The Journal of Laryngology and Otology
October 1998, Vol. **112,** pp. 976–978

Double epiglottis in Weyer's acrofacial dysostosis

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Abstract
While evaluating a 61-year-old patient for stridor we incidentally detected a double epiglottis. The patient was also diagnosed of having Weyer's acrofacial dysostosis which is characterized by hexadactyly affecting all four extremities, small and deeply set nails, dental deformities with small, conical teeth and mandibular hypoplasia. The double epiglottis was not the cause for the stridor. Because of the covert symptomatology of double epiglottis it is suggested that the association with Weyer's syndrome is common. Embryological evidence and a review of the literature on laryngeal abnormalities is discussed.

Key words: Epiglottis; Congenital defects

Introduction

Congenital laryngeal abnormalities are infrequent and are usually detected during childhood. Congenital syndromes eg: Cri du Chat syndrome, may be associated with laryngeal abnormalities (Manning, 1977). We report an unusual case of a double epiglottis in a patient with Weyer's syndrome.

Case report

A 61-year-old woman presented with sore throat, hoarseness and stridor. She had had antibiotic treatment for an upper respiratory tract infection for four weeks prior to presentation. There has been some improvement in her symptoms. The patient was known to have the autosomal dominant Weyer's acrofacial dysostosis, or Curry Hall syndrome, with the following features: hexadactyly affecting all four extremities, small and deeply set nails, dental deformities with small, conical teeth and mandibular hypoplasia. She also suffered from sensorineural hearing loss (40–70 dB) and a gelastic epilepsy with tonic/dystonic mime spasms, controlled with Tegretol and clonazepam. On examination she had mild biphasic stridor; all her other signs were normal. A soft tissue X-ray of the neck did not show any abnormality, but on laryngoscopy there was a small larynx with a double epiglottis and a tendency to adduct the vocal folds on inspiration (Figures 1 and 2). After conservative treatment the stridor subsided. It was felt that her stridor was a functional problem – the patient was referred for speech therapy.

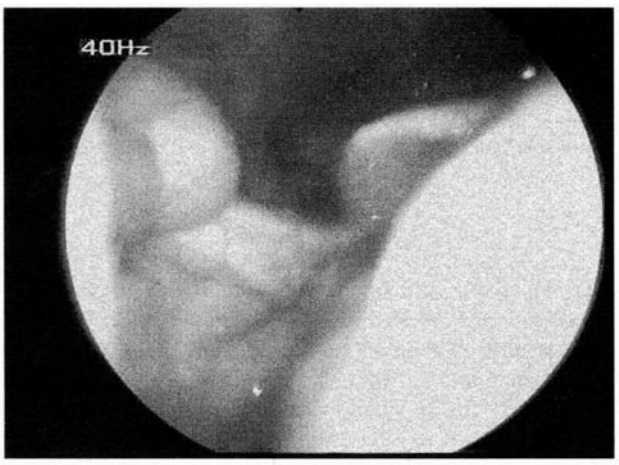

Fig. 1
Endoscopic picture of the double epiglottis.

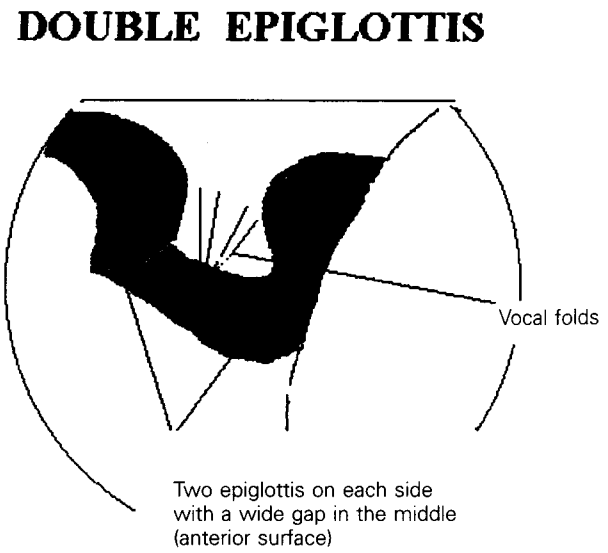

Fig. 2
Schematic representation of the endoscopic picture.

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Accepted for publication: 20 July 1998.

Discussion

The accidental detection of laryngeal abnormalities in adults is unusual. These abnormalities are usually found in infants or little children who are evaluated for stridor (Graham *et al.*, 1985; Goldenberg *et al.*, 1996; McClay *et al.*, 1997).

The commonest congenital laryngeal abnormality is laryngomalacia, accounting for 76 per cent of laryngeal abnormalities in a large series (Smith and Catlin, 1984). Vocal fold paralysis accounts for 10 per cent of congenital laryngeal abnormalities (Smith and Catlin, 1984). Subglottic stenosis, the third most common laryngeal abnormality occurs in approximately one per cent of the general neonatal population (Smith and Catlin, 1984). Atresias are very rare and mostly accompanied by other fatal abnormalities. Other rare conditions include laryngotracheo-oesophageal clefts and laryngeal cysts and laryngoceles.

Chromosomal abnormalities with associated laryngeal defects are very rare. Plott's syndrome presents with laryngeal abductor paralysis at birth causing severe impairment of phonation and inspiratory stridor when the paralysis is bilateral. In cri du chat syndrome the larynx may show laryngomalacia. The characteristic "cat-like" cry is probably central in origin (Manning, 1977).

The Cockayne syndrome is a disease of childhood characterized by mental retardation and premature aging. Abnormal stiffness of the larynx is found in this syndrome. There is no consensus on aetiology but the underlying pathology appears to be a metabolic dysfunction (O'Brien and Ginsberg, 1994).

Graham described three infants with bifid epiglottis. They had also different hand and foot anomalies – including polydactyly, syndactyly, clinodactyly, small hands with short fingers, absent finger creases, short metacarpals and dorsiflexed halux – and congenital hypopituitarism (Graham *et al.*, 1985). Two of the infants died shortly after birth probably because of unrecognized hypopituitarism and necroscopy revealed the bifid epiglottis. In the third infant the bifid epiglottis was noted on intubation. None of the children had stridor. The authors suggest an *in utero* insult during the 32nd and 42nd day of gestation because at this time the epiglottis as well as the hand plate and the hypothalamus develop. Like these three infants our patient also had anomalities of the extremities.

McClay *et al.* reported a family spanning three generations with bifid epiglottis and polydactyly of hands and feet (McClay *et al.*, 1997). In this family various degrees of bifid epiglottis were found leading to stridor: one six-week-old boy had a malacic half of the epiglottis which prolapsed into the glottis on inspiration. This was managed with supraglottoplasty with amputation of the malacic half of the epiglottis. Flexible fibreoptic evaluation of the child's relatives (grandmother, mother and three aunts) revealed variable degrees of bifid epiglottis. All were asymptomatic. The authors suggest a new genetic syndrome.

Goldenberg *et al.* reviewed the occurrence of bifid epiglottis (Goldenberg *et al.*, 1996) and found it to be rare and usually discovered after birth in the evaluation of stridor. It is not classified as a specific syndrome, but there are frequent associations with other congenital anomalies such as midline defects, endocrine disorders and central nervous system neoplasms such as hypothalamic hamartoma. The Pallister-Hall syndrome, mainly characterized by hypothalamic hamartoma, polydactyly and dysplastic nails is also associated with bifid epiglottis as well as with pituitary abnormalities and visceral anomalies including imperforate anus (Biesecker and Graham, 1996) Interestingly our patients otherwise asymptomatic son was suffering imperforate anus at birth. Accordingly Pallister-Hall syndrome shares a number of features with oral-facial-digital syndromes although an association with Weyer's syndrome has not been described thus far.

We report a patient with double epiglottis who had previously been diagnosed as having Weyer's acrofacial dysostosis. Weyer's syndrome was described first in 1952 and is characterized by postaxial hexadactyly, bony cleft of the mandibular symphysis (not seen in our patient), abnormalities of the lower incisors and the oral vestibule (Roubicek and Spranger, 1984). Curry and Hall described a large Spanish-Mexican family with the above mentioned abnormalities and short limbs (Curry and Hall, 1979). Shapiro *et al.*, reported another patient with the same features from a different geographic and ethnic background (Shapiro *et al.*, 1984). Roubicek and Spranger reported a four generation family with similar findings and additionally prominent ear antihelices, hypoplastic and dysplastic nails and a mild shortness of stature. To differentiate it from other acrofacial dysostosis they suggested calling the condition 'acrodental dysostosis' (Roubicek and Spranger, 1984) Ellis-van Creveld syndrome, an autosomal recessive disorder characterized by disproportionate dwarfism, polydactyly, cleft palate, natal teeth and congenital heart disease, has also been associated with Weyer's syndrome (Spranger and Tariverdian, 1995). In summary it appears, that Weyer's syndrome is a pleiotropic autosomal dominant disorder which can be associated with a number of other deformities. A laryngeal abnormality has not been previously described in this syndrome.

Our patient had obvious signs of Weyer's syndrome. The laryngeal abnormalities did not appear to cause any significant clinical symptoms. The coincidental finding of a double epiglottis (Figure 1) on laryngoscopy in a patient with apparently functional biphasic stridor does not imply that the obvious epiglottic abnormality is of functional importance. Thus it is possible that its presence is more common in Weyer's syndrome than previously thought, as such it may remain clinically covert. The finding of a double epiglottis in Weyer's acrofacial dysostosis also may not be incidental because of the embryological fact that the hand plate and the epiglottis develop during the same time, eg. from the 32nd to the 42nd days of gestation (Smith and Catlin, 1984; Graham *et al.*, 1985). This would suggest an *in utero* insult rather than a chromosomal abnormality. Clarification of this matter can only come with laryngoscopy in all patients known to have Weyer's syndrome.

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