

## Spina Bifida

**Condition:** Spina bifida is a birth defect that occurs when the spinal cord does not completely close during fetal development. Types of defects include anencephaly, spina bifida occulta, meningocele, and myelomeningocele (MM). The MM form is typically the condition referred to as “spina bifida” and involves protrusion of the membrane that covers the spinal cord and the spinal cord itself from the back. This requires a surgical closure within the first few days after birth.

**Background:** MM results from failure of normal prenatal development of the neural tube, which is usually completely closed by 1<sup>st</sup> month of pregnancy. MM is a non-progressive disorder, but associated conditions can progress over time.

**Risk Factors:** Most cases occur occasionally but have many causes. Risk factors in the mother include folic acid deficiency, obesity, diabetes, increased body temperature, and certain medications. Folate is added to food items like cereal and bread products to counter folate deficiency in diet. The risk for recurrence in a family is 3-5%, meaning if parents have a child with MM, there is a 3-5% chance of having another child with MM.

**History and Symptoms:** In newborns, a build-up of fluid in the brain causing increased pressure (hydrocephalus) is usually present, requiring a shunt to be placed in the brain. Difficulties with feeding, breathing, or swallowing are typical in babies. Developmental delays are also common. Weakness (even paralysis) in the legs, bowel and bladder problems, seizures, and scoliosis are often present. Vision and mental impairments may be present. During adolescence, problems with walking and physical activity are common. Patients may also have a “tethering” of their repaired spinal cord, where the spinal cord is stretched due to attachment to the scar tissues, which can lead to further pain and weakness. In adults, obesity, blood clots, and metabolic syndrome commonly occur.

**Physical Exam:** A physical exam will be performed to evaluate the nerves, mental ability, sensation, strength, reflexes, and walking ability of the patient. The spine, shoulders, hips, knees, and feet will be examined. Head circumference will be measured in combination with evaluation of growth of the patient. Skin integrity at the site of the open spinal cord will be assessed as well. Developmental (gross motor, fine motor, language, and mental ability), behavior, ability to take care of oneself, and mood evaluations are essential.

**Diagnostic Process:** A blood test of the mother during pregnancy at 16-18 weeks gestation can be used for screening, and diagnosis of spina bifida can be confirmed with ultrasound and amniocentesis, which involves testing a small sample of amniotic fluid. In patients with suspected spina bifida, MRI of the spine can be used to evaluate for complications. Brain CT scans can be used to evaluate hydrocephalus and viability of a shunt, and X-rays can be used to evaluate scoliosis. Other tests may be used to investigate specific symptoms in patients with spina bifida.

**Rehab Management:** A physical medicine and rehabilitation (PM&R) physician is trained in the management of this specific patient population. Rehab plan varies based on the level of the lesion, developmental age of the child, other factors like cognitive impairment and family resources. After initial surgical management, patients are provided with therapies with focus on functional activities, including walking, bladder and bowel management. Appropriate braces, wheelchairs, walkers, adaptive gait aids are

prescribed. Involvement of schools is important in ensuring the child receives adequate resources and assistance for education. Bladder studies, Vision and hearing screens, Neuropsychological assessments are performed to monitor function.

**Other Resources for Patients and Families:** Families of children with spina bifida require extensive education on the complexity of the defect and the medical needs of the child. Support and advocacy groups, counseling, and respite care are available for families. Community resources for recreational activities and support may be helpful for patients.

## Frequently Asked Questions

### What is PM&R?

Physical medicine and rehabilitation (PM&R), also known as physiatry, is a primary medical specialty that aims to enhance and restore functional ability and improve quality of life to those with injuries, physical impairments or disabilities affecting the brain, spinal cord, nerves, bones, joints, ligaments, muscles and tendons. PM&R physicians, known as physiatrists, evaluate and treat the whole body, maximize patients' independence in their daily life and are experts in designing comprehensive, patient-centered treatment plans to empower patients to achieve their goals. By taking the whole body into account, they can accurately pin-point problems, decrease pain, assist in recovery from devastating injuries and maximize overall outcomes and performance with non-surgical and peri-surgical options. To learn more, visit [www.aapmr.org/aboutpmr](http://www.aapmr.org/aboutpmr).

### What makes PM&R physicians unique?

PM&R physicians' training focuses not just on treating medical conditions, but on enhancing the patient's performance and quality of life in the context of those medical conditions. They focus not only on one part of the body, but instead on the development of a comprehensive program for putting the pieces of a person's life back together – medically, socially, emotionally and vocationally – after injury or disease. PM&R physicians make and manage medical diagnoses, design a treatment plan and prescribe the therapies that physical therapists or other allied therapists perform or that are carried out by the patients themselves. By providing an appropriate treatment plan, PM&R physicians help patients stay as active as possible at any age. Their broad medical expertise allows them to treat disabling conditions throughout a person's lifetime.

### Why see a PM&R physician?

A PM&R physician will thoroughly assess your condition, needs, and expectations and rule out any serious medical illnesses to develop a treatment plan. By understanding your condition and goals, you and your PM&R physician can develop a treatment plan suited to your unique needs.

### How do I find a PM&R physician near me?

Visit [www.aapmr.org/findapmrphysician](http://www.aapmr.org/findapmrphysician) or contact your primary care physician for a referral.