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Case report: Causes and complications of myelomeningocele

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Abstract

Neural tube defects are congenital anomalies that occur when the neural tube fails to close during development [1]. They have varied presentations and prevalences worldwide however, predominant risk factors include obesity, poorly controlled diabetes, exposure to certain medications, and folate deficiency [2]. The neural tube typically forms within the first 28 days of life; if formation is impaired, these defects present as anencephaly, encephalocele and spina bifida [3]. Myelomeningocele, a form of spina bifida, affects approximately .5 per 1000 live and stillborn babies within the United States [4]. This report presents a case of a female neonate, who was born at term, and presented with a congenital anomaly. Local and systemic examinations were done, and along with imaging studies, a diagnosis of myelomeningocele was made.

Keywords

myelomeningocele; spina bifida; neural tube defect; folate deficiency.

Introduction

Neural tube defects are the second most common congenital defects seen in the central nervous system. Though relatively uncommon, neural tube defects have a prevalence among live births of 1 in 1200 in the United States [5]. They are typically classified as open or closed defects, the most commonly noted ones being anencephaly and spina bifida.

The central nervous system, consisting of the brain and spinal cord, forms through a process called neurulation, which occurs between weeks 3 and 4 of development in humans. Different neural tube defects reflect the different stages in which neurulation was interrupted. For example, craniorachischisis arises from the failure of the initial neural tube closure site, which results in an open brain and spine. Anencephaly, by comparison, occurs due to abnormalities solely in the cranial neurulation process, leading to cranial anomalies [6].

Spina bifida is a congenital malformation in which the neural tube fails to close caudally, the most severe form being myelomeningocele. These individuals will often have motor and sensory deficits below the level of the lesion since the lesion's anatomic location roughly correlates with the patient's neurologic, motor and sensory deficits [7]. Chiari II malformations, hydrocephalus, urinary and fecal incontinence are often associated with this condition.

Chiari II malformations are classified as herniated cerebellar tonsils and vermis, along with brainstem herniations through the foramen magnum with accompanying towering of the cerebellum. This leads to the blockage of cerebrospinal fluid absorption resulting in hydrocephalus, which is present in 90% of cases of myelomeningocele [8]. Comparatively, cerebellar volume reduction is affiliated with thoracic level spinal lesions more so than that of lumbar or sacral lesions [9].

Most neural tube defects are diagnosed in utero via fetal ultrasonography, blood work (assessing maternal serum first trimester and quadruple screening) and/or amniocentesis as needed [10]. Early diagnosis allows for proper care plans to be established. Diagnoses postnatally are typically done via MRI, X-ray or a CT scan to assess the degree of severity. This report presents a case of myelomeningocele in a full term female neonate, who was diagnosed postnatally.

Case Report

A newborn baby girl presented to the neonatal intensive care unit due to presentation of a notable outpouching in her lower back. She was delivered via spontaneous vaginal delivery at 39 weeks gestation to a 17-year old primigravida. Prenatal care was delayed due the mother being homeless at the time. Maternal HIV, syphilis, HSV and group B streptococcus statuses were all unknown. Prophylactic antibiotics were given for group B streptococcus colonizations.

At birth, the baby presented with a notable outpouching located on the lower back. Vitals signs were noted as 97.6 F temperature, blood pressure of 79/40 mmHg, respiratory rate of 66 breaths per minute, pulse rate of 140 beats per minute and pulse ox measured 100%.

Apgar scores at birth were 7 at 1 min and 8 at 5 mins. Birth weight was 2892 grams, length was 49 cm and head circumference was 29.5 cm. Physical exam was notable for a 3.5 cm soft mass located in the thoracolumbar region that was covered by overlying skin.

An ultrasound of the pediatric spinal canal showed a soft mass over the lumbar spine with intact skin. There was an outpouching of the lower end of the lumbar spinal canal that protruded within the soft tissue at the level of the sacral dimple. Internal echoes were noted within this area that were surrounded by fluid. The ultrasound showed no significant movement of the conus medullaris. Findings were highly suspicious of a myelomeningocele with possible tethering of the spinal cord.

The baby was transferred to the nearby hospital for surgery follow-up three days later.

Discussion

Both genetic and non-genetic factors contribute to the development of neural tube defects. Chromosomal anomalies such as trisomies 13 and 18 along with triploidy account for less than 10% of all neural tube defect cases. The majority of cases, however, arise from non-syndromic, isolated causes, which exhibit sporadic patterns of occurrence [11]. In this particular case, the lack of prenatal care served as a catalyst for the development of myelomeningoceles in the newborn. Various randomized clinical trials have all provided evidence that folic acid supplements can prevent neural tube defects in pregnancies [12]. High risk women, such as those with a previous history of affected pregnancies, should take 4mg of folic acid, compared to those at low risk who are required to take 0.4 mg [13].

Other maternal risk factors include alcohol and caffeine consumption, smoking, elevated glycaemic index and low levels of vitamin C, vitamin B12, choline and zinc [14]. In addition, hyperthermia during the first trimester, whether it is internal such as a febrile illness, or external such as hot tubs and saunas, imitate teratogens and can contribute to the development of neural tube defects [15]. Maternal stress levels which included job changes, residential moves, injuries and lack of social support also strengthened the NTD risk with an odds ratio of 2.9 (95% confidence interval = 1.8-4.7) compared to mothers experiencing no events [16]. Our patient's mother also had a complicated prenatal course, in which prenatal care was unavailable due to inadequate social and housing support. Until the third trimester, the mother was homeless and had no access to obstetrical healthcare. In addition to the lack of folic acid supplementation, maternal stress levels were also increased due to her unforeseen circumstances, leading to an increased risk for the formation of a neural tube defect.

Neural tube defects were first diagnosed prenatally in the 1970s with the discovery of elevated concentrations of alpha-fetoprotein in amniotic fluid samples from pregnancies with anencephaly or myelomeningocele [17,18]. Later, acetylcholinesterase was also shown to be diagnostic [19]. However, biochemical screening for myelomeningocele became unnecessary as ultrasounds offered greater sensitivity and specificity. Currently, biochemical screening is indicated for cases of maternal obesity due to the inability to accurately observe the details of the fetal anatomy during ultrasound examinations [14]. Obesity associated risks are stronger for spina bifida than for anencephaly and cannot be reduced with folic acid supplements [20].

After prenatal diagnosing, individuals are then assessed for the use of prenatal or postnatal surgical procedures. There have been numerous cases accounting for the success of prenatal repairs of myelomeningoceles. As such, it is suggested that the neural tube defects present may be progressive in utero, and early interventions may lead to inhibiting further loss of functioning. The mortality rate of prenatal myelomeningocele repair is about 10-15%, with most deaths occurring before the age of 4. At least 70% of those who survive the procedure will have normal intelligence, but they may present with a high prevalence of learning or seizure disorders when compared to the general population [21].

Medical management of individuals affected with spina bifida is best provided by a multidisciplinary team consisting of a physician, neurosurgeon, urologist, physical therapist, social worker, orthopedic surgeon and nurses who are specialized in taking care of disabled children. Myelomeningoceles are typically managed with surgery within 48 hrs of birth when done postnatally. This minimizes the risk of an ascending infection that can result in meningitis. Orthopedic deformities are also treated with long term follow up. Urodynamic studies are monitored in order to detect complications caused by bladder dysfunction [14].

The Management of Myelomeningocele Study identified that prenatal repair of myelomeningocele reduced hindbrain herniation and the need for cerebrospinal fluid shunting at 12 months of age. It also noted that prenatal repair showed an increase in motor function in children at age 30 months when compared to those who had postnatal myelomeningocele repair [22].

In a study comparing school aged children, ages 5.9 - 10.3 years, who had either prenatal or postnatal myelomeningocele repair, it was noted there were statistically significant differences between surgery groups in overall adaptive behaviors. In comparison to postnatal repair, long-term benefits of prenatal surgery showed continued improvement in mobility and independent functioning along with fewer surgeries needed for shunt replacement and revisions. There was, however, no evidence to show noted improved cognitive functioning when comparing either groups [23].

Despite surgical repair, myelomeningocele is one of the most prevalent causes of neurogenic bladder in children, presenting with life-long functional disabilities. They can also present with paraplegia, sphincter anomalies as well as sensory and motor impairments that worsen with age and abnormal sexual development in adulthood [24]. Mortality rates increase in those who also have respiratory, cardiac or renal anomalies as well [25]. Other clinical features include hypertonia, pain, vertebral anomalies, tethered cord, as well as psychological and cognitive complications. The degree of neurological impairment is, however, predominantly linked to the level at which the malformation is present [26].

Individuals with spina bifida have a reduced health-related quality of life compared to those without it. These individuals exhibit higher levels of depression and lower levels of self-esteem [27]. They also have difficulties in their social lives and tend to have fewer friends, as well as date less frequently in their adolescent years [27,28]. Research on families with children who have spina bifida clinically exhibit between 10-15% of a disruption in normative family functioning [29]. Parents of these individuals feel incompetent, isolated and have a less optimistic view about the future than comparison parents [30,32]. The best predictors for successful adult milestones in individuals with spina bifida include an absence of hydrocephalus, intrinsic motivation, socioeconomic status and accessibility to healthcare [33,35].

Conclusion

Spina bifida is one of the most common forms of neural tube defects, with myelomeningocele being the most severe form. While typically diagnosed in utero via prenatal screenings and ultrasounds, for those who lack prenatal care or support, such as our patient, myelomeningoceles can be diagnosed postnatally.

Postnatal diagnosing may lead to further or worsened motor and neurological complications due to the late onset of repair. Prenatal folate supplementation has been shown to decrease the prevalence and in most cases prevent the formation of neural tube defects [12]. For individuals who are diagnosed postnatally, or opt for postnatal management, surgical interventions are available, though prenatal interventions have been clinically proven to show a more improved way of life [23].

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