PLATYBASIA AND KLIPPEL FEIL-SYNDROME: CASE REPORT

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SUMMARY – A case is presented of a 35-year-old woman diagnosed with platybasia associated with Klippel-Feil syndrome type I. She was admitted to University Department of Neurology for clinical examination because of walking difficulties, dizziness, and intermittent vision disturbances. Neurological examination revealed a predominance of cerebellar symptomatology. Relevant diagnostic work-up included craniogram, cervical spine x-ray, computed tomography (CT) of the brain and craniocervical junction, magnetic resonance imaging of the brain, electroencephalography, ophthalmologic examination, urinary tract ultrasonography, laboratory tests, and psychological testing. CT of the craniocervical junction showed platybasia, congenital fusion of the second and third cervical vertebrae, and basilar invagination of dens axis. Platybasia is leveling of the angle between the floor of the anterior cranial fossa and posterior cranial fossa in the area of sella turcica, which is normally at 115-140 degrees. Basilar impression or invagination is moving up of the basis of the occiput and occipital condyles into the cranium, which means that the borders of the foramen magnum, condyles and adjacent bone are invaginated into the posterior fossa. Klippel-Feil syndrome type II is massive fusion of two of seven cervical vertebrae associated with short neck and low hair line.

Key words: *Platybasia; Klippel-Feil syndrome; Case report; Bosnia and Herzegovina*

Introduction

Platybasia is flattening of the angle between the floor of the anterior and superior cranial fossae in the area of sella turcica, which normally ranges between 115 and 140 degrees. A less severe degree of such anomaly has no clinical significance. Basilar impression or invagination represents an upward shift of the occipital base and its condyles into cranial fossa, which means that the edges of the foramen magnum, condyles and adjacent bone are invaginated upwards into the posterior cranial fossa. The etiology of primary basilar impression is unknown. Genetic factors probably play a role. Secondary impression is a consequence of bone malacia (Paget's disease, osteomalacia, hyperthyroidism, osteogenesis imperfecta)[1]. Clinical signs usually appear between age 10 and 40 years, with painful neck movements, vertigo, hand and leg palsy, and in many patients palsy of cranial nerves, especially of the accessory nerve, with dysarthria and dysphagia. Leg palsy can be accompanied by functional disorder of the sphincter muscle. In some cases, there are signs of increased intracranial pressure together with signs of insufficient vertebrobasilar circulation. Such defects are often combined with Arnold-Chiari's malformation. Treatment is directed toward decreasing compression in the posterior cranial fossa, by employing surgical methods[2,3].

Klippel-Feil syndrome includes massive fusion of at least 2-7 cervical vertebrae, accompanied by short neck and lowered hair line. Klippel and Feil published their first work about this syndrome in 1912. The etiology of the disease is not well known. What is known is that there is a disorder of the cervical segmentation in the 3rd and 8th week of gestation.

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According to literature data incidence of the disorder is around 0.71% to 0.2 cases *per* 1000 population[4,5]. In 50% of cases, clinical signs include short neck, lowered hair line, and limited movements of the neck. The body is capable of compensating for the defect partially, by hypermobility of the movable segments. Twenty percent of patients have facial asymmetry and torticollis. Ptosis, horizontal nystagmus and cleft palate are common findings. Syndactyly or polydactyly can be seen as well. Numerous anomalies can be associated with this syndrome, most commonly scoliosis, kyphosis, genitourinary anomalies, cardiopulmonary anomalies and craniocervical anomalies (such as basilar invagination, cervical stenosis, syringomyelia, Chiari I malformation and intracranial tumors). Feil divided this syndrome into 3 categories: type I presenting as massive fusion of cervical vertebrae; type II presenting fusion of 2 cervical vertebrae; and type III presenting anomalies of thoracic and lumbar spine associated with type III anomalies[6]. Symptoms vary and include symptoms due to increased mobility of the free segments with possible development of osteoarthritis of these segments. Symptoms due to mechanical irritation of the roots, together with neurological symptoms due to irritation and compression of spinal cord, can develop as well. The worst neurological sequel can be death or quadriplegia, caused by minor trauma in high-risk patients[7]. A smaller percentage of patients have minimal symptoms[1]. These patients can lead normal life provided that they follow given recommendations (avoiding head trauma, i.e. contact sports, etc.)[1,8]. Conservative therapy includes analgesics, nonsteroidal antiinflammatory drugs, collar, or careful traction. In severe cases, therapy is surgical with postoperative physical treatment[9,10].

Case Report

A 35-year-old female patient was referred for clinical examination by her family physician due to problems with walking, vertigo and instability persisting for 3-4 years. Family history showed that both parents had died (father from heart attack and mother from kidney tumor). Her brother committed suicide by poisoning. Her father's sister had suffered a stroke and her uncle died from cancer. Personal history revealed normal early psychosomatic development; good elementary school performance. The patient reported no serious disease (only flu); menarche at age 15, regular menstruation. In 1990, she fell and injured her head, but did not lose consciousness.

Current symptoms: fatigue and weakness persisting for the 3-4 years, vision problems (reduced sight on both eyes, with occasional diplopia), vertigo, instability while walking, staggering and falling, all these more pronounced in the past winter; backache with propagation to both legs (more to the right one). Findings at admission: conscious, oriented, communicable, eupneic at rest, normocardiac, physical examination of her heart and lungs within the normal limits. Blood pressure 110/70, pulse 74/min. Abdomen at the same level as her chest cavity, soft, not sensitive to palpation. No edema on her extremities.

Neurological findings: her head had normal configuration; somewhat narrower left rima oculi; bilateral horizontal nystagmus when looking aside; limited movement and rotation of the neck. Upper extremities in antigravity position: left hand slightly in pronation. Myotatic reflexes provoked bilaterally, somewhat weaker on her left side. Lower extremities: loss of motor strength; she could lift her legs in antigravity position and shortly keep them in such position; bilaterally, reflexes hardly provoked; walking unstable, ataxic, "tandem" walking cannot be performed. In Romberg's test, there was a tendency toward falling ahead; coordination test with open eyes insecure, and with closed eyes she missed. She claimed good sphincter control. Laboratory findings: blood tests, proteinogram, min-

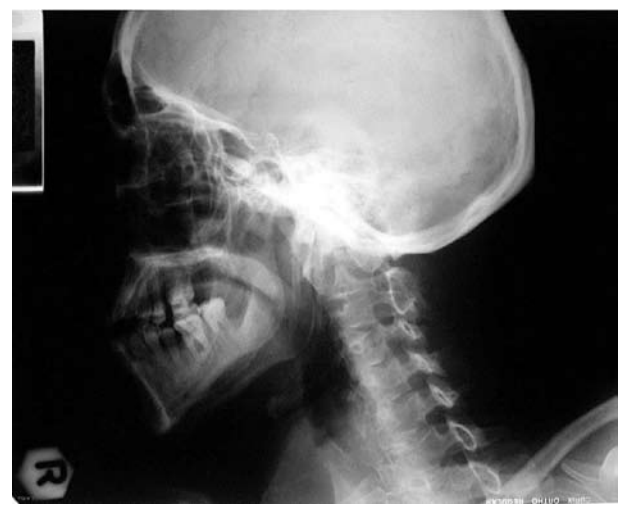

Fig. 1. Cranium x-ray.

eralogram and transaminases within the normal limits. Urine results pathologic. Urine cultures remained sterile. Ophthalmologic examination normal. Lung and heart x-ray within the normal limits. Cranium x-ray is shown in Figure 1.

Computed tomography (CT) of the cranium: infratentorially, on most caudal scanning of the medulla oblongata region, there is a hypoechogenic zone of bone density, which belongs to dens. Density of brain tissue in the supratentorial area is normal; ventricular system is in midline and of normal width. Sulci at the brain convexity are of normal depth (Fig. 2).

CT of the cervical spine: during previously performed CT of the brain, a significant prominence of dens axis (cranially) was noticed, and additional examinations were recommended. On additional examination of the cervical spine, using magnetic reconstructions, coronally and transversally (with bone and soft tissue windows), there was a bone blockage between dens axis, corpus of the dens and third cervical vertebra.

Dens axis invaginated cranially by about 2 cm (basilar invagination, platybasia). Anterior arch of atlas is in connection with occipital foramen, so the distance between dens and anterior arch of atlas is wider. The posterior part of atlas arches is also in connection with occipital bone. On more distal parts, other cervical vertebrae have normal continuity, appear normal

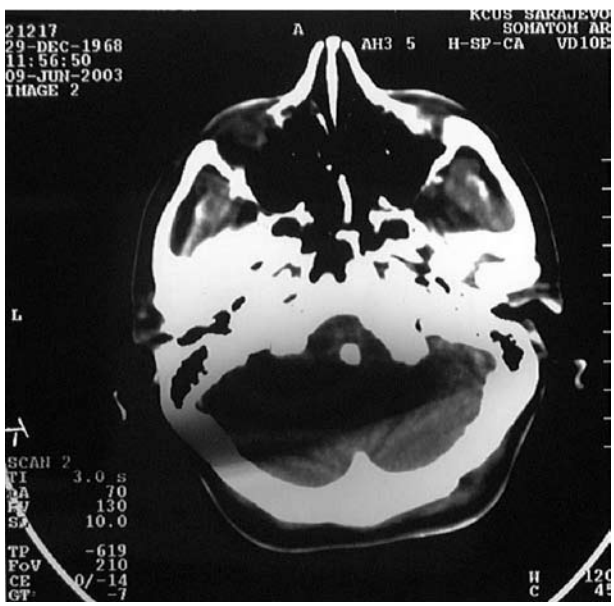

Fig. 2. Computed tomography scan of the cervical spine.

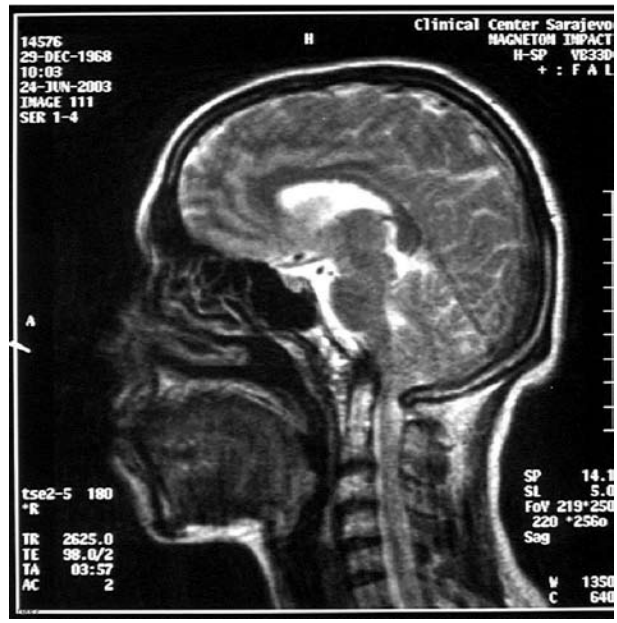

Fig. 3. Magnetic resonance image of the cranium.

and with normal density. On reconstructive scanning, there were no signs of reduction of the vertebral canal width.

Magnetic resonance imaging (MRI) of the cranium and craniocervical junction: 1.0 Tesla: axially T1 SE, T2 SE and PD, sagittally T1 SE, T2 SE and

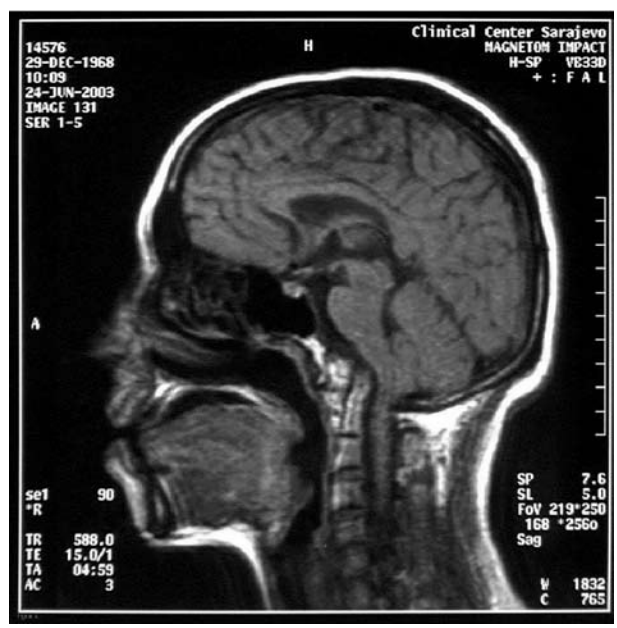

Fig. 4. Magnetic resonance image of the craniocervical junction.

PD. Oblique MRI of the cranium: signal intensity within the normal limits both infratentorially and supratentorially. The forth, third and lateral ventricles are in normal position, of normal width and signal intensity. Sulci on the convexity of the brain together with basal cisterns have adequate depth. When MRI of the craniocervical junction was performed up to C6 segment, it was noted that dens had changed signal intensity (type of degenerative changes) and was in retroposition; it was obvious that there was flattening of the occipital bone, which led to narrowing of the vertebral canal in the craniocervical junction (down to 1 cm). On the scans of medullary canal, there was no zone with changed signal intensity (Figs. 3 and 4).

Psychology test (conclusion): based on the test and behavioral results performed in the study, there are indicators of cognitive dysfunctions (primary and global) with additional limitations due to the problems in coordination of movements.

Neurosurgeon's opinion: surgical treatment is recommended.

Discussion

In practice, platybasia and Klippel-Feil syndrome are very rare disorders, when associated even rarer. That is why we considered it useful to present the case of a female patient suffering from both disorders. After first neurological examination of the patient, differential diagnosis included tumors of the anterior cranial fossa and multiple sclerosis, and both were excluded by employing relevant technique (CT of the cranium and cervical spine). After establishing the diagnosis, we consulted neurosurgeons who recommended surgical treatment, which has been refused by the patient so far. According to the literature, surgical treatment achieves good recovery of neurological deficit and improves the quality of life. Goncalves da Silva and Goncalves da Silva report on 53 cases of basilar impression, treated surgically either as a unique entity or in association with Chiari's malformation and syringomyelia[11]. Urculo-Bareno *et al.* report on a case of a 65-year-old female patient whose neurological status improved significantly after surgical treatment of basilar impression[8]. Ghosh *et al.* report on platybasia and basilar invagination in osteogenesis imperfecta[12]. Turgut has described Klippel-Feil syndrome in association with posterior fossa dermoid tumor[6].

Conclusion

Klippel-Feil syndrome is a very rare disorder of congenital etiology. In our patient, platybasia or basilar impression was associated with Klippel-Feil syndrome. Individuals with Klippel-Feil syndrome should be warned of the potential for sustaining a neurologic deficit after minor trauma. The disorder was diagnosed by employing relevant techniques. Because of the anomaly severity and presence of neurological deficit, surgical treatment is considered appropriate.

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Sažetak

PLATIBAZIJA I KLIPPEL-FEILOV SINDROM: PRIKAZ SLUČAJA

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Prikazan je slučaj 35-godišnje žene s dijagnozom platibazije povezane s Klippel-Feilovim sindromom tip I. Ona je primljena na Kliniku za neurologiju zbog poteškoća u kretanju, vrtoglavica i povremenih smetnji vida. Neurološki pregled otkrio je dominantnu cerebelarnu simptomatologiju. Provedene su relevantne dijagnostičke pretrage: kraniogram, rendgenski snimak vratne kralježnice, kompjutorska tomografija (CT) mozga i kraniocervikalnog spoja, magnetska rezonanca mozga, elektroencefalografija, oftalmološki pregled, pregled mokraćnog sustava ultrazvukom, laboratorijski testovi i psihološko testiranje. CT kraniocervikalnog spoja pokazao je platibaziju, prirođenu fuziju drugog i trećeg vratnog kralješka i bazilarnu invaginaciju. Platibazija je izravnavanje kuta između poda prednje lubanje i stražnje lubanjske jame u području sedlaste jame, koji je inače na 115-140 stupnjeva. Klippel-Feilov sindrom tip II. je masivan spoj dvaju od sedam vratnih kralježaka povezan s kratkim vratom.

Ključne riječi: *Platibazija; Klippel-Feilov sindrom; Prikaz slučaja; Bosna i Hercegovina*