Klippel-Feil syndrome (dystrophia brevicolis) was described for the first time in 1912, by Maurice Klippel and Andre Feil. It is known for the triad of symptoms such as short neck, limitation of neck movements and low posterior hairline. It may be associated with some somatic diseases and defects such as scoliosis, anomalies in urogenital system, Spergel sequence, congenital heart defects, deafness, facial asymmetry, flat neck, mental deficiency, synkinesis or mirror movements. Klippel-Feil syndrome is often manifested in oral cavity with cleft palate, limitation of mouth opening, hipodontia, oligodontia, micrognathia or prisms. The goal of this study was to systemize the knowledge on Klippel-Feil syndrome (Dent. Med. Probl. 2007, 44, 4, 491–494).

Key words: Klippel-Feil syndrome, congenital short neck, wry neck, torticollis, cleft palate.
fused occurs between the third and the seventh week of embryonic development) [1]. Authors claim that the reason for the Klippel-Feil syndrome may be the mutation of either p12 or q34 loci of chromosome 2 [4]. Others observed this syndrome occurring in the whole family. In this case, the disease was related to the paracentric inversion on chromosome 8 (q22.2q23.3) [5]. Usually, the mutation of gene appears de novo. It is rarely inherited [6]. There is a thesis, that fusion of vertebrae is likely related to disturbance of PAX-1 gene expression, responsible for developing of vertebral column [7]. It is thought that types I (fusion of many cervical vertebrae) and III (characteristic for both cervical and lower thoracic or lumbar fusion) are inherited autosomal recessively. The type II (fusion of two or three vertebrae) is rather an autosomal dominant [1, 8].

The literature reports on several severe abnormalities of vertebra column, which are: the changes of number of vertebrae (8 cervical, 11 thoracic, 6 lumbar, 5 sacral) and their segmentation and morphology [6]. Changes of number and morphology of vertebrae as well as fusion of some of them results in an asymmetry of posture [9].

The syndrome, beside its typical features, may also have some Orofacial manifestations, such as cleft palate, hipodontia or oligodontia, micrognathia and prismus [4, 10]. Sometimes the syndrome is associated to difficulties in opening mouth or limitation of mandible movements with a normal mouth opening [2, 3].

The aim of this study was to systemize the knowledge on Klippel-Feil syndrome.

**Skeletal Malformations**

Malformations of skeleton are one of the typical symptoms in Klippel-Feil syndrome. These may be mild cosmetic deformities or even severe disabilities [2]. Number of vertebrae may be either reduced or increased [6, 8]. Beside the fusion of vertebrae, also scoliosis is an often skeleton’s malformation and it occurs at 60% of cases of Klippel-Feil syndrome. A typical malformation of this disease is so called Spiegel’s deformity, occurring at 35% of cases [11]. Spiegel’s deformity is considered as an elevation or malposition of the scapula which results in various degrees of shoulders asymmetry and dysplastic manifestations that cause limitations in motion and function of shoulder girdle [9]. Rib defects were also observed. This is usually so called cervical rib, widely seen together with Klippel-Feil syndrome [1, 8]. Some cases report on moderate degree of kyphosis [2]. Rarely, other anomalies of craniovertebral spine were observed. Those were: “fixed” atlantoaxial dislocation, assimilation of the atlas, malformations of the occipital bone, fusion of other than vertebrae cervical bodies [12]. Some authors described also a case of absence of radius and first metacarpal bone as well as elongated-curved ulna and triphalangeal thumb [11]. The others reported on syndactilia or polydactyly [8, 13]. Literature also reports on the possibility of coexisting of Klippel-Feil syndrome with Poland anomaly, which consist of unilateral aplasia of the chest wall muscles and ipsilateral anomalies of upper extremity [14].

The literature reports on several severe abnormalities of vertebra column, which are: the changes of number of vertebrae (8 cervical, 11 thoracic, 6 lumbar, 5 sacral) and their segmentation and morphology. A hemivertebrae (C0) may be in this case fused to the occipital condyle as well as with the foramen magnum. Atlas and axis are usually deformed and hypoplastic. These malformations of spine as well as fusion of two or more vertebrae may correspond to an abnormal division of mesodermal somites and an inhibition or necrosis of the chondrogenic centres of vertebrae [6]. Changes of number and morphology of vertebrae as well as fusion of some of them results in an asymmetry of posture [9].

The malformations of spine lead not only to asymmetry of posture but also torticollis and facial asymmetry (also the orbital area) which create a typical aspect of the syndrome [10].

**Coexisting Somatic Diseases**

Beside the typical image of the Klippel-Feil syndrome, also some somatic disease may be present. The most common are: anomalies in urogenital system (65%), anomalies of cardiovascular system (15.35%), deafness or hearing lesion which usually refers to low tones (36%). Some common coexisting values were facial asymmetry and platybasia (20%), synkinesis or mirror movements (20%), anomalies of gastrointestinal tract (6%), congenital heart defects (4.2–14%) [9–11]. Delayed psychical and physical development was also observed in literature as coexisting with Klippel-Feil syndrome [7]. The delayed physical and physical development is not obligatory though [10]. The anomalies of urogenital system are various. Those may be renal agenesis, asymptomatic renalal cyst. The most common disorders of cardiovascular system are: aortic insufficiency, mitral valve prolapse, mitral insufficiency, aortic insufficiency and atrial septal defect [8].

Some neurological diseases such as paresis, parenthesiae, discrepancies in sensation, spasticity,
weakness, hyperflexia, quadriplegia or other disabili-
ties are possible [8, 15]. Mirror movements are also quite often seen in this syndrome [15]. Mirror movements refer to involuntary movements in a group of muscles or limb of one side of the body as a response to intentionally made move of the opposite side of the body (corresponding muscle group or limb) [16]. Usually mirror movements affect generally the hands, although may affect entire upper extremity or even legs. Mirror movements and cervicomedullary neuroschisis have a strong association to Klippel–Feil syndrome [17]. Also some developmental milestones such as standing and walking may be delayed [10]. There was a report of a case of a patient with epilepsy and some cases of hydrocephaly [8]. Some authors claim that the neuropathologic changes have a quite high prevalence in Klippel–Feil Syndrome [17]. The other though do not see any direct junction between neuropathological changes and dystrophia brevicolis [18].

Orofacial Manifestation

Klippel–Feil syndrome, beside the triad of symptoms (short neck, limitation of neck movements and low posterior hairline) is also known for facial asymmetry. Asymmetry in the orbital area, a deficient midface as well as ptosis and downward obliquity of the palpebral fissures are easily noticed. Absence of eyebrows was also reported [10]. Patients may exhibit a smaller lower third of the face [1]. Also hypoplasia of depressor angularis oris muscle was reported [3, 11]. Patients with Klippel–Feil syndrome often show mouth breathing [1]. Several authors report underdeveloped low-set ears coexisting with facial asymmetry [1, 5, 19]. The head is usually short, stenofacial [20].

Oral findings usually refer to clef palate (15% of cases), but may also include oligodontia, micrognathia, trismus and bruxism [4, 10]. Cleft may refer only to an uvula [3]. Mouth opening may be restricted and temporomandibular joints may be clicking. Cleft palate may be followed by nasal speech [10].

The roentgenograms may reveal asymmetry within the mandible. This may be the shortening of its branch and malformations of the articular tuberculum [3].

Intraorally, Klippel–Feil syndrome is known for late dentition [10]. Also cases of oligo- and hypodontia or tooth agenesis both in primary and permanent dentition were observed [3, 4, 10]. Some authors claim that no dental implications excluding caries were observed [1].

Some bite disorders were also described as following the syndrome. These may be: crowding of teeth, vertical open bite and deep palate [1]. There were also reports on: minimal overjet, deep bite, crossbite and Angle Class II Div I [10]. Therefore, usually, some orthodontic treatment is required.

Treatment

The diagnostics of Klippel–Feil syndrome should be based on the typical image of the disease with triad of symptoms: short neck, low posterior hairline, limitation of neck movements. The basic step of diagnostics is though a roentgenogram of the cervical vertebrae. The diagnosis should be followed with auricular, oculist and dental examination, but also should be followed with medical examination [3].

The treatment of Klippel–Feil syndrome used to be only a correction of some facial asymmetry. This usually were closure of cleft palate, correction of ears or nose, sometimes correcting the mandible due to its one-sided deficiency. The spine used to be unoperated due to difficulties in lying and intubating the patients [21]. Technological advances make it possible to operate those patients. The vertebral discectomy is followed by flexibility and motion of the patients body. The artificial cervical disc arthroplasty helps to maintain and preserve mobility and function of the cervical motion segments [22].

There is no scheme of dental treatment of the patients with Klippel–Feil syndrome. Childen with Klippel–Feil syndrome are patients in at high-risk caries category. Therefore some materials releasing fluoride to the oral cavity should be used at children [1]. For orthodontic treatment extraction of primary canines and Hawley appliance with coffin springs were delivered [10].

Both dental and medical treatment are difficult due to limited neck movements and limited mouth opening [10]. Some authors report that patients should not be intubated due to neck anomalies [1, 23]. The failure of intubation under direct laryngoscopy and loss of airway control is also possible. The insertion of laryngeal mask may solve this problem in life-threatening situation [12]. The arthroplasty or discectomy halps patients with Klippel–Feil syndrome in normal living, but the operation is very risky. That is why the indications for operative treatment of cervical diastematomyelia remains controversial [9].
References


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