Core Messages

- Spinal cord malformations (= spinal dysraphisms) are usually diagnosed at birth or early infancy (open spinal dysraphism, closed spinal dysraphisms with a back mass) but are sometimes not discovered before adulthood.
- Spinal cord malformations arise from defects occurring in the embryological stages of gastrulation (weeks 2–3), neurulation (weeks 3–6) and caudal regression.
- The term “spina bifida” merely refers to a defective fusion of posterior spinal bony elements but is still incorrectly used to refer to spinal dysraphism in general.
- “Tethered spinal cord” is a broadly used umbrella term for numerous spinal cord abnormalities, such as lipomyelomeningocele, previously operated on myelomeningoceles, or thickened filum terminale, which tether (fasten, fix) the spinal cord in the spinal canal.
- Tethered cord syndrome is a stretch-induced functional disorder of the spinal cord worsened by daily, repeated mechanical stretching, and distortion may even occur in patients who have the conus at normal level.
- Patients with spinal cord malformation are either diagnosed at birth or present later because of unexplained pain, neurological deficits, unclear recurrent urologic infections, cutaneous markers or orthopedic deformities.
- MRI is the imaging modality of choice and has increased the number of tethered spinal cord diagnoses.
- Prenatal treatment encompasses prophylactic folic acid substitution and intrauterine surgery.
- Open spinal dysraphism is best surgically treated postpartum to untether the spinal cord, prevent infections, repair the dural/cutaneous defect, and restore normal anatomy as far as possible.
- Closed spinal dysraphism with tethered spinal cord warrants early untethering, when minimum or mild symptoms are detected.
- Surgery after development of the deficits only stops progression, but symptoms may even further progress after detethering.
- Individuals with spinal malformations need both lifelong surgical and medical management, which should be provided by a multidisciplinary team.

Epidemiology

Spine and spinal cord malformations are often collectively summarized under the term of spinal dysraphisms [39]. This term was first employed by Lichtenstein (1940) [36]. Open spinal dysraphism is a common congenital midline defect of the nervous system and has been historically reported in 2–4/1000 live births [14]. However, the true incidence of spinal dysraphism is not well studied. Myelomeningocele accounts for the vast majority of open spinal dysraphisms (98.8%) [32, 39].

Myelomeningocele occurs in 0.6 patients per 1 000 live births, and females are affected slightly more often than males (by a ratio of 1.3 to 3), with the first-born usually affected [5, 39]. Myelocele is a rare malformation and represents only 1.2% of all open spinal dysraphisms [39]. The most common locations for these malformations are, in decreasing frequency, lumbosacral, thoracolumbar and...
Case Introduction

A 17-year-old patient presented with progressive tethered cord syndrome with worsening of hand functions and some leg weakness and increasing spasticity. Postnatally he had had a cervical myelomeningocele and had had only “cosmetic” closure after the birth. The MRI showed a widened spinal canal at C6–C1 (a, c), cord tethering dorsally at C6–7 and dorsal limited myeloschisis. It is possible to see the hypotrophic right hand (b). This clinical worsening recovered after an intradural exploration and dissection of the stalk placode.

Spina bifida is present in 90–100% of patients with tethered cord. The incidence of myelomeningocele varies from country to country and from one geographical region to another [20]. Since the early 1980s, estimation of the prevalence of open spinal dysraphism in many industrialized countries has been decreased by folic acid administration to pregnant women and the availability of prenatal diagnosis and elective termination [20, 29, 48]. Patients with open spinal dysraphism almost always have associated Chiari II malformation. There are also reports in the medical literature of an association between closed spinal dysraphisms and Chiari II [41].

Spina bifida occulta occurs in approximately 17–30% of the total population and is present in 90–100% of patients with tethered cord [35, 61]. The dermal sinus is a common abnormality and accounts for 23.7% of all closed spinal dysraphisms. Overall, caudal regression syndrome is not uncommon, accounting for...