

# Case Report on Myelomeningocele

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## ABSTRACT:

Myelomeningocele is a type of spina bifida, a congenital condition characterized by the incomplete closure of the neural tube during fetal development. Specifically, myelomeningocele involves the protrusion of the spinal cord and its covering membranes through a defect in the vertebrae, resulting in a sac or cyst on the baby's back. This sac may contain cerebrospinal fluid, part of the spinal cord, and nerves, leading to varying degrees of neurological impairment below the defect. Common complications associated with myelomeningocele include paralysis, loss of sensation, bladder and bowel dysfunction, orthopaedic issues, and hydrocephalus. Treatment typically involves surgical repair of the spinal defect shortly after birth to minimize the risk of infection and further damage to the spinal cord. Long-term management may require multidisciplinary care involving neurosurgeons, orthopaedic surgeons, urologists, physical therapists, and other specialists to address the complex needs of individuals affected by myelomeningocele. Early intervention and ongoing support are crucial to optimize outcomes and quality of life for affected individuals.

**KEYWORDS:** Myelomeningocele, spina bifida, neural tube, cerebrospinal fluid, paralysis, spinal cord, quality of life.

## INTRODUCTION:

**Myelomeningocele** represents a neural tube defect (NTD) that emerges during fetal development. This congenital anomaly is characterized by the inadequate closure of the neural tube, the precursor to the spinal cord and its surrounding structures. Specifically, myelomeningocele manifests as an improper closure of the lower back portion of the neural tube, resulting in a noticeable gap or opening in the spinal column. In individuals affected by myelomeningocele, the spinal cord and the meninges (protective layers covering the spinal cord) extend through the back opening, forming a sac or cyst. This sac may contain cerebrospinal fluid, nerve tissue, or a combination of both. The severity of myelomeningocele can vary, often leading to neurological and musculoskeletal complications, including paralysis, sensory deficits, and other issues that depend on the location and extent of the spinal cord involvement. Typically, myelomeningocele is diagnosed prenatally using screening techniques such as ultrasound. Treatment commonly involves surgical intervention to repair the spinal defect shortly after birth. Subsequent medical management and rehabilitation aim to address associated complications and support the child's development.

The **history** of myelomeningocele, like many congenital conditions, is intertwined with the broader understanding of embryonic development and medical advancements over the years. Here is an overview of the historical aspects:

## 1. Early Observations (Antiquity to Middle Ages):

Before the understanding of embryology, historical records do not specifically describe myelomeningocele. However, certain historical accounts may have documented instances of babies born with visible spinal abnormalities.

## 2. Renaissance and Early Modern Period (15th-18th centuries):

Medical knowledge began to advance during the Renaissance, but specific details about myelomeningocele were likely limited. Anatomical studies by figures like Andreas Vesalius provided a more detailed understanding of human anatomy.

## 3. 19th Century:

As medical science progressed, there was a growing interest in developmental anomalies. The concept of neural tube defects, including myelomeningocele, became better understood during this period. However, surgical interventions were limited, and the prognosis for affected individuals remained poor.

## 4. 20th Century:

- The early 20th century saw significant progress in surgical techniques and medical imaging. With the development of technologies like X-rays and later ultrasound, it became possible to detect and diagnose myelomeningocele before birth. This allowed for better preparation and planning for surgical interventions.
- In the mid-20th century, pioneering surgeons, such as Dr. Kenneth Salyer, contributed to the improvement of surgical techniques for repairing myelomeningocele. Advancements in anaesthesia, infection control, and perioperative care also enhanced the outcomes of surgeries.
- The understanding of the association between folic acid deficiency during pregnancy and the increased risk of neural tube defects, including myelomeningocele, became clearer in the late 20th century. This knowledge led to public health initiatives promoting folic acid supplementation for pregnant women to reduce the incidence of these defects.

## 5. 21st Century:

Ongoing research continues to explore genetic and environmental factors contributing to myelomeningocele, as well as improved treatment options and long-term care strategies.

The **aetiology** of myelomeningocele encompasses a blend of genetic, environmental, and nutritional elements. The genesis of myelomeningocele is linked to a failure in the closure of the neural tube during embryonic development. The neural tube takes shape early in pregnancy, and inadequate closure can lead to various neural tube defects, including myelomeningocele. The following are key contributors to the aetiology of myelomeningocele:

## 1. Genetic Factors:

- Evidence suggests that genetic factors contribute to myelomeningocele development. Certain genetic mutations or variations may heighten the risk of neural tube defects in offspring.
  - Family history is a potential contributing factor, as individuals with a familial predisposition to neural tube defects may face an increased likelihood of having a child with myelomeningocele.
- 2. Folic Acid Deficiency:**
- Inadequate intake of folic acid, a B-vitamin, during early pregnancy is a well-established risk factor for neural tube defects, including myelomeningocele.
  - Folic acid plays a critical role in proper neural tube closure, and supplementation before and during pregnancy has demonstrated a significant reduction in the risk of these defects.
- 3. Environmental Factors:**
- Exposure to certain environmental factors during pregnancy may contribute to myelomeningocele development, including certain medications, toxins, and substances that can disrupt embryonic development.
  - Maternal conditions such as diabetes and obesity have been linked to an increased risk of neural tube defects.
- 4. Multifactorial Nature:**
- Myelomeningocele is often considered a multifactorial condition, where both genetic and environmental factors likely interact to influence its occurrence.
  - The specific combination of genetic susceptibility and environmental exposures may vary among individuals, contributing to the diversity of myelomeningocele cases.
- 5. Unknown Factors:**
- Despite significant strides in understanding myelomeningocele's aetiology, the precise cause remains unknown in many cases. It is probable that a combination of genetic and environmental factors, along with complex interactions, contributes to the condition's development.

The **epidemiology** of myelomeningocele involves studying its occurrence, distribution, and prevalence in populations. Key points include:

1. **Incidence:** Myelomeningocele's occurrence varies globally, estimated at 1 to 2 per 1,000 live births, with differences based on ethnicity, location, and socioeconomic status.
2. **Geographical Variation:** Rates may differ due to genetic, environmental, or nutritional factors, with some regions implementing folate fortification programs to reduce incidence.
3. **Ethnic and Racial Differences:** Certain groups, like Hispanic populations, may have higher rates due to genetic factors.
4. **Gender and Age Patterns:** It affects both genders equally and is typically diagnosed prenatally or after birth, depending on medical resources.
5. **Socioeconomic Factors:** Limited healthcare access, poor nutrition, and lack of awareness about folic acid supplementation contribute to higher rates in some socioeconomic groups.
6. **Trends and Prevention Efforts:** Folic acid programs have reduced neural tube defects, and ongoing efforts focus on raising awareness about folic acid's importance in pregnancy.

The **pathophysiology** of myelomeningocele involves abnormalities in the development of the neural tube during embryogenesis, resulting in a structural defect affecting the spinal cord and its surrounding structures. Here's a breakdown of the key aspects:

**1. Normal Neural Tube Development:**

- The neural tube forms from the ectodermal layer in early embryonic development, eventually forming the brain and spinal cord through a process called neurulation.

**2. Failure of Neural Tube Closure:**

- Myelomeningocele involves incomplete closure of the neural tube, particularly in the lower back region, often occurring in the first few weeks of pregnancy.
- This incomplete closure leads to an opening in the spinal column, allowing the spinal cord and meninges to protrude.

**3. Sac Formation:**

- Protrusion of neural tissue and meninges through the opening creates a sac or cyst on the individual's back, varying in size and contents.

**4. Location and Severity:**

- The severity and location of the defect vary along the spine, with higher defects resulting in more significant neurological dysfunction.
- The amount of exposed neural tissue and involvement of motor and sensory nerves determine the functional deficits.

**5. Neurological and Musculoskeletal Complications:**

- Exposed neural tissue is prone to damage, leading to paralysis, sensory impairment, and lower limb dysfunction.
- Abnormal muscle and bone development in the affected area can cause orthopaedic issues.

**6. Hydrocephalus:**

- About 80-90% of individuals with myelomeningocele develop hydrocephalus due to obstruction in cerebrospinal fluid flow, leading to increased pressure within the skull.

**DIAGNOSIS:**

- 1. Prenatal Screening:** Myelomeningocele is often detected during routine prenatal ultrasound examinations. Elevated levels of alpha-fetoprotein (AFP) in the amniotic fluid or maternal serum may also indicate a neural tube defect, prompting further investigation.
- 2. Diagnostic Imaging:** After an abnormal ultrasound or AFP test result, more detailed imaging studies such as fetal MRI (magnetic resonance imaging) may be performed to confirm the diagnosis and assess the severity of the defect. MRI provides clearer images of the fetal spine and associated abnormalities.
- 3. Physical Examination:** Upon birth, a physical examination of the newborn is conducted. Myelomeningocele is typically evident as a visible sac or lump on the back, often covered by a thin membrane, at the site of the spinal defect. The sac may contain neural tissue, spinal cord, and meninges.

4. **Neurological Assessment:** A thorough neurological assessment is crucial to evaluate the extent of spinal cord involvement and associated neurological deficits. This may include testing reflexes, muscle strength, sensation, and coordination.
5. **Additional Tests:** Depending on the individual case, additional tests such as ultrasound of the kidneys, echocardiogram, and other imaging studies may be performed to assess associated anomalies. Since myelomeningocele is often associated with other congenital abnormalities affecting various organ systems, a comprehensive evaluation is essential.

The **signs and symptoms** of myelomeningocele, a congenital condition characterized by the protrusion of the spinal cord and meninges through a spinal opening, vary in severity depending on the extent and location of the defect. Common manifestations include:

**1. Visible Spinal Defect:**

- A noticeable sac or cyst on the back, typically in the lumbar or sacral region, containing cerebrospinal fluid, nerves, and meninges.

**2. Neurological Deficits:**

- Paralysis or weakness in the lower limbs, with the degree of impairment determined by the level of the spinal defect.
- Sensory deficits, such as altered or diminished sensation below the defect.

**3. Orthopaedic Complications:**

- Muscle dysfunction, joint contractures, and orthopedic problems due to abnormal muscle and bone development.
- Possible development of clubfoot resulting from muscle imbalances.

**4. Bladder and Bowel Dysfunction:**

- Impaired bladder and bowel control, leading to incontinence or difficulty with voluntary function.
- Increased risk of urinary tract infections due to incomplete bladder emptying.

**5. Hydrocephalus:**

- Seen in 80-90% of cases, characterized by an accumulation of cerebrospinal fluid in the brain, presenting symptoms like an enlarged head, vomiting, irritability, and changes in consciousness.

**6. Skin Complications:**

- Vulnerability to injury and infection of the exposed spinal cord necessitates meticulous skin care to prevent pressure sores and infections.

**7. Motor and Developmental Delays:**

- Delayed motor development and achievement of developmental milestones due to neurological deficits and associated challenges.

**8. Tethered Cord Syndrome:**

- Scar tissue or abnormal spinal cord growth post-surgery can tether the spinal cord, leading to symptoms like back pain, leg weakness, and changes in bowel and bladder function.

**MANAGEMENT:**

Treating myelomeningocele typically involves a comprehensive approach, addressing surgical repair of the spinal defect and managing associated issues. The aim is to reduce neurological deficits, prevent infections, and enhance overall quality of life. Here's an outline of the treatment:

**1. Surgical Repair:**

- Closing the Spinal Defect: Surgery is the primary treatment, performed shortly after birth to close the spinal opening and protect the exposed nerves.
- Timing: Surgery timing varies based on the infant's health, often within the first 24 to 72 hours.

**2. Hydrocephalus Management:**

- Ventriculoperitoneal Shunt: If hydrocephalus occurs, a shunt may be implanted to drain excess fluid and relieve pressure.

**3. Orthopaedic and Neurological Care:**

- Physical Therapy: Rehabilitation, including physical therapy, helps with musculoskeletal issues and motor function.
- Orthopaedic Interventions: Bracing or surgeries may be needed for proper skeletal development.

**4. Bladder and Bowel Management:**

- Catheterization and Medications: Managing bladder and bowel dysfunction with catheterization and medications is common.

**5. Skin Care and Infection Prevention:**

- Skin Protection: Vigilant skin care prevents pressure sores and infections due to the exposed spinal cord.
- Antibiotics: Prescribed to prevent postoperative infections, especially meningitis.

**6. Supportive Services:**

- Educational and Psychosocial Support: Counselling and educational support help address developmental and psychosocial challenges.
- Social Services: Aid in accessing resources and support groups for caregivers and affected individuals.

**7. Ongoing Monitoring and Follow-Up:**

- Lifelong monitoring and multidisciplinary care ensure evolving needs are addressed and interventions provided.

**CASE REPORT:**

Master SMA 4 months 9 days old male child weighing 4.61kgs, a case of conservatively managed meningomyelocele and s/p right parietal VP shunt, was brought by parents as they noticed gradual increase in the size of head for 10 days. On examination his vitals are normal. Sodium:138, Potassium – 5.4mmol- high, Creatinine- on 01/02/2024- 0.34mg/dl and on 02/02/2024- 0.31 mg/dl.Hb- 10.7%, platelets: 3.75. Anterior fontanelle was full and sunset eye sign was positive. Diagnosis of hydrocephalus secondary to meningomyelocele was considered. Child was noted to have hyperkalaemia and was corrected with salbutamol nebulisation and calcium gluconate. Under sedation MRI was done and it showed myelomeningocele at lumbosacral region with fatty filum terminale

causing tethered cord and gross ventriculomegaly without any obvious obstruction and right shunt tube in situ. Patient diagnosis supports the criteria of **“sacral limited dorsal myeloschisis (saccular type), Type 2 Chiari malformation, congenital communicating hydrocephalus”**.

Discussion:

A 4months 9days male child was brought to the hospital by his parents after noticing the gradual increase in the size of his head. Parents were counselled regarding diagnosis and explained requirement of repair of meningomyelocele with untethering cord by excising filum terminale and shunt revision as possibility of shunt non-functioning was there. After preoperative anaesthetic assessment and evaluation, he underwent EXCISION OF SACCULAR LIMITED DORSAL MYELOSHISIS AND SECTIONING OF FATTY FILUM TERMINALE AND REVISION OF VP SHUNT ON RIGHT SIDE AND PLACEMENT OF LEFT FRONTAL VP SHUNT ON 02/02/2024. As right ventricular catheter was placed in right temporal and left behind and new catheter were placed in right temporal and left frontal and connected to Y catheter. Post operative period was uneventful. Child is stable, playing and alert. There is no new onset neurological deficit. He is being discharged in stable condition with following medications-

1. Taxim O 10ML drops- 0.80ml BD- PO- 5days
2. Lizoforce 30ml dry syrup- 2ml- TID- PO- 5days
3. Calpol drops 15ml- 0.60ml- TID- PO- 5days

Surgery notes: repair of MMC excision of dermal sinus tract and untethering of fatty filum terminale on 02/02/2024.

Findings: 2 x 2 cm swelling over sacral area with tuft of hair over the swelling and area of hyperpigmentation.

Swelling was connected to the fatty filum terminale through the dural defect and densely adherent to surrounding tissue.

Posterior bony elements were deficit in the lumbo-sacral area.

Filum was thick and densely adherent to the dura which was separated and cut.

Swelling was excised gross totally which contains fat and fibrous tissue

Dura repaired primarily and closure done in layers.

Revision of ventricular end on right side and placement of left frontal ventricular shunt on 02/02/2024

Findings

Right parietal VP shunt, cranial end was exposed, No CSF flow was noted from ventricular end.

Ventricular catheter was adherent to the brain parenchyma so it was left behind.

New burr hole made posterior to the previous burr hole, ventricular was tapped and connected to low pressure system with Y catheter

Left frontal burr hole made and connected to the Y connector at right parietal side.

Abdominal end was not explored.

CONCLUSION:

The child was diagnosed with Sacral limited dorsal myeloschisis (saccular type, Type 2 Chiari malformation, congenital communicating hydrocephalus.As it represents a neural tube defect (NTD) that emerges during fetal development. This congenital anomaly is characterized by the inadequate



closure of the neural tube, the precursor to the spinal cord and its surrounding structures. He underwent surgery and was managed with antibiotics to prevent infection, paracetamol for pain and normal saline for hydration.

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