Clinics in Surgery

Case Study
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Bow-Hunter Anomaly in a Patient with Ehlers Danlos Syndrome

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Presentation

A-20-year-old-girl with a history of ligamentous hyper-laxity and joint hypermobility compatible with 9 out of 9 points in correlation with Beighton scoring test with the diagnosis of Ehlers Danlos syndrome -hypermobile type (hEDS) [1,2]. Since her early childhood she complained of persistent headache (migraine-like), joint pain (early osteoarthritis). In addition she suffered from frequent bouts of syncope accompanied with easy tiredness with drastic episodes of leg weakness. At the age of nine years she started to develop episodes of sudden involuntary head rattling movements associated with sudden rolling of both eyes. These episodes occurred in response to sudden forceful head turning/rotation. Sagittal MRI showed tip of the odontoid is more cephalad and protrudes into the spinal cord, resulting in partial anterior compression of the vertebro-basilar artery (arrow). This sort of compression falls within the group of compression syndromes of the vertebral artery that occur at the cranio-cervical junction. This compression leads to dynamic vertebral artery occlusion where the vessel courses around the atlas and the axis-the so-called V3 segment. This in turn leads to in insufficient collateral flow to the posterior fossa. The clinical picture is a vertebra-basilar insufficiency disorder. Clinical manifestations are broad ranging from vertigo, syncope and possibly to posterior fossa stroke. The condition is termed as “bow hunter's” syndrome (Figure 1). 3D reformatted sagittal CT scan showed atlanto-axial instability causing effectively impingement of the brain stem. The patient´s natural history of the disease and her recent neurological deficits are highly likely in favor of basilar invagination/impression secondary to associated laxity of the ligaments. Basilar invagination [3] is a life threatening deformity when the bulging of odontoid is more than 3 mm above Chamberlain´s line. This patient showed abnormal bulging of the dens more than 15 mm into the brain stem (Figure 2).

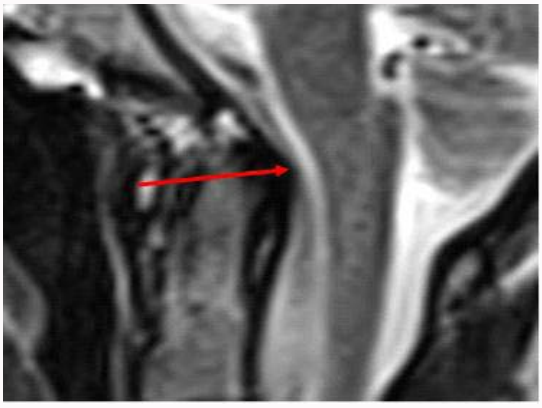

Figure 1: A- 20-year-old-girl, developed episodes of sudden involuntary head rattling movements associated with sudden rolling of both eyes since she was 9-year-old. Sagittal Craniocervical MRI showed: Tip of the odontoid is more cephalad and protrudes into the foramen magnum, resulting in partial anterior compression of the vertebro-basilar artery (arrow). This sort of compression falls within the group of compression syndromes of the vertebral artery that occur at the craniocervical junction. This compression leads to dynamic vertebral artery occlusion where the vessel courses around the atlas and the axis-the so-called V3 segment. This in turn leads to in insufficient collateral flow to the posterior fossa. The clinical picture is a vertebra-basilar insufficiency disorder. Clinical manifestations are broad ranging from vertigo, syncope and possibly to posterior fossa stroke. The condition is termed as ‘bow hunter's syndrome.

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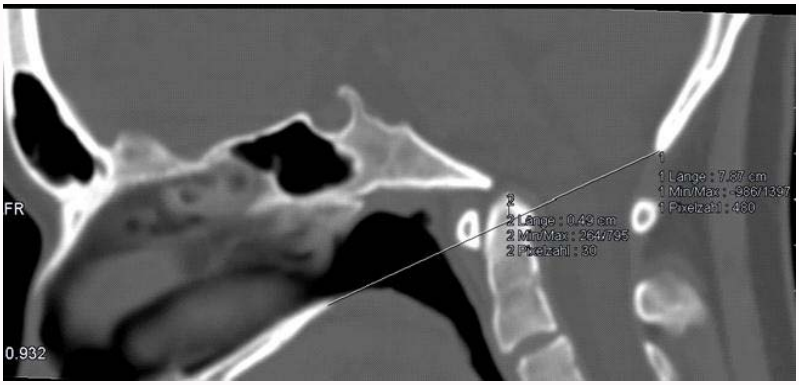

Figure 2: 3D sagittal CT scan showed Chamberlain´s line, which is joining the back of hard palate with the opisthion (Chamberlain´s line). This patient showed basilar invagination in connection with atlanto-axial instability.

Discussion

Bow Hunter's syndrome classically refers to mechanical occlusion of the vertebral artery during head rotation secondary to numerous etiologies including congenital or acquired bony malformations or neck muscle hypertrophy [4]. Patients with connective tissue disorders are believed to be at a higher risk for cerebrovascular malformations, dissections and basilar invagination [5]. Our current patient needs distinctive management to overcome the unpredictable morbid or mortal complications. Firstly; the generalized ligamentous hyperlaxity can lead to laxity of the transverse ligament with subsequent C1-2 instability. Secondly; Basilar invagination [3], which is a serious craniocervical junction anomaly represented by a bulging of C2 into the skull-base (brain stem and spinal cord) that can result in severe neurological impairment. The suggested surgical intervention for this patient is fusion of C1-2. The diagnosis of hypermobile Ehlers-Danlos syndrome is mostly based on clinical and radiological phenotypic characterization. This patient underwent whole exome sequencing and no genetic mutation has been encountered.

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