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Caring for children with septo-optic dysplasia

Nurses involved in the care of children may encounter this brain disorder, which occurs in 1 out of 10,000 live births.

By Marcia K. Julian, MSN, RN, and Akilia Hang Nguyen, OD

Septo-optic dysplasia (SOD), also known as de Morsier syndrome, is a disorder of early brain development. One of the leading causes of congenital blindness and occurring equally in boys and girls, this disorder is part of a group of midline brain malformations. In the infant with SOD, a disruption in brain development is thought to occur sometime between 4- and 6-weeks' gestation.

This article takes a closer look at SOD, as well as the common challenges associated with it. Patients with SOD benefit from a multidisciplinary approach to care that's tailored to their specific needs, with nurses playing a key role by advocating for and educating families.

What causes SOD?

In most cases of SOD, the precise cause isn't known. It's believed that the disorder is multifactorial, with prenatal environmental factors and genetic predisposition implicated as potential causes. Young maternal age, usually less than age 22, and drug use have been identified in some cases. Other risk factors that have been studied and posited are viral infections, maternal diabetes, certain medications, and a disruption of blood flow to the fetal brain during development.

There are also several gene mutations that have been linked to SOD. The involved genes are HESX1, OTX2, SOX2, and SOX3. These genes have a role in the development of the eyes, pituitary gland, and forebrain. Mutations can result in alterations in the development of these structures. It should be noted that gene mutations have only been found in 1% of patients with SOD. The genetic mutations are also sporadic in nature; therefore, genetic counseling may not be necessary or helpful.

The classic triad

There are three diagnostic features of SOD: optic nerve hypoplasia (ONH), agenesis of the midline brain structures (septum pellucidum and/or corpus callosum), and hypoplasia of the hypothalamic-pituitary axis. Two of the three features must be present for a diagnosis. Approximately 30% of those diagnosed with SOD will have all three components.

The most common manifestation of SOD is ONH; however, a patient may have ONH but not meet the diagnostic criteria

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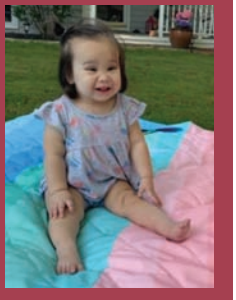
One family's experience with SOD: Maia's story

Maia came into the world 7 weeks early and spent the first month of her life in the neonatal ICU. Her parents knew to expect some delays due to her prematurity; what they didn't know initially was that she was born with SOD.

Despite being born at 33 weeks' gestation, Maia weighed 4 lb, 11 oz at birth. From a respiratory standpoint, Maia did well. She had an isolated episode of apnea in the hospital but, overall, she didn't require supplemental oxygen. Her primary issue early on was feeding. Maia preferred to sleep and it took time and persistence before she was feeding well enough to go home.

It wasn't long after Maia was home that her father voiced concern that he didn't think Maia could see. She didn't look at her parents when they spoke to her and didn't show interest in rattles or toys. At age 4 months, Maia developed irregular eye movements that occurred while she was being fed. Her eyes would dart rapidly, which worsened with fatigue or when her parents would engage in conversation with her. This prompted a visit to her pediatrician, who in turn made a referral to a pediatric neuro-ophthalmologist for a comprehensive evaluation. It was during this visit that Maia was diagnosed with ONH, nystagmus, and legal blindness.

It was determined that Maia would need to have an MRI to rule out intracranial lesions. The results of the MRI showed that Maia had SOD; in fact, Maia is one of the 30% of cases who has all three of the diagnostic criteria for SOD. She has bilateral ONH, hypoplasia of the corpus callosum, and hypoplasia of the pituitary gland. Maia's MRI also revealed a 7 mm intraventricular cyst within the atrium of the left lateral ventricle in her brain and a 4 mm lesion in the right cerebellar hemisphere, thought to possibly be a cavernous venous formation (cavernoma).



Maia was referred to both a pediatric neurologist and pediatric endocrinologist. Her neurologist wasn't concerned about the cavernoma; however, her parents were advised to observe for potential seizure activity. Maia sees a pediatric endocrinologist on a regular basis and has lab studies every 4 months to evaluate her hormone levels. So far, Maia's pituitary gland is keeping up and she hasn't experienced any hormone deficiencies.

It's been over a year since Maia's diagnosis. She was enrolled in several state care programs related to her visual impairment and legal blindness. Her parents were instructed on techniques to maximize her vision. A referral for physical therapy was also made. Maia receives biweekly physical therapy visits, which have been instrumental in helping her achieve developmental milestones and have provided education and support to her parents.

At age 11 months, occupational therapy was added to Maia's care plan. Several months before, Maia developed a food aversion and stopped taking food from a spoon. By incorporating counting and presenting food in a rhythmic pattern, Maia's food aversion has improved and she's more accepting of food. She continues to accept a bottle and can get adequate nutrition with bottle feedings.

Although it isn't clear exactly how much Maia is able to see, it appears that she can see light and movement in the presence of bright light. At age 16 months, she's crawling and exploring her home environment. Although Maia hasn't taken her first step, she's standing and scaling. Maia is a social baby, always happy to laugh and play with her family.

for SOD. With ONH, there are fewer ganglion cells in the optic nerve, which is responsible for carrying information from the eyes to the brain. In patients with SOD, the optic nerve head is one-third to one-half of the normal size, resulting in fewer connections occurring between the eyes and the brain.

Patients who have ONH often present to the healthcare provider with symptoms of visual impairment, such as poor fixation, searching nystagmus (involuntary side-to-side eye movement), visual inattentiveness, or strabismus (eyes that don't look in the same direction). ONH can occur in one or both eyes and visual acuity is highly variable, ranging from 20/20 to no light perception at all. Most ONH cases are bilateral; 80% of these patients are legally blind. Visual acuity and symptoms are generally less severe in unilateral cases and directly proportional to the size of the optic disc.

The midline brain abnormalities associated with SOD involve the septum pellucidum and/or the corpus callosum. The septum pellucidum separates the fluid-filled ventricles in the brain, whereas the

corpus callosum consists of a band of tissue that separates the right and left sides of the brain. Developmental delays are associated with changes in the corpus callosum.

The last feature associated with SOD is pituitary hypoplasia. Located at the base of the brain, the pituitary gland produces hormones needed for growth and reproduction. When the pituitary gland is underdeveloped, there's a risk of inadequate production of necessary hormones, such as growth hormone, adrenocorticotropic hormone, thyroid-stimulating hormone, antidiuretic hormone, luteinizing hormone, and follicle-stimulating hormone (see *Endocrine abnormalities*).

Early detection and treatment of hormone deficits are crucial. Inadequate hormone production can lead to medical problems, such as adrenal insufficiency and diabetes insipidus. In individuals with SOD, the most common hormone deficiency is growth hormone followed by a decrease in thyroid hormones. With a deficiency in growth hormone, slow growth and short stature are seen. When the pituitary gland doesn't produce any hormones, it's known as panhypopituitarism. In an individual with panhypopituitarism, you can expect slow growth, hypoglycemia, genital abnormalities, and problems with sexual development.

Endocrine abnormalities

Endocrine abnormality	Clinical signs
Growth hormone deficiency	• Decreased growth rate • Neonatal hypoglycemia
Hypothyroidism	• Prolonged neonatal jaundice • Decreased growth rate • Developmental delay
Hypocortisolism	• Neonatal hypoglycemia • Hypotension • Recurrent infections • Seizures • Developmental delay • Poikilothermia (inability to regulate body temperature)
Diabetes insipidus	• Polydipsia • Polyuria • Hypernatremia

Clinical presentation

Patients with SOD can exhibit a wide spectrum of symptoms. Some patients may be asymptomatic, whereas others will display severely disabling symptoms. It should be noted that patients with SOD can have normal cognition. The most common manifestations of SOD are visual impairment in one or both eyes, developmental delays related to visual impairment or neurologic problems, seizures, sleep disturbances, and precocious puberty. Other features include short stature, heart problems, obesity, jaundice, anosmia, and low blood glucose levels. In fact, hypoglycemia can be one of the first indicators of pituitary insufficiency. The individual with SOD may also have difficulty regulating thirst, hunger, and body temperature when the hypothalamus is underdeveloped.

In infants, the first indication of SOD may be the development of nystagmus or strabismus, which are common findings in patients with ONH. When an infant presents to the healthcare provider with irregular eye movements, a referral should be made for a thorough assessment by a neuro-ophthalmologist. Initial clinical evaluation should include assessment of direct pupillary response. Sluggish or nonreactive pupils can indicate poor visual acuity or damage to the optic pathway. Evaluation should also include ocular alignment for nystagmus and/or strabismus, which often occurs between ages 3 and 6 months in individuals with this condition.

In cases of ONH where both eyes are affected, nystagmus consisting of involuntary, rapid, repetitive eye movements is most common. In unilateral cases of ONH, strabismus or "eye turn" is more likely to occur. With strabismus, the eye can turn in (esotropia) or turn out (exotropia) and

sometimes shifts between these two patterns. The healthcare provider may observe that infants with these visual impairments prefer a particular head or gaze position to decrease the amplitude of eye movements and create a more stable image.

Once it's determined that a patient has ONH, he or she should have an MRI to evaluate for other indicators of SOD. After the diagnosis of SOD is confirmed, an endocrinology referral should be initiated to ensure the best outcomes.

Managing SOD

There's no cure for SOD, so treatment is focused on management of each patient's individual symptoms. It's imperative that the patient with SOD have access to healthcare. The multidisciplinary care team includes primary care providers, nurses, dietitians, and early childhood intervention specialists, as well as an endocrinologist and ophthalmologist.

An endocrinologist is needed for a comprehensive screening of pituitary function at the time of diagnosis, with follow-up screenings every 6 months so hormone deficits are detected early and appropriate hormone replacement therapy can be initiated. Regular ophthalmologic exams are also recommended to maintain optimum vision. Associated visual conditions, such as strabismus or astigmatism, should be treated. Surgery for the treatment of strabismus is indicated in cases where better visual outcomes can be obtained.

Growth patterns should be carefully monitored in children with SOD, especially when it's known that there's a decrease in the amount of growth hormone produced by the pituitary gland. It should be considered that there's a propensity for obesity in children with SOD, so a dietetic consultation may be helpful to ensure good nutrition and help maintain a healthy weight.

In addition, most children with SOD benefit from physical, occupational, and speech therapies. Early intervention is important to prevent or minimize developmental delays. These patients are at risk for delays in gross motor functions, fine motor functions, language, and social domains. A comprehensive therapeutic approach is essential for management of SOD throughout the lifespan and to improve the patient's long-term prognosis.

IDEA Part C programs

IDEA provides support for children with disabilities from birth through age 21. Intended for children identified as having delays or those at risk for delays, Part C of IDEA consists of early intervention programs for children from birth to age 3. Federal funds are distributed to states to develop programs and offer services. Individual states determine eligibility criteria, and many states have a cost share component. No one can be denied service due to the inability to pay.

The goal of Part C programs is to enhance the development of infants and toddlers with disabilities. An individualized family service plan is developed to provide support and education to parents and caregivers. In Part C programs, the emphasis is placed on the home and community; services are delivered in the child's natural environment.

Family support

Caring for a new baby is challenging enough and learning that their baby has SOD can be overwhelming for new parents. Keep in mind that not all patients with SOD will have the same manifestations and challenges due to the wide variation in symptoms. The eager anticipation that accompanies the birth of a child may now be superseded with fear of the unknown or grief related to the loss of what was expected. It's important for new parents to remember that their baby's most important need is the same as any baby—to be loved and nurtured.

Nurses play an important role by offering reassurance and providing needed education on caring for a newborn with special needs. There's evidence that

infants with special needs and their families benefit from early intervention services.

The Individuals with Disabilities Education Act (IDEA) of 2004 addresses the federal requirement for services and offers support to children with disabilities from birth through age 21. IDEA provides federal funding for Part C, which consists of early intervention programs for children from birth to age 3 (see *IDEA Part C programs*). The goal of an IDEA Part C program is to facilitate the development of infants and toddlers with disabilities. Individual states are tasked with developing programs for individuals identified as having developmental delays or being at risk for a delay. Children with visual impairments often qualify for these types of programs. When enrolled in a Part C program, parents are provided with practical, professional guidance to support their child's development.

Common challenges

Caring for a child with a visual impairment can present unique challenges. He or she may take longer to master developmental milestones, such as sitting, crawling, and walking. Delays may also be present in the following areas: sound localization and orientation, posture and motor skills, tactile exploration skills, receptive and expressive language skills, and social/emotional understanding.

Sleeping is a common area of concern in children with SOD because sleep problems are more common in children who have neurologic disabilities. The child with a congenital malformation of the central nervous system is at higher risk for sleep problems, occurring at a rate of 75% to 80% of affected children. Lack of sleep can have an adverse effect on learning, memory, and reasoning abilities. Mood can also be negatively impacted by lack of sleep, resulting in depression and irritability. In addition, health can be affected by sleep deficits, specifically the immune system and metabolic functions such as glucose metabolism and endocrine function.

Children with SOD are at high risk for sleep difficulties due to decreased light perception and the inability to detect light changes in the environment. Light is important to the maintenance of the circadian rhythm; reduced light perception can lead to arrhythmicity and fragmented sleep. Individuals with agenesis of the corpus callosum have a high incidence of sleep problems, such as greater sleep onset delay, less sleep duration, greater resistance to bedtime, sleep anxiety, night

Sleep promotion measures

Quick tips for developing good sleep patterns in children

- Develop a consistent bedtime routine.
- Use a crib for sleeping only.
- Allow time for self-soothing during the night.
- Limit the length of daytime naps.
- Provide engaging activities during the day.
- Keep a diary of sleep-wake patterns.

The four categories of sleep hygiene

Environmental

- Control the temperature, noise, and lights.
- White noise may help mask unwanted noise.
- Create a comfortable, secure sleeping environment.

Scheduling

- Maintain a regular sleep schedule.
- Keep a regular bedtime and wake-up schedule.
- Maintain a consistent weekend and weekday schedule.

Sleep practice

- Develop a bedtime routine.
- Choose calming activities, such as bathing or reading before bedtime.
- Use soft music or gentle rhythmic touch.

Physiologic

- Be mindful of exercise, meal timing, and caffeine intake.
- Promote physical activity during the daylight hours.
- Encourage daytime exposure to bright light to improve nocturnal melatonin levels.

on the web

American Foundation for the Blind:
www.afb.org

Center for Parent Information and Resources:
www.parentcenterhub.org

National Center for Advancing Translational Services Genetic and Rare Diseases Information Center:
https://rarediseases.info.nih.gov/diseases/7627/septo-optic-dysplasia-spectrum

The Magic Foundation:
www.magicfoundation.org

Wonder Baby: www.wonderbaby.org

waking, parasomnias, sleep disordered breathing (abnormal breathing patterns or insufficient ventilation), and excess daytime sleepiness. Developing a regular sleep routine is especially important for these children.

When a sleep problem is suspected, a comprehensive evaluation should be performed to include current sleep patterns, sleep/wake schedule, and sleep duration. It's helpful to look for factors such as caffeine intake and the sleep environment. Keeping a sleep diary may reveal factors contributing to sleep problems.

Hypnotic medications to induce sleep aren't ideal because they have associated adverse reactions and aren't recommended for use in children. Melatonin has been recommended for disorders related to abnormal circadian rhythm. The first-line treatment for sleep disorders in children with neurologic disorders is to improve sleep hygiene.

Sleep hygiene involves activities to prepare for and promote sleep. There are four categories of sleep promotion measures: environmental, scheduling, sleep practices, and physiologic. These measures are thought to promote sleep by entraining the intrinsic circadian rhythm; however, success could also be the result of behavioral conditioning. Children with SOD may benefit from consistent patterns of stimulation and rest each day (see *Sleep promotion measures*).

It's often normal for children with sensory deficits to self-soothe by rocking or performing repetitive movements during bedtime to help fall asleep. It may become a safety concern if soothing techniques lead to self-harm such as head banging. Safety adaptations should be considered, such as a portable mesh crib versus a traditional crib to prevent injury. Self-soothing may continue through the teenage years or longer in some individuals.

Many children with visual impairments also have sensory food aversions. They often don't tolerate the introduction of food with different textures or may be reluctant to try new foods. Reactions to the aversive foods can range from grimacing to gagging, vomiting, or spitting out the food. The child may become very distressed if forced to eat. Occupational therapists can be beneficial in working with parents to overcome feeding difficulties. Therapy techniques are similar to those used when treating picky eaters. Foods with a variety of tastes, textures, smells, and appearances are presented using rhythm and repetition. It's important to present food in small amounts and be patient to prevent distress associated with food.

Developing a safety plan for the home is especially important with a child who's visually impaired. The safety plan needs to include many of the usual measures that families take when there are young children in the household, such as covering outlets and keeping medications locked and out of reach. Gates need to be in place at the top and bottom of stairs, furniture needs to be secured to prevent tipping, and children shouldn't be left alone with any source of standing water.

Your positive impact

Nurses can have a positive impact on families of children with SOD by recognizing the early signs and symptoms, carefully monitoring growth patterns and development once a diagnosis of SOD is

made, and educating families on available resources to ensure the best outcomes possible. ■

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