Klippel-FEIL Syndrome-Appropos of A Case

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Abstract

Klippel-FEIL syndrome is a congenital skeletal malformation characterised by the fusion of various cervical and occasionally thoracic vertebrae, leading to low hair line and short neck. Occasionally it is associated with hearing loss and Sprengel deformity with winging of scapulae. This case is being reported for its rarity.

Klippel-FEIL syndrome is also known by its synonyms Brevicollis, congenital osseous torticollis and congenital synostosis of cervico-thoracic vertebrae. Although documented by Vlysses Aldrovandi (1642), Haller (1743) and Morganni (1746), the first definitive detail description was made by Klippel FEIL (1912). Jalladeau (1936) reported the

demonstrated inner ear anomalies in Klippel-FEIL syndrome, by polytomography.

Case Report

A 7 year old female patient presented with the complaints of limitation of neck movements since birth, impaired hearing, difficulty in swallowing and unclear speech.

Fig Showing the photograph of the patient

Fig. 2 Showing high arched plate bifid uvula and cleft plate

association of deaf mutism. Wildervanck (1952) described abducens paralysis, retraction of eyeballs and deafness, associated with this condition-termed cervico-oculoacoustic syndrome. Everberg et-al (1963)

History revealed that the child was born with a short neck and low birth weight. In the post natal period, baby had nasal regurgitation of milk on breast feeding.

She continued to have difficulty in swallowing liquids and semisolid food. The onset of speech occurred at age of 2 years and was described as nasal and unclear. Milestones were delayed. She suffered recurrent attacks of cold. She complained of impaired hearing since 1 year.

Examination

The child was of short stature with a short and webbed neck. The head appeared to be placed directly on the trunk, between the shoulders, with restricted neck movements. A low hair line was noted. Scapulae were higher than normal and arms could not be raised more than 90 degrees (Sprengel's deformity).

Examination of oral cavity revealed high arched palate, bifid uvula and partial cleft palate. (Fig 2) Auricles were normal but external auditory canal was narrowed. Bilateral retraction of tympanic membrane with impaired mobility was noted. A 512 CPS tuning fork test revealed bilateral negative Rinne. Audiometry

Fig. 3. Showing Fusion of cervical and vertebrae/ Hemivertebra into a single mass

revealed bilateral moderate conductive hearing loss. Lateral view X-ray of cervical spines showed fusion of bodies of C2 to C5 vertebrae. (Fig 3)

A diagnosis of Klippel-FEIL syndrome with cleft palate and bilateral conductive hearing loss (Secretory otitis media) was made. Bilateral myringotomy with aspiration of serous fluid was performed. This was followed by grommet insertion in both ears. The child was simultaneously treated with systemic and local decongestants and mucolytics. The patient was referred to plastic surgeon for management of cleft palate.

Discussion

It is suggested that Klippel FEIL Syndrome arises from faulty segmentation of the mesodermal somites between the third and seventh week of embryogenesis (Shoul Ritro 1952). The Syndrome is probably inherited as an autosomal dominant trait with poor penetrance and variable expression. It is twice as common in females than males. The Syndrome is characterized by fusion of cervical and occasionally upper thoracic vertebrae/ hemivertebrae into a single osseous mass, leading to short neck, low hair line. Other associated skeletal anomalies described are spina bifida, cervical rib, Sprengel's deformity, and platybasia. The skull is usually normal but high arched palate, cleft palate, bifid uvula and multiple jaw cysts have been reported.

A variety of neurological complications including cranial nerve paresis, syringomyelia or syringobulbia, chorio retinal atrophy, nystagmus and intellectual retardation may occur. Hemiplegia or quadriplegia and a curious symptom of bilateral synkinesis or mirror movements in the upper limbs have been described. Otic findings of malformations of external, middle and inner ear occur.

Hearing loss in Klippel FEIL syndrome is generally severe and of sensory neural type but occasionally a conductive component owing to ankylosis of ossicles can be present. Associated ventricular septal defect and Eisenmenger's complex have also been reported (Nora et al 1961).

In the differential diagnosis of Klippel FEIL Syndrome one must consider cleido cranial dysostoses, Sprengel's deformity, Wildervanck's syndrome and those abnormalities associated with webbing of the

neck without bony defect like Turner's syndrome and ptergium colli.

Treatment for cervical deformity as a rule is not indicated, though plastic operation may produce marked improvement. Sensory neural deafness is managed with auditory and speech training. If deafness is conductive, the cause should be evaluated and treated accordingly.

Summary

A 7 year old girl presented with Klipper-FEIL Syndrome associated with cleft palate and hearing impairment in both ears. The hearing loss was due to bilateral secretory otitis media. Hearing improved after bilateral myringotomy and grommet insertion, while the cleft palate was repaired by plastic surgery.

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