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Cervico-Ocula-Acusticus Syndrome of Wildervanck

Case Report*

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The association of Klippel-Feil, Duane's syndrome and deaf-mutism was first described as a new entity by Wildervanck in 1952.[1] Although over 50 cases have subsequently been described from different parts of the world,[2] there is a surprising lack of case reports in the American medical literature. The main purpose of this communication is to present a patient with this syndrome who also had a dumb-bell shaped cystic cranial lesion. It is suggested that more detailed examinations of these patients could result in uncovering an increased number of similar cases.

Case Report

F. O., A five-year-old girl was brought to this hospital with the chief complaints of swelling on the back of the neck and delay in starting to talk. According to the parents, the patient was the product of an uneventful pregnancy and labor with a normal delivery. The mother denied taking any drugs during the pregnancy. There was no history of cyanosis or jaundice following birth. The patient's walking was delayed until the age of 4 1/2 years. Deafness since birth was reported.

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The family history revealed that the father's paternal and the mother's maternal grandfathers were brothers. The mother was 20 years of age and the father was 25 at the time of the patient's birth. There was no one on either side of the family with congenital defects. The patient's younger brother was in good health.

Physical examination showed her weight to be 17 kg. (25th percentile), height 96 cm (<3 percentile), and head circumference 53 cm. The head was slightly tilted to the left and posteriorly. The face was asymmetrical, the left side being smaller than the right. The patient's neck was short with webbing and a low hair-line. A mass 3 x 3 cm in size, was seen on the back of the back of the neck. The left scapula was located higher than the right and there was also a moderate kyphoscoliosis. Neurological examination in brief showed the pupils to be round and equal in size with a normal reaction to both light and accomodation. The ocular fundi were normal. Visual acuity was not measured. She was unable to turn both her eyes laterally beyond the midline. Retraction of the globe and narrowing of the palpebral fissure on adduction (Duane's syndrome) was evident. She had no response to noise. The rest of the cranial nerves were intact. Neck movements were limited due to congenital deformity of the cervical region but there was no evidence of muscle weakness. Rapid alternating movement of the hands was grossly impaired due to mirror movements but no ataxia, dysmetria or intentional tremors were present. The deep tendon reflexes as well as planter responses were symmetrically normal. She was able to stand on one foot and the Romberg's sign was negative. There was no obvious abnormality of gait. Superficial sensation was normal, but a satisfactory estimate of vibration and position senses could not be made.

Laboratory findings: Routine urinalyses, blood counts, various blood chemistry estimations and urine chromatography for amino acids revealed no abnormalities. Buccal smears and chromosomal studies using peripheral leukocytes showed a normal female karyotype. X-ray studies demonstrated congenital fusion involving cervical vertebrae from 2-7 ((Fig. 1). There was a midline round osseous defect in the occipital bone measuring 1.5 cms in diameter. A small mass was seen protruding out of this defect. There was also spina bifida over the sacral area. Other findings included absence of pneumatisation of the petrous bones bilaterally and slight hydronephrosis and hydroureter on the right. Pneumoencephalography was carried out and revealed hydrocephaly. Surgical exploration disclosed the extension of the cystic mass in the neck through the bone defect into the posterior

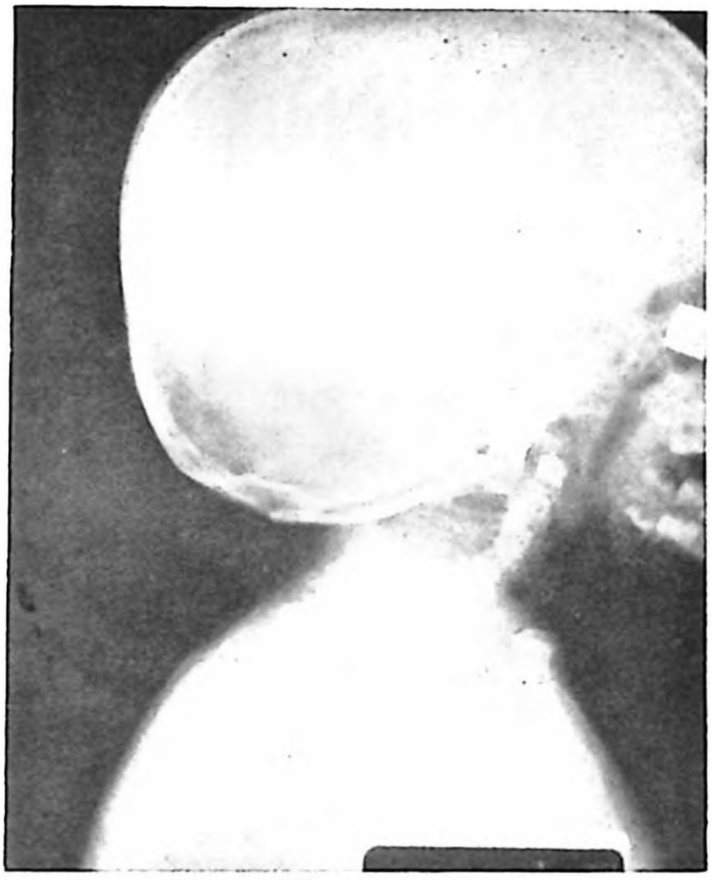

Figure 1

fossa between the two cerebellar hemispheres. The dumb-bell shaped cystic mass was 6 x 7 cm in size. Grossly, the pathological specimen was a grayish-white, firm, irregular membranous tissue, 2.3 x 0.6 cm in size. Histopathological studies showed rather simple histology, consisting chiefly of dense membranous collagenic fibers which tended to run in parallel fashions corresponding to the dura mater (Fig. 2). In some areas it was associated with a cobwebby net-work and a cerebellar of the arachnoid membrane which contained also calcospherites (Fig. 3). No neurological or other type tissue was present in any of the various sections examined. There was no suggestion of neoplasia but a somewhat disorganized appearance with prominent vascularity (Fig. 4) suggested that the lesion was the result of a developmental abnormality such as is seen in cystic lesions of dysrhaphic states. In the light of clinical information and X-ray findings the diagnosis of occipital meningocele was proposed for this lesion by the pathologist.

Audiometric testing in the "free field" using pure tones and frequency-calibrated sound effects yielded no reliable responses up to levels of approximately 90 dB level (100-110 dB Sound Pressure Level). Hearing screening on the parents were normal bilaterally.

Various X-rays, including the cranium and the spine in the parents and the maternal uncle and grandfather showed no abnormalities.

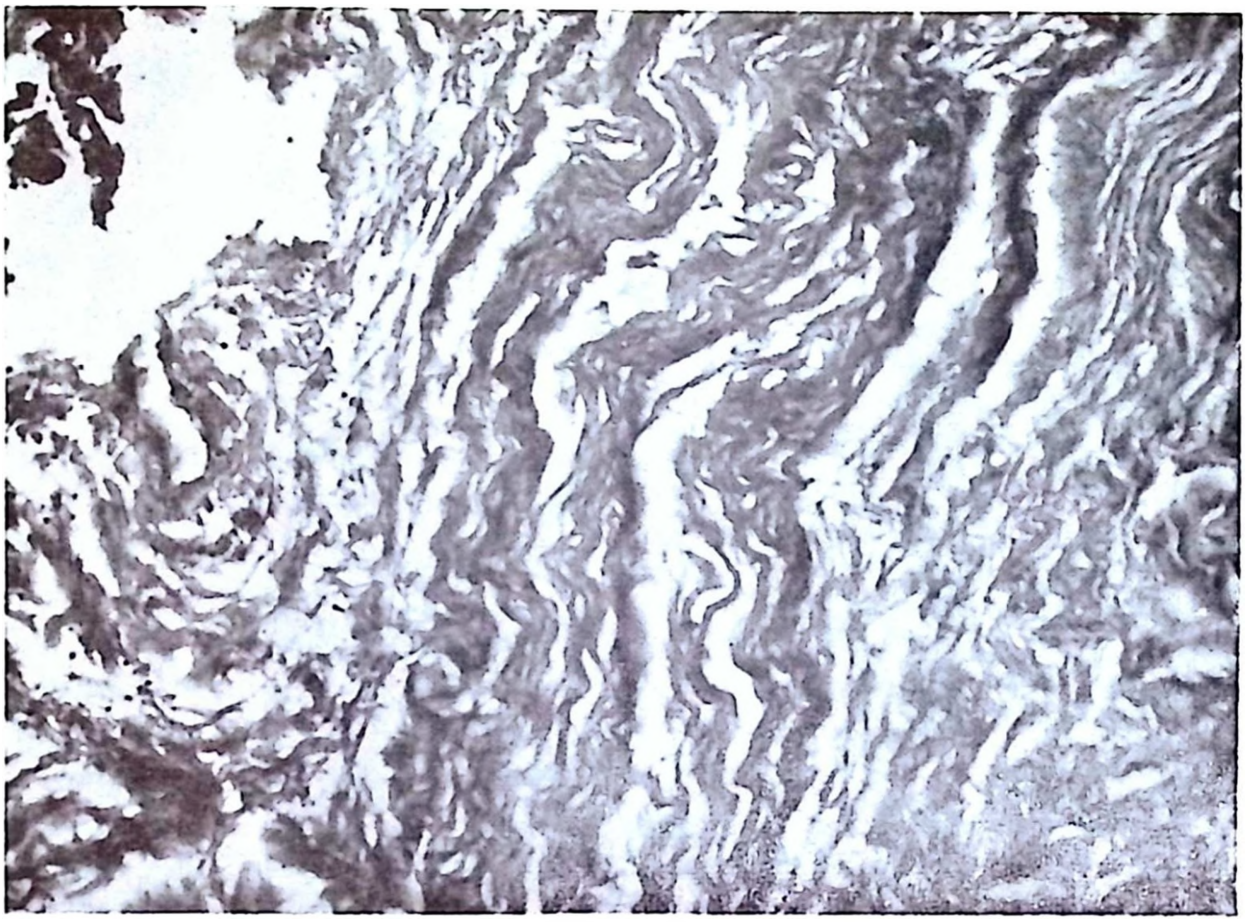

Figure 2

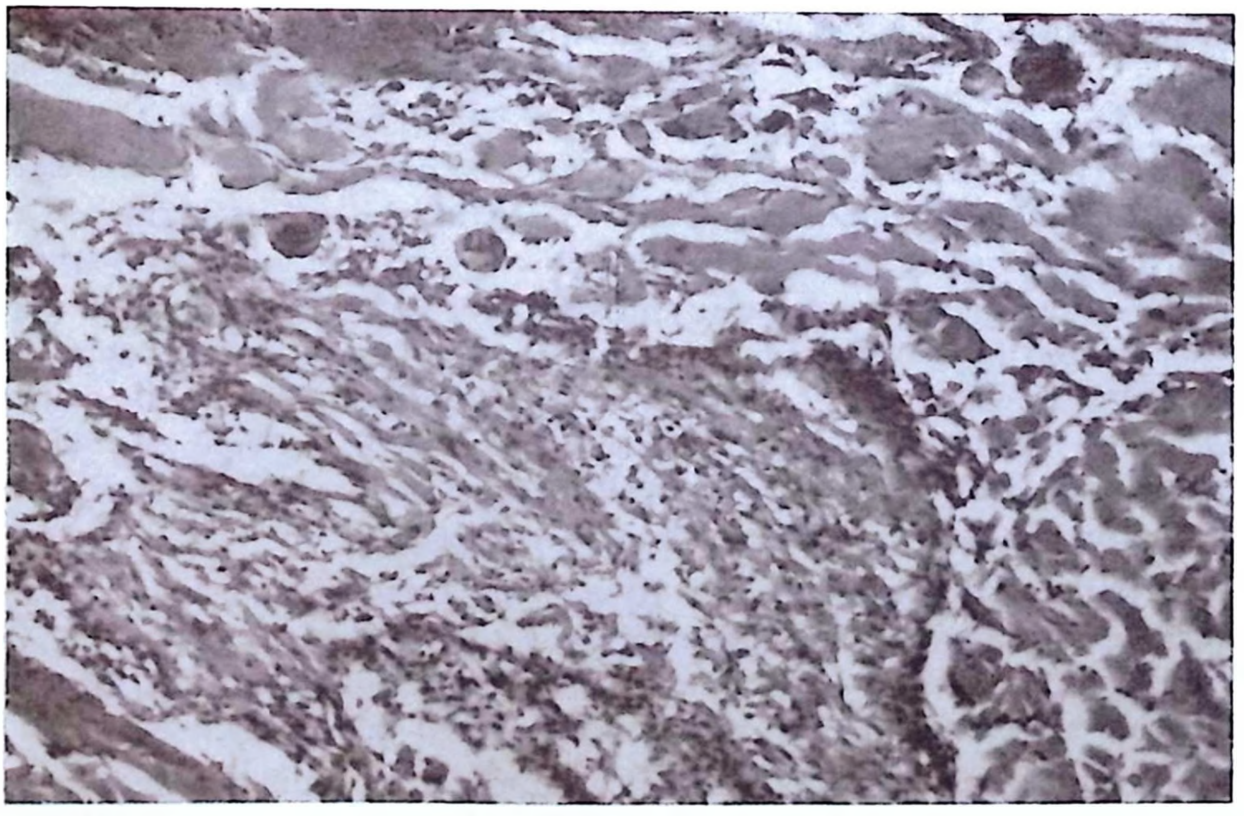

Figure 3

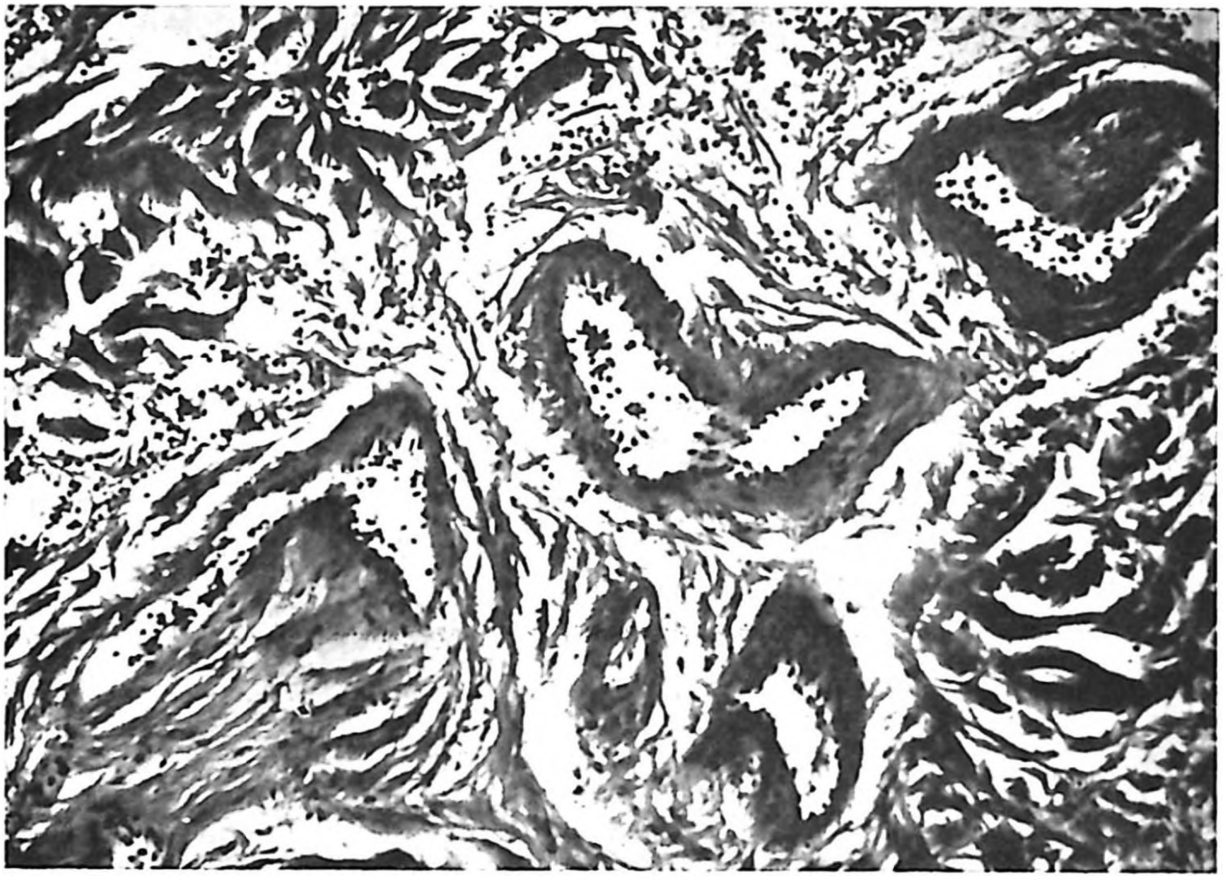

Figure 4

Discussion

It is generally accepted that this syndrome is of genetic origin, however, there is no agreement among the investigators as to its pattern of inheritance. McKusick[3] classified it among the sex-linked entities, while Wildervanck himself favored polygenic inheritance. It is also possible, as suggested by Fraser,[4] that the syndrome of Wildervanck is due to an undetectable chromosomal abnormality. It is of interest in this respect that the parents of our case were relatives.

Our patient had an occipital cystic mass in addition to the cardinal findings of the Wildervanck syndrome. We could find no case of this syndrome with a similar association in the literature. However, it should be noted that cases with extracranial benign lesions have been described. Franschesetti and Klein[5] a patient with Wildervanck's syndrome who also had subconjunctival lipoma. A patient in Wildervanck's own series had limbal dermoid.[2] Preauricular dermoids were found in two other cases.[4, 6] More interesting is the fact that two patients have been described by Kirkham in both of whom papilloedema was observed.[7] These findings, although not progressive and therefore considered by the authors as pseudopapilloedema, may, in fact, suggest the existence of a small intracranial mass lesion.

A partial explanation which would encompass some of the findings in our case may be the so-called "split notochord syndrome." Bentley and Smith[8] postulated a sagittal split of the notochord, as has been demonstrated previously in animals, as the probably cause of various spinal malformations, including accessory or persistent neuroenteric canal, dermoid cyst and sinuses, spinal and cranial cysts and Klippel-Feil syndrome. According to this theory, after the split takes place, it may close without trace during the process of development. However, when the closure is not satisfactory, it will result in various degrees of spinal defects. More severe lesions may enclose enteric or cutaneous elements giving rise to various malformations such as neuroenteric canals, dermoid cysts and/or sinuses. Hemivertebrae in these patients growing individually may show a fusion of the medial particles which constitutes the osseous element in the Klippel-Feil syndrome. Posterior meningoceles, as seen in our case, are due to a protrusion of the meninges through the occipital defect.

Finally, it would be of interest to know whether any of the reported cases would have revealed similar intracranial lesions if they had been investigated in detail, including surgical exploration. The case presented here indicates that such studies may be useful in detecting similar lesions in patients with various forms of spinal dysrhaphism.

Summary

A patient with Wildervanck's syndrome who also had an occipital meningocele is presented. It is suggested that more detailed examination of patients with this syndrome as well as with other forms of spinal dysrhaphism will result in uncovering an increased number of cases with internal lesions.

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REFERENCES

1. Wildervanck, L. S.: Een geval van aandovening van Klippel-Feil gecombineerd met abducens paralyse, retractio bulbi en doofstomheid, Nedel. Tijdschr. geneeskd. 16: 2752, 1952.
2. Wildervanck, L. S., Hoeksema, P. E., and Penning, L.: Radiological examination of the inner ear of deaf-mutes presenting the cervico-oculo-acusticus syndrome. Acta Otolaryngol. (Stockh.) 61: 445, 1966.
3. McKusick, V. A.: Mendelian Inheritance in Man, ed. 2, Baltimore, Johns Hopkins Press, 1968, p. 428.

4. Fraser, W. I., and MacGillivray, R. C.: Cervico-oculo-acoustic dysplasia. (The syndroma of Wildervanck), J. Ment. Defic. Res. 12: 322,, 1968.
5. Franceschetti, A., and Klein, D.: Dysmorphie cervico-oculo-faciale avec surdite familiale, J. Genet. Hum. 3: 176, 1954.
6. Fraser, G. R.: Profound childhood deafness, J. Med. Genet. 1: 118, 1964.
7. Kirkham, T. H.: Cervico-oculo-acusticus syndrome with pseudopapilloedema, Arch. Dis. Child. 44: 504, 1969.
8. Bentley, J. F. R., and Smith, J. R.: Developmental posterior enteric remnants and spinal malformations: the split notochord syndrome, Arch. Dis. Child. 35: 76, 1960.