Spina Bifida

Developed by Quinlan Pon, Brianna Salverda, and Dr. Cynthia Gunaratnam for PedsCases.com.
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Introduction:

A: Hello and welcome to this PedsCases podcast on Spina Bifida. My name is Quin Pon, and “my name is Brianna Salverda”, we are 3rd year medical students at the University of Alberta. This podcast was developed in conjunction with Dr. Cynthia Gunaratnam, a general pediatrician at the Stollery Children’s Hospital in Edmonton, Alberta, Canada. In this PedsCases podcast, we will provide you with an overview of spina bifida. To keep it relevant, we’ll take you through some cases.

B: By the end of this podcast, we will cover the following objectives:

1. Define and differentiate: neural tube defects, spina bifida, and myelomeningocele.
2. Discuss the etiology of spina bifida, and how the risk of spina bifida can be reduced.
3. Delineate the diagnostic workup for spina bifida.
4. Discuss the neonatal management of myelomeningocele.
5. Describe the complications associated with spina bifida and what routine surveillance is recommended.

A: During my obstetrics rotation, working in Maternal Fetal Medicine clinic for the day, I met a mother who had an abnormal prenatal genetic screen. Specifically, her alpha-fetoprotein level was elevated. That day, she was coming in for a high-resolution ultrasound and to discuss the implications of these findings for her pregnancy. While reviewing the ultrasound, my preceptor pointed out the out-pouching on the fetus’ back as a neural tube defect and asked me what the most common diagnosis would be.

B: As a very smart medical student, you were able to correctly answer: spina bifida?!

A: Well, yes, but she wanted me to be even more specific. The correct answer was myelomeningocele!

B: What’s the difference?

A: Turns out, “neural tube defects”, is an umbrella term describing congenital abnormalities of the brain and spinal cord caused by incomplete neural tube closure during embryonic development. This subsequently causes abnormal neurulation which

leads to a spectrum of post-natal outcomes varying from anencephaly, where parts of the brain and skull are missing, to spina bifida occulta. Spina Bifida is a subcategory of neural tube defects used to describe abnormalities of the spinal cord specifically. There are two ways Spina Bifida can present: open, meaning there is an opening in the overlying vertebrae *and* skin, and closed (aka spina bifida occulta), where there is an opening in the vertebrae, but the overlying skin is closed. Myelomeningoceles are a specific type of open spina bifida where the meninges and spinal cord are completely exposed! Because myelomeningoceles are the most common type of spina bifida, people often say spina bifida when they mean myelomeningocele, or vice versa.

B: Okay, so myelomeningoceles are a type of spina bifida, and spina bifida is a type of neural tube defect, is that right?

A: Yes!

B: Wow, all of these diagnoses sound very serious. I remember learning something about folic acid supplementation to prevent neural tube defects. Did your patient not take folic acid? Is that why this happened?

A: Well, about 1 in 1000 newborns continue to be affected by myelomeningoceles today. Strategies *have* been implemented to decrease the prevalence in some countries. However, we still don't know exactly what causes spina bifida, so we can't completely prevent it.

We know there's a large genetic component, because monozygotic twins are very likely to share the abnormality. Also, the familial recurrence rate is much higher than the risk rate for the general population. For example, if a couple already has one child with a myelomeningocele, their second baby has a 4% chance of having the same diagnosis (instead of 1 in 1000), and if two children are affected, the third baby has a 10% chance!

B: Apart from genetics, is there anything else? How does folic acid play a role?

A: Yes, other risk factors include decreased maternal folate intake, maternal anti-convulsant therapy, maternal diabetes, and maternal obesity. It's also worth mentioning that while most spina bifida is non-syndromic, a minority of cases are associated with genetic conditions in the fetus such as Trisomy 13 or 18.

Since a major modifiable risk factor is folic acid deficiency, in Canada, women of child-bearing age and pregnant women are advised to take supplements starting at least 3 months prior to conception. The recommended dose varies based on overall risk. For example, if there is a family history of neural tube defects or maternal anticonvulsant use, these women would be recommended a higher dose compared to an otherwise healthy woman. Starting supplementation early is important because primary neurulation is usually complete by just 27 days post conception - before most women even know they're pregnant!

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For my patient, she had no family members with neural tube defects and this was an unplanned pregnancy, so she only started taking her folic acid once she found out she was pregnant at 12 weeks. My MFM preceptor discussed the ultrasound findings with her and offered her amniocentesis with karyotyping. We then briefly talked about the possibilities of 1) fetal surgery 2) post-natal surgery, 3) therapeutic abortion or 4) adoption. She was very overwhelmed and said she would wait for the amniocentesis before making any further decisions.

B: I didn't realize there was all this prenatal evaluation and prevention related to spina bifida. On my NICU rotation, I saw a baby with spina bifida. The mother had poor prenatal care and her only ultrasound was prior to delivery. The ultrasound was suspicious, so the NICU team was at the delivery and we made the diagnosis during my newborn exam. I'll never forget it! The myelomeningocele looked like a raw, red, fleshy membranous outpouching on the baby's lower back. Apparently, the lumbosacral region is where 80% of myelomeningoceles occur.

A: I already find examining newborn babies scary. That must've been even more nerve-racking! What did you do then?

B: Well, the most important thing was preventing infection. When the baby was born, I remember my preceptor instructing me to cover the open lesion with a sterile towel and position the baby on his side. He ordered broad spectrum IV antibiotics and we prepared the baby for urgent transfer to the surgical NICU. While we waited, my preceptor demonstrated some of the neurologic abnormalities to me – this baby's legs were quite limp. Stroking at the thighs evoked some movement, but we didn't notice much of a plantar grasp or response to stroking the feet. He told me that the presentation varies with the location of the myelomeningocele. All nerves at the level of myelomeningocele and below are affected. Apparently, serial exams characterizing motor and sensory function is an important way to prognosticate future outcomes. Higher lesions have a poorer prognosis. It makes sense, since patients with lumbosacral lesions only have the lower extremities, bowel, and bladder function affected, but for patients with higher lesions, other important functions like diaphragmatic innervation, and therefore breathing, can be also be affected.

A: That sounds very different from the typical route of diagnosis my MFM preceptor explained to me. I guess it's because this mother had poor prenatal care. My understanding is that typically the diagnosis of myelomeningoceles is confirmed prenatally. She said usually neural tube defects are suspected when there is an elevated alpha-fetoprotein as part of the QUAD or integrated prenatal screens women are offered at 15-20 weeks. For women who decline these screens, myelomeningoceles and more serious neural tube defects can be detected on the detailed anatomic ultrasound (which is typically done at 18 – 22 weeks). In either case, if there is concern, amniocentesis is offered. If alpha-fetoprotein and acetylcholinesterase levels are elevated in amniocentesis, diagnosis of a neural tube defect is confirmed with 96%

positive predictive value. During the amniocentesis, karyotyping is also done to investigate for associated chromosomal abnormalities, like Trisomy 13 and 18. Molecular genetic testing and a fetal MRI are sometimes done and can help with counselling and therapeutic decisions. Typically, families have this information early enough, to consider a therapeutic abortion. For those who continue with the pregnancy, there are now certain centers offering fetal surgery. Apparently, long-term outcomes are improved with this technique, but complication rates are still high and specific criteria need to be met before mother-babe dyads are given this option. In Canada, post-natal closure is still the treatment of choice for most infants. Regardless of the outcome, I imagine that an earlier diagnosis offers families a chance to plan for their babies' arrival and post-natal journey in a much more controlled and informed way.

B: Agreed! For the family I took care of, it was very shocking and emotional. I learnt that the baby would have surgery to close the lesion within the first 72 hours of life. Apparently, neurosurgery does the initial closure but other specialties such as orthopedics and urology also get involved early.

A: Oh yeah, what are some of those important initial consultations and investigations again?

B: Well, because of the potential for infection and risk of intracranial complications, these babies are admitted to the NICU and the neonatologist usually coordinates their care.

The most important initial consultant is neurosurgery of course! They will prioritize closing the spinal opening within the first 72 hours of life. A post-natal MRI brain and spine is also completed early to assess for associated structural brain abnormalities that may need surgical intervention or increased monitoring. Chiari malformations are common, occurring in 90% of patients with spina bifida. These put infants at risk for brain herniation and may require posterior fossa decompressions by neurosurgery. Hydrocephalus is another concerning complication and ventriculo-peritoneal (VP) shunts may need to be placed. Healthcare providers use serial head circumferences, serial neurologic exams, and radiographic imaging to monitor progression and make therapeutic decisions.

Orthopedics is also typically involved. They look for related spinal curvature abnormalities and associated joint contractures such as club foot and developmental dysplasia of the hip that can result from decreased intrauterine fetal movement. These findings can require early casting or surgery.

Urology is often involved early. This is because all children with spina bifida have neurogenic bladders of varying degrees. Some can have significant vesicoureteric reflux and hydronephrosis leading to kidney injury early on. Typical initial investigations include renal investigations including labs and a renal bladder ultrasound. If there are concerns, a voiding cystourethrogram (VCUG) is also warranted. If a neonate appears

unwell, a related important consideration is urosepsis. Some patients may require intermittent catheterization, vesicostomies, or prophylactic antibiotic therapy and these decisions are typically made in consultation with Urology and Nephrology.

A: The same multi-disciplinary follow-up is also required throughout life! Neurosurgery, Orthopedics, and Urology continue to be involved, but surveillance changes throughout the patient's lifespan, and differs depending on the level of the myelomeningocele and associated complications.

Neurosurgical complications to monitor for include VP shunt malfunction, progression of the Chiari malformation, scar-tissue related tethered cord, and hydromyelia. Imaging and clinical parameters are used accordingly. If seizures arise, neurology may also become involved to take on medical management with anti-epileptics.

If there were any congenital orthopedic deformities, these will require on-going care. In addition, acquired issues resulting from muscle imbalance, paralysis, and decreased sensation often require orthopedic evaluation. Ortho's priority is supporting ambulation by correcting deformities so that patients can use orthoses comfortably. Following spinal balance and intervening to prevent or correct functionally debilitating scoliosis and kyphosis falls under orthopedics' domain as well.

Urology's priorities are to preserve renal function, achieve urinary continence as early as socially acceptable, and to maximize urologic independence throughout life. With neurogenic bladder, chronic urinary retention and overflow urinary incontinence can occur. Anticholinergic agents can be used to help with urinary incontinence, while clean intermittent catheterization can be used to help reduce urinary retention. Managing urinary tract infections carefully in order to protect renal function and reduce antibiotic resistance is also an important consideration. Though operative bladder reconstructions can be done, minimizing these if possible is a priority of urologic surveillance.

The primary pediatrician plays an important role as care coordinator, and monitoring/managing complications with the aid of these specialists as required. Constipation and fecal incontinence are very common childhood concerns that also occur with spina bifida. Any combination of laxatives, stimulants, suppositories, and retrograde colonic enemas can be necessary. Additionally, neurobehavioural monitoring, weight monitoring, skin care, facilitation of mobility and locomotion, and prevention of latex sensitization are all important aspects for the primary pediatrician to focus on. They will typically enlist the support of physiotherapists, occupational therapists, speech pathologists and social workers as well. If there is an intellectual disability or a specific learning disorder, a supportive teacher and school-based team are tremendously important. Often, there are specialized spina bifida clinics where the patient can see many of the members of this multidisciplinary team in one day!

B: Wow! That makes things much more convenient for the patient. With this much monitoring, what does life for patients and their families with spina bifida look like?

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A: Well 75% of patients who undergo myelomeningocele repair in infancy survive into adulthood. Typical life span is currently 30-40 years of age and mortality at this time is most often the result of renal failure. Poorer prognosis is still related to a higher level of lesion, presence of Chiari II malformation with posterior fossa herniation, or presence of hydrocephalus.

Quality of life is poorer in patients with spina bifida compared to healthy individuals and compared to those with other chronic diseases. But almost 50% go on to finish post secondary education, become employed, and have intimate relationships. Some modifiable factors that diminish health-related quality of life include lack of mobility, parenting stress, and pain management. Interestingly, outcomes of surgery, continence status, and level of lesion did not correlate with health-related quality of life.

As healthcare providers, it is important for us to continue to prioritize patient-centered care when we care for our patients with spina bifida. We can do this by understanding the disease, listening to our patients, and prioritizing strategies that improve mortality and their quality of life.

Thank you for listening to our Peds Podcast. Before finishing, we'd like to review our objectives and take-home points:

1. Define and differentiate neural tube defects, spina bifida, and myelomeningocele:
   Spina bifida, often used interchangeably for myelomeningocele, is a type of neural tube defect that affects the spinal cord and leads to exposure of the meninges and spinal cord.
2. Discuss the etiology of spina bifida, and how the risk of spina bifida can be reduced:
   The cause of spina bifida is multifactorial but increasing folic acid intake in pregnancy can help with prevention.
3. Delineate the diagnostic work up for spina bifida
   Typically, the diagnosis is made prenatally via prenatal screening markers and routine or detailed anatomic ultrasounds. More definitively diagnosis and potential genetic diagnoses are made via amniocentesis levels of alpha-feta protein and acetylcholinesterase if the family desires to pursue this more invasive procedure.
4. Discuss the neonatal management of myelomeningocele
   Important aspects of management are careful positioning following birth, preventing CNS and renal infection, and closing the open lesion.
5. Describe the complications associated with spina bifida and what routine surveillance is recommended: Management requires collaboration between several disciplines, with the main players being general pediatrics, neurosurgery, orthopedics, and urology. Exact surveillance depends on the level of the lesion and associated complications. Some complications include Chiari II malformation, hydrocephalus, tethered cord, recurrent UTIs, chronic renal failure, and difficult to manage constipation. General Pediatricians should also monitor

for constipation, learning difficulties, mobilization issues, obesity, skin breakdown, and latex sensitization.

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