

Klippel Feil syndrome: A case report and review of literature

Soumeya Fedala¹, Haddam Ali el Mahdi², Tadjine Aicha³, Ahmed Ali Leyla⁴
^{1,2,3,4}Department Of Endocrinology, Bab El Oued Hospital, Algiers

Abstract: Klippel Feil syndrome (SKF) is a rare congenital disease. It is characterized mainly by the incorrect union or fusion of two cervical vertebrae or more. This malformation is responsible for limitation of movement of the head and a significant risk of spinal cord injury. Other variable anomalies may be associated with increased morbidity. We report the case of a child who consulted us for short stature with abnormal sexual differentiation. The existence of short and palmed neck associated with congenital strabismus oriented diagnosis and has linked abnormalities present to Klippel Feil syndrome

Keywords: cervical vertebrae; short neck; radiography of the cervical spine; malformative anomalies

I. Introduction

Klippel Feil syndrome (SKF) is a rare congenital disease. It has a prevalence of 1 in 42,000 births. It is characterized mainly by the incorrect union or fusion of two cervical vertebrae or more. This malformation is responsible for limitation of movement of the head and a significant risk of spinal cord injury.¹ Other variables anomalies of a patient to another may be associated with increased morbidity.^{2,3} We report a case of SKF remembering the clinical and laboratory characteristics of the syndrome.

II. Observation

The child BS aged three years and ten months from a non-consanguineous marriage is hospitalized for exploration and therapeutic management of abnormality of sexual differentiation associated with short stature. The examination did not find any similar case in the family. Pregnancy and childbirth were without incident. The weight and height at birth were normal (2.8 kg and 50cm). Sexual ambiguity found at birth. However no exploration is undertaken to the present hospitalization.

Clinical examination on admission noted a good psychomotor development. The children had a significant short stature: Size -3.5 DS / M (Sempé) and - 2,5DS / TC, abnormal sexual development with bilateral cryptorchidism, hypo plastic scrotum welded but surmounted by a genital bud (0.5 cm) with a perineoscrotal hypospadias.

The somatic exam found multiple malformations anomalies: Asymmetry of the face, épicanthus, strabismus right convergent asymmetric upper lip, arched palate, short neck, limited range of motion of the neck, low hairline and hypo plastic pavilions ears (fig1).

The rest of the clinical examination did not find signs of adrenal hormone insufficiency, hypothyroidism or growth hormone delay.

The diagnosis is suggested by SKF clinical and confirmed by radiography of the cervical spine which revealed a fusion of C1 and C2 (fig 2). Supplementary examinations in search of other associated malformations revealed severe bilateral sensor neural hearing loss with the magnetic resonance imaging of the posterior fossa malformation and rocks of the two systems labyrinthine type almost unique vesicle (Geysler ear). Cardiovascular Exploration found dilatation of the aortic bulb with a non-compaction and without left ventricular dysfunction. On the genital level, cytogenetics, radiological assessments and hormonal balance sheets were in favor of a XY pure gonadal dysgenesis (Table I). The skeletal radiographs showed no other bone deformities.

Therapeutically, the child is oriented to pediatric surgery for the treatment of cryptorchidism and hypospadias, pediatric cardiology monitoring for his heart, in otorhinolaryngologie for equipment and functional reeducation with neurological follow.

III. Discussion

The first description of the SKF dates back to 1912 (M. Klippel) who reported the case of a patient with a very shortened neck associated with other malformations anomalies.¹ The condition is characterized mainly by the improper union or fusion of two cervical vertebrae or more. Other associated symptoms of the syndrome vary greatly from person to person, can be diagnosed at birth and requires clinical evaluation and in-depth explorations. The observed anomalies include scoliosis, vertebral abnormalities, including half vertebrae, other skeletal abnormalities, hearing damage, some craniofacial malformations and congenital anomalies of the heart. Moreover, in some cases, there may be complications. Neurological lesions resulting from spinal cord due to the instability of the vertebrae.⁴ includes three main categories of KFS. Type I is characterized by long fusion

of the cervical and upper thoracic vertebrae.

Type II includes the most common form in addition to the merger of two cervical or thoracic vertebrae, additional skeletal abnormalities, such as incomplete development of one half of certain vertebrae and the fusion of the first cervical vertebra (atlas) with the occipital bone.

Type III is characterized by the fusion of neck vertebrae as well as thoracic or lumbar vertebrae. In addition, a fourth category was mentioned in which KFS is associated to agenesis normally fused bones that form the sacrum^{4,5} In some patients with KFS, a part of the spinal cord can be exposed due to incomplete closure of vertebrae (spina bifida occulta). The presence of a tuft of hair can be found on the underlying abnormality and, in some cases, the weakness of a foot and some urinary incontinence.^{6,7}

25 to 50 percent of individuals with KFS also have hearing loss. Deafness can be of transmission, sensory, or both.⁸

Several eye abnormalities may also be associated with KFS, such as strabismus, nystagmus, or coloboma. In addition, some affected individuals may have other craniofacial anomalies: asymmetrical face, lateral tilt of the head, cleft palate. KFS may have other defects. : Congenital malformations of the heart, often characterized by the presence of an incorrect opening in the wall between the two lower chambers of the heart; hypoplasia or agenesis of one or both kidneys, renal incorrect rotation (hydronephrosis); Testicular agenesis or dysgenesis with abnormal sexual differentiation. Very rarely, an intellectual disability is reported in these children.⁹

On the etiopathogenic level, the path physiological mechanisms of disease are not yet known. KF syndrome is a congenital disorder often sporadic, some familial cases were reported, however, which may suggest the dominant or recessive autosomal dominant pathology. Various genetic elements and probably exogenous factors may be involved in the onset of pathology. This results in a failure of the appropriate segmentation of embryonic tissue that develops in the cervical vertebrae in the first weeks of pregnancy.^{4,11}

Research is still needed to learn more about the various underlying mechanisms that may be responsible for KFS. When the diagnosis of KFS is mentioned should be eliminated as part of the differential diagnosis, surgical fusion of the vertebrae. It must also differentiate KFS ankylosing spondylitis, juvenile rheumatoid arthritis and fibrodysplasia ossificans.

Treatment is directed toward specific symptoms that present in each individual. Such treatment requires a coordinated multidisciplinary care which includes pediatricians, endocrinologists, surgeons, orthopedists, neurologists, cardiologists, otorhinolaryngologists, ophthalmologists, physiotherapists, occupational therapists and / or other doctors various specialties.¹³ Most affected individuals have a good prognosis if the syndrome is diagnosed early and the symptoms are treated in time. Activities that can cause neck injuries should be avoided.

IV. Conclusion

KFS is a very rare congenital disorder often sporadic clinical diagnosis. It needs a thorough clinical examination and para clinical looking deformities that can be associated to it. The treatment of this syndrome is towards the specific symptoms that present themselves in each individual.

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Figures



Fig 1 : short neck and hypoplastic pavilions ears



Fig3 : fusion of C1 and C2

Table I: Paraclinical results of patients

Exploration	Results	Normes
Cortisol sanguin (nmol/l)	509,8	367-631
ACTH sanguin (pmol/l)	53	154- 638
SDHEA (ug/dl)	1,98	0,7-3,9
17 OH P (ng/ml)	0,12	0,16-0,42
A4 (ng/ml)	0,13	0,05-0,17
Testosterone (ng/ml) Before HCG	0,03	0,04- 1
AFTER HCG	0,1	
abdominopelvic Echography	hypotrophic testicles inguinal TDroit : 9mm ; TGauche : 10 mm	
GenitoCystogram	Absence of Cavities and other relics. of the male type urethra. bilateral Vesicoureteral reflux	