity during regular daytime activities. Brace wear then increases to 22 hours per day, allowing an extra hour or two out-of-brace for physical activities. When the brace is first fitted, initial radiographs in brace should be taken to verify that the curve shows correction as compared to the out-of-brace radiograph. Smaller spine curves in children with Prader-Willi syndrome are often flexible and the goal should be to obtain a 50% correction, although this is not possible for all curves. Follow-up out-of-brace radiographs are obtained every 4 to 6 months, having the child remain out of brace overnight, until the time of the radiograph.

Surgical Intervention for Scoliosis

The goal of scoliosis treatment is to try to keep the curve under 40° to 50°, because curves larger than that will continue to progress (worsen) even after growth has ended. More importantly, severe curves at any age deform the chest cage, which can seriously affect how the lungs develop, leading to breathing problems. To prevent curve progression in adulthood, or to improve chest shape, curves that are, or will be, over 50° at the end of growth likely need surgical intervention.

Head forward alignment and junctional kyphosis

People with PWS balance themselves differently from their peers, with a characteristic head-forward position. On x-ray, this is seen as either an increased thoracic kyphosis or a cervical-thoracic (base of the neck) junction kyphosis. Experience has shown that attempting to correct the posture to “normal” can lead to failure of the surgery, as the patient works to reestablish their comfortable posture. They will lean forwards, compensating for the rigidly fixed part of the spine by bending above or below the spine rods. This can lead to a sharp kyphosis at these areas, called a proximal (upper) or distal (lower) junctional kyphosis, possibly followed by the spine pulling away from the rods. To avoid this, the proximal extent of the fixation should be as low as possible (the 3rd or 4th thoracic vertebra, called T3 or T4), and a moderate amount of existing thoracic kyphosis can be accepted.

Surgery for the growing spine

In a young, growing child, performing a definitive spinal fusion may improve the child's deformity, but will restrict spine and chest growth. At maturity, that child will be adult sized but with a child sized chest. For these children expandable implants (rods) can control the shape of the spine while allowing for growth. In practice, I try to postpone surgery by bracing until an x-ray of the curve in its brace cannot be held below 50°. The goal of expandable implant surgery is to decrease the curve's size initially, then prevent it from progressing while allowing for spinal growth. There are two kinds of expandable implants, one is the “growth rods” or “non-fusion spinal instrumentation” (NFSI), the other being the “vertical expandable prosthetic titanium rib” (VEPTR) device. The VEPTR device has proven to be problematic in children with PWS, primarily due to the poor bone strength of their ribs, leading to frequent failure from rib fractures. For that reason, we recommend the use of a NFSI. For PWS, the construct that has worked well is having the upper part anchored to two vertebrae in the upper spine, and the lower part anchored to two vertebrae in the lower spine, each with bilateral pedicle screws, for four pedicle screws anchor above and below. The segments are spanned with dual rods. A typical construct would be from the third thoracic vertebra (T3) to the third lumbar vertebra (L3), fusing T3 and T4 to act as the upper anchor, and L2 and L3 as the lower anchor. Except in certain difficult cases, a magnetically actuated rod can be used, so that the rods can be lengthened during a regular office visit, not requiring surgery.

Magnetically actuated rods can be lengthened every 2-4 months in the clinic/office setting, whereas the manually expandable rods are usually lengthened in the operating room every 6 months, to keep up with spinal growth. Near skeletal maturity, the spine may need to undergo a definitive fusion.

Definitive spinal fusion

In the older child, a definite fusion should be performed for curves 50° or more out of brace. A curve of this size has a 95% chance of progressing, even after skeletal maturity. Curves as small as 40° may need to be fused if there is a concern of progression or poor spine balance. Whereas in idiopathic scoliosis the lower age threshold for a definitive procedure is 10-12 years of age, children with PWS often have a later growth spurt, with delayed maturation, possibly related to the use of supplemental growth hormone. For that reason, it is advantageous to wait until 12 years of age for girls and 14 years for boys prior to fusion. Bone mineral density in children with PWS is frequently low, so multi-segmental pedicle screw constructs are recommended, maximizing the number of fixation points. In the typical scoliosis seen in adolescents without PWS, the scoliosis usually has a lordo-scoliosis pattern, meaning that the upper back is actually straighter than usual when seen from the side. In children with PWS most commonly have a kypho-scoliosis, in that the rounding of the upper back as seen from the side is exaggerated. As mentioned above, care should be taken not to over correct the kyphotic deformity, as this may lead to worsening of their cervical-thoracic kyphosis. Also, as with the expandable implants, it is best to keep the upper level of the fusion no higher than T3 or T4, if possible, to prevent proximal junctional kyphosis from developing.

Surgical Planning

In children requiring anesthesia, whether for spinal casting, spine surgery with rods, or even planned interval lengthening of a NFSI, the special characteristics of children with PWS should be well understood by the entire treatment team.

Respiratory

Children with PWS have a number of respiratory issues, some related to their hypotonia, and others to obstructive and/or central sleep apnea. They are at high risk for postoperative pneumonia. The literature reflects a high rate of complications with anterior spinal procedures, and it is strongly recommended to avoid surgeries that enter the chest. Preoperatively, a consultation with a pulmonologist is required, and often a sleep study. A tonsillectomy may be indicated preoperatively for obstructive sleep apnea, and CPAP or BiPAP may be prescribed for after the operation. Postoperatively, extubation (removing the breathing tube) may need to be delayed for a few hours, or even overnight, until a patient has sufficiently awoken for a strong respiratory effort. Hypotonia may cause the patient to have a weak cough, and chest physical therapy should be aggressive.

Bone Mineral Density

Children with PWS have decreased bone mineral density and bone strength, which may lead to surgical hardware losing fixation (pulling out) or bone not fusing (pseudo arthrosis). Pre-operatively, it is important to have vitamin D and calcium levels optimized. Children with PWS who have been actively managed by their endocrinologist will likely have been on long term growth hormone and possibly sex steroid replacement. This treatment can optimize bone strength by puberty, but only if vitamin D and calcium levels have been sufficient. It is a good idea for patients to supplement their vitamin D and calcium intake, especially if surgery is being considered. Although radiographs notoriously underestimate bone mineral quality, a sure sign of an abnormality is if bowel gas pattern on a spine x-ray actually obscures the vertebra it overlaps. Another warning sign is a diet low in dairy products (milk, yogurt, cheese). A DEXA scan may be helpful if 1) there is concern that the bone mineral density is critically low, such as a history of fractures, and 2) there is enough time prior to surgery for the treatments to have an effect. In those critical cases, intravenous pamidronate and vitamin D may make a meaningful difference.

Operative planning should maximize the number of spinal fixation points for performing the instrumentation for fusion. A multi-segmental pedicle screw construct, with fixation at nearly every level has worked well. Occasionally protective bracing after surgery is needed to prevent proximal or distal junctional kyphosis.

Pain Tolerance

Children with PWS have an increased pain tolerance, which may be helpful when attempting to mobilize them after the surgery. But it may also be a reason why they awake slower from anesthesia.

Food Seeking and Gastrointestinal Issues

The family and hospital staff needs to be vigilant for food seeking behavior, and its possible life-threatening consequences. Preoperatively, the patient might violate the NPO instructions, making induction of anesthesia dangerous. Postoperatively, the children should have their diet advanced very slowly until they have normally active bowel sounds and flatus. Usually, they are hungry much earlier than other postoperative patients, but their ileus resolves a day or so later than expected. A typical protocol is 2 ounces of clear fluids every 4 hours starting immediately after surgery. If that is tolerated, it is increased to 4 ounces every 4 hours, and then gradually a soft diet is introduced. Daily abdominal radiographs are used to confirm that the ileus is improving, which may take up to a week to occur. Also, the patient's preoperative calorie restrictions should be well known by the hospital's nutrition staff and observed postoperatively. All treating staff should be aware that children with PWS do not vomit, nor lose appetite. Should either of those occur, they require an emergent nasogastric tube insertion, followed by an abdominal radiograph to evaluate for gastric dilatation,

and possibly a CT scan to rule out free air. Gastroparesis leading to gastric necrosis is unfortunately a common cause of death in children with PWS.

Skin-Picking

A constant threat to the surgical incision is the well-known characteristic of patients with PWS to skin pick, in essence scratching their wounds until they break down, leading to a dehiscence and a deep infection. This is probably the most common post-surgical PWS complication seen. Skin picking is usually direct gouging of the surgical site with the fingers, but it may take the form of rubbing their back against a wall or furniture. We use a light post-operative brace to protect the skin for the first several weeks, even for those children without a history of skin picking, as the stress of surgery may bring about new maladaptive “coping” behavior.

If the above points are carefully observed, surgery can be performed safely and uneventfully in children with PWS.

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