Cleidocranial Dysostosis - A Case Report and Review of the Literature

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SUMMARY: Cleidocranial dysostosis is a generalised dysplasia of bone and teeth with predominantly autosomal dominant inheritance. A new case referred to the Paediatric Department at the British Military Hospital, Rinteln is reported and a review of the literature discussed.

Introduction

Cleidocranial dysostosis is a rare disorder of bone development and less than 1000 cases have been described since 1899. The aetiology and pathogenesis of the disease are unknown, although it has been described as a manifestation of both recessive and dominant inherited traits. Patients often consult the physician for something else and the disease is discovered incidentally. In the majority of cases the late emergence of teeth causes these patients to present to a dentist. Indeed the majority of reports occur in dental journals. It rarely presents in early life to a paediatric department.

Case Report

A two and a half year old boy was referred to the Paediatric Department at BMH Rinteln because of an irregular shaped head and a palpable anterior fontanelle. He had been a full term normal delivery, but was noticed to have wide sutures at birth and hence a cranial ultrasound was performed which was reported as normal. His head circumference at birth was 35cm and serial measurements from then on showed a head that grew in size at a rate such that it was above the 97th Centile, at one year (Fig 1).

The only other relevant history was that the father also had a large head, but in proportion to his overall build. He had a normal sister. Developmentally he had reached all his appropriate milestones.

On examination he was a playful, happy child who was

Fig 1. Head circumference of case.

Fig 2. Case pressing shoulders together.

easy to examine. While standing he showed a marked lordosis, a big head with a wide metopic suture and low set ears. Other abnormalities included a reshaped palate, hypertelorism eyes, bilateral clinodactyly and a protuberant abdomen. He was able to demonstrate the classical feature of being able to press his shoulders together (Fig 2).

Further investigations revealed normal biochemistry, but radiological investigation demonstrated in the case of the skull, Wormian bones, an anterior fontanelle still open, and supernumerary teeth (Fig 3).

Clavicles (Fig 4) were absent and in the pelvis, the ilia

Fig 3. Lateral skull X-ray of case.

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Fig 4. Chest X-ray showing absent clavicles of case.

(Fig 5) were hypoplastic. These radiological changes were consistent with the diagnosis of cleidocranial dysostosis.

Discussion

Cleidocranial dysostosis involves the bones that are ossified earliest in fetal life; the clavicle is the first bone to show ossification (1). It occurs with equal frequency among males and females and there are no racial differences. The genetics have been studied extensively and it is believed to be transmitted as an autosomal dominant, and various reports have estimated that between 16 and 39 per cent of cases are sporadic. There are reports, however, that indicate a rarer autosomal recessive inheritance pattern (2). The recessive form is characterised by being more severe and has dwarfism and extensive classical bony changes throughout the body, as its clinical features.

Patients with cleidocranial dysostosis have a bulging forehead and prominent cranial bossing. The facial bone and paranasal sinuses are hypoplastic, which causes the face to appear short. Hypertelorism is often present. There is delayed closure of the anterior fontanelle and sutures and indeed these may remain open into old age. There may be decreased auditory acuity as a result of narrowing of the external auditory canals due to hyperplasia of the mastoid bones. Maxillary hypoplasia gives the mandible a prognathic appearance. The palate is high and narrow (3,4). Because there is partial to total absence of the clavicles, the patient can press his

Fig 5. Pelvis X-ray showing hypoplastic ilia of case.

Fig 6. Clindodactyly of case.

shoulders together in front of the sternum to varying degrees (Fig 2). The clavicles are completely absent in about 10 per cent of cases. When there is unilateral absence it is usually the right clavicle (2). The thorax is 'cone shaped' and with the sloping shoulders may lead to respiratory distress in the neonatal period. In the pelvis pubic bones ossify late, often in the third decade. When the mother is affected, carrying a foetus with cleidocranial dysostosis i.e. a wide biparietal diameter - delivery must be by caesarean section because of cephalopelvic disproportion. In the hands, the terminal phalanges are pointed, the second metacarpal is lengthened by an epiphysis, and many have brachymesophalangy and clindodactyly of the fifth finger (Fig 6).

Although primary dentition is normal, permanent dentition is not. There is a delay in eruption of permanent teeth, even though they are all present in the jaw. The delay may be very long and there is one case of an edentulous lady cutting a tooth at the age of seventy eight years (4). The management of dental anomalies is the only area of treatment that needs to be addressed, and there are various modalities available.

Development and intelligence is usually normal in cleidocranial dysostosis, except in instances where there has been intrapartum brain damage from cephalopelvic disproportion. In the neonatal period the condition is sometimes misdiagnosed as hydrocephalus. Despite the absence or hypoplasia of the clavicles there appears to be no significant loss of strength, nor susceptibility to injury of the shoulder girdle, and work capacity is normal. Final height attainment though is significantly reduced in all forms, in both males and females. Life span is not reduced.

Diagnosis is principally made by radiograph, and often by the radiologist before referral to a physician. In addition to the clavicular changes, which are variable, the finding of Wormian bones in the skull together with dysplasia of the pubic symphysis can be considered

diagnostic of the disease (5). Biochemical investigations are usually normal.

This case illustrates two important features. Firstly, that this is not an autosomal dominant case unless there is incomplete penetrance. It could of course be autosomal recessive or a sporadic mutation. And finally it shows that no patient should be dismissed despite being developmentally normal with an odd feature without being investigated.

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