

**Editorial**  
**What is Spina Bifida?**  
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I have been asked as to why I object to the term “**Spina Bifida**”. I will attempt to explain. The term was introduced by Nicolas Tulpius in 1463 as a result of the autopsy examinations of babies that died with an open form of Spina Bifida Aperta. meaning obvious spinous processes in two parts; normally the fetal tissues that are precursors to bone and muscle (mesoderm) and its covering, ectoderm, rise up as folds on each side of what becomes the spinal cord and fuse in the middle. This folding and fusion leaves primitive ectoderm that becomes the nervous system and spinal cord encased in an inner layer of mesoderm that fuses to become the bony arches that surround the spinal cord and its accompanying muscle. The resulting outer layer of primitive ectoderm becomes skin. If, in this form, the process of folding and fusion in the mid line fails, there remains an area of bone in two parts (Spina Bifida) and a failure of skin to cover the intervening space [Aperta].). Today this type lesion is called Myelomeningocele (Myelo {nerves} menigo {membranes that cover the spinal cord and brain}cele{sac}) or Meningomyelocele. Since the time of Tulpius, a number of scientific advances have demonstrated that the term, “Spina Bifida”, is insufficient to describe the many normal and other types of “Spina Bifida”.

The most benign, or condition with no symptoms, involves the separation of the boney portions of the usually closed spinal vertebral arch not being closed and, hence “spine in two parts = spina bifida”. One in every 7 persons **without a family history** of an “open spinal vertebral defect – “Spina Bifida Aperta” has x-ray evidence of posterior vertebral spine failure to close. Nicholas Tulpius, of course, did not know this since we did not learn of this frequency of this isolated boney abnormality without symptoms until 500 years after his death when x-rays were introduced.

Why is “without a family history” in bold? Simply because **some** families have more than one relative with Spina Bifida Aperta and their immediate relatives will have both an increased frequency of the failure of the posterior vertebral spinous processes to fuse with out symptoms as well as their fsmily’s type of Spina Bifida Aperts.

Is there a way to determine this? Yes! Ordinarily the spine is comprised of 8 cervical (neck) vertebrae, 12 or 13 thoracic vertebrae (vertebrae with ribs), 4 or 5 lumbar vertebrae (without ribs and just above the pelvis) and the 5 or 4 sacral vertebrae that are fused into one bone, the sacrum, that is part of the pelvis separated by the sacroiliac joints. On average, there are 9 vertebrae below the ribs. Hence, if one has 4 lumbar then there will be 5 sacral and if 5 lumbar then 4 sacral. Persons who will later have “normal” x-rays have no x-ray evidence of posterior vertebral arch of the 4<sup>th</sup> or 5<sup>th</sup> lumbar arch fusion until women, on average, are 18 years of age and men by 21 years of age. Until that age the area of fusion is cartilage and does not have sufficient calcium to be visible by x-ray. Families with multiple cases of Myelomeningocele will have relatives with posterior vertebral arches open at above the lumbar 4 and below the 1<sup>st</sup> sacral vertebra more frequently than members of families without a family member with a Myelomeningocele.

How long has this phenomenon been known? Not until the discovery of x-ray. However, Paleolithic finds have demonstrated bony evidence of these abnormal “spina bifida” amongst Northern European skeletons dating to 3-400 years BC, Spanish skeletons dating to 10,000 BC, Egyptians to 3000 BC and USA natives, Modoc Indians, to 2000 BC as well as Eskimos of the North American Continent of this latter era. Were these Paleolithic finds representative of Myelomeningocele, (Spina Bifida Aperta)? The only evidence I know is the Egyptian hieroglyphics that portray babies with anencephaly or encephalocele (brain anomalies) amongst holy objects along with hibiscus and monkeys.

What is the connection between these last two brain abnormalities and “Spina Bifida”? A number of family and epidemiologic studies have identified these two abnormalities of the central nervous system (brain) and Myelomeningocele. These lesions are more common in the families of patients with Myelomeningocele and are the brain equivalents of Myelomeningocele with brain protruding into the sac (cele) or absent with no sac (anen {no} cephalo {head}). They are collectively called “neural tube defects”.

Why is such a relationship of importance? These two severe brain abnormalities are fatal shortly after birth and are reduced in family recurrence and the general population occurrence of both these brain anomalies as well as Myelomeningocele by administration of large (100 x the daily recommended dose of Folic Acid – 4 grams by mouth daily beginning three months prior to conception by the mother).

What about the other types of “Spina Bifida” that are neither Myelomeningocele nor “normal”? They are the so called “**Spina Bifida Occulta**” lesions. The most important reason is that they do not, in general, involve the same embryologic process of development. They do not include abnormalities of the brain. They arise as an abnormality of the second process whereby the lower spine and lower body nerves develop. The Myelomeningocele (Spina Bifida Aperta) lesions cause not only abnormalities of the function of lower body nerves but, more importantly, cause malformations of the brain that range from fatal in infancy as described in the three previous paragraphs to interference with mental function as children and adults.

These **Spina Bifida Occulta** lesions are usually skin covered and not associated with hydrocephalus. Eighty to 90 percent of affected persons have the ability to ambulate without aids and have normal intellect. Their medical diagnoses are: **Meningocele** (skin covered fluid filled sac of spinal cord coverings and spinal fluid but no nerves), **Myelocystocele** (the same as Meningocele but with several fluid filled sacs that are not connected to one another or the central nervous system), **Caudal Regression Syndrome** (absence of the development of the nerves, muscle, bones and tissues of the lumbar spine and/or sacrum, with or without fat tissue in the spinal canal surrounding the nerves) and, the most common of these “**Spina Bifida Occulta**” lesions, **Lipomyelomeningocele**. These lipomyelomeningocele lesions are both the most difficult to diagnose in some cases and the cause of recurrent episodes of loss of lower body nerve function loss that can be prevented by medical intervention. Fortunately, this loss of function can be detected by careful follow up examinations by experienced health professionals and prevented by surgical intervention. This loss of function is a sign of “spinal cord tethering” and is common to all forms of “Spina Bifida”.

Other Lipomyelomeningoceles can present at birth as a huge protruding, skin covered lesion. Between the barely perceptible Lipomyelomeningoceles and the grossly obvious is a continuum of gradations of presentation starting from a dimple above the gluteal cleft but off to one side of the midline or abnormal skin pigment.

These **Spina Bifida Occulta** lesions are not known to be prevented by excessive doses of folic acid, rarely are associated with hydrocephalus, are associated with lesser degrees of paralysis, obesity and intellectual impairments such as learning disorders, are much less frequently familial in occurrence and carry a much better prognosis for future adjustment as a normal person than the Myelomeningocele lesions.

These are the reasons, as your medical advisor, I state you should know your diagnosis. It is not sufficient to state that you have “Spina Bifida”. The only way you can be sure of your diagnosis is to obtain a report of your own, or your child’s original operative report and pathologic findings. This information is important for you, your child and the entire family. Medical recommendations are directly related to the original diagnosis as to the type of “Spina Bifida”.