Differentiating Tethered Cord Syndrome, Neuro-Cranio-Vertebral Syndrome, and Filum Disease

Author: Dr. Miguel B. Royo Salvador, Director of the Institut Chiari & Siringomielia & Escoliosis de Barcelona

Having encountered frequent inquiries about the similarities and differences between neuro-cranio-vertebral syndrome, filum disease and tethered cord syndrome, I would like to take the opportunity offered by *WSCTF* and the *Rare Disease Report* to address the definitions of the different conditions and how they relate to each other.

Tethered Cord Syndrome (TCS)

Tethered cord syndrome (TCS) consists of an abnormal tethering of the spine due to a spinal and spinal cord malformation (meningocele or myelomeningocele, spina bifida) that, due to traction, can cause a descent of the cerebellar tonsils, syringomyelia and scoliosis. It is characterized by neurological manifestations of sensory and motor deficiencies, predominantly in the lower extremities, by orthopedic deformities in feet and skin stigmata. The external visibility of the latter upon simple inspection makes the TCS different from ***tethered spinal cord*** **(TSC).**

Figure 1.- Tethered Cord Syndrome with myelomeningocele in a fetus where the spinal cord (yellow in the image) exerts an important traction on the encephalon which in turn produces a descent of the cerebellar tonsils.

TCS is an affectation of little frequency; it is either diagnosed upon birth or late in the adult and can be mistaken with other spine and spinal cord anomalies. The symptomatology induced by the spinal cord traction was initially suggested in patients with myelomeningocele by Fuchs in 1910 (1) and Lichtenstein in 1940 (2).

The term ***filum terminale syndrome*** or ***cord-traction syndrome*** was first described by McKenzie KG in 1949, defined by Garceau in 1953 (3), and named ***tght filum terminale*** by Jones and Love in 1956 (4*)*. It is a neurological disease with symptoms and signs of a suffering spinal cord accompanied by idiopathic scoliosis without other associated neurovertebral malformations and is treated with the ***sectioning of the filum terminale*** (SFT), the ligament that connects the end of the spinal cord with the spine at the coccyx through an posterior opening of the back. Garceau applied this in three patients with good results.

The term ***tethered spinal cord*** was first used in 1976 by Hoffman and colleagues (5); who published the improvement of neurological deficit after the SFT through lumbar laminectomy in 31 children with an elongated spinal cord by the traction from an abnormal filum terminale or another clearly abnormal structure that is not visible externally (Figure 2). The treatment for these conditions tends to be the release of the tethered spinal cord of the myelo-meningo-vertebral malformation or in less severe cases, the sectioning of the abnormal filum terminale, almost always with the lumbar laminectomy as the surgical approach.

The incidence of these neuro-vertebral malformations is low, it represents 20-50% of spina bifida cases (spina bifida: lack of posterior closure of the spinal canal), and these in turn have an incidence of 1/2000 live births in France and 6/1000 in Anglo-Saxon countries. The incidence of treatment indication is even lower (10-20%), around 70,000 cases per annum worldwide.

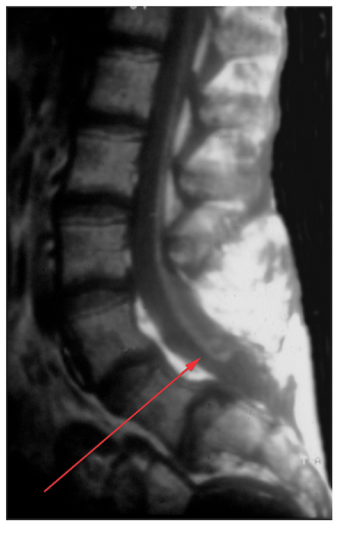

Figure 2.- Spinal cord tethered to the posterior wall dura mater meninges of the spinal canal at the height of L4 and L5 with an absence of the spinous process as well of the bony posterior wall of the spinal canal from L4 to the coccyx.

Neuro-Cranio-Vertebral Syndrome (NCVS)

Neuro-cranio-vertebral syndrome (NCVS), described together with Filum Disease in 1996 and named in 2009 by Royo-Salvador MB (6,7), is a consequence of the abnormal traction exerted by the filum terminale and expresses itself with primary manifestations of the entire nervous system (brain, spinal cord and nerve roots, skull and spine} and secondary manifestations of the entire organism such as ocular, oropharyngeal, circulatory, urinary, digestive, hormonal consequences.

The abnormal traction of the spinal cord and of the entire nervous system can be due to a congenital deformation of the spinal canal or vertebral alterations (fusions, hemi-vertebrae, or others); to traumatic causes such as fractures, compressions, disc extrusions or others; postsurgical causes; infectious causes; tumorous causes.

That is to say that all those causes that produce an asynchrony in the dimensions of the spinal canal and the neuroaxis, with a tense filum terminale as a result. The most frequent NCVS form is the congenital one, called the Filum Disease.

Filum Disease (FD)

Filum disease (FD) is the congenital form of NCVS or when the cause of the cord traction can be due to a congenital anomaly of an apparently normal filum terminale, without that any anomalies of the ligament or of other conventional neurovertebral malformations can be seen on the neuroimaging.

FD manifests clinically at its highest intensity inthe form of the Arnold-Chiari I syndrome, idiopathic syringomyelia and idiopathic scoliosis, basilar invagination, platybasia, odontoid retroflexion, brainstem kinking, disc disease, cerebral multiinfarctions, sudden death, nocturnal enuresis, sphincter alterations, hormone dysfunctions and others.

FD is a condition where the patient in the majority of cases is not aware that he/she is suffering from it. The symptomatology tends to be faint, diffused, and latent, and together with a manifest but unperceivable physical deterioration, it does not allow, neither the doctor nor the patient, to consider it as a disease. For example, an adult has a grip strength of 25 to 70 kilos in the hands, the patient does not become aware of his or her limitation until it is less than 8 kilos. The majority of patients that we diagnose with the FD have less than half of the corresponding grip strength in their hands; they would however never know this if they were not examined with dynamometer tests (8).

The suggested treatment for FD is the application of the Filum System® with 12 protocols for the diagnosis, treatment and follow up (8). Of the 12 protocols, the 4th is concerned with the sectioning of the filum terminale (SFT) at sacrum level with a minimally invasive technique (9). The SFT procedure is indicated once the FD diagnosis has been established and there are signs of neurological deterioration, or it has a progressive character.

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Images provided by Dr Royo Salvador