Occulocerebrocutaneous Syndrome: a case report

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Occulocerebrocutaneous syndrome is a rare condition characterized by orbital cysts and skin tags. The presence of supernumerary teeth has not previously been associated with this syndrome. A primary supernumerary tooth with a permanent supernumerary successor was found in this case. This highlights the importance of very careful examination when assessing children with syndromes.
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INTRODUCTION

Occulocerebrocutaneous Syndrome was delineated by Delleman *et al*.[1,2] The syndrome is characterized by the presence of orbital cysts, micropthalmia/ anophthalmia, focal dermal defects, aplastic skin defects, skin appendages and cerebral malformations.[3-6] Although there is a definite genetic component, the exact inheritance pattern of the syndrome is unknown.

Patients with this syndrome often have asymmetric facial features and this is due to the underdevelopment of their facial bones. Bones that are usually affected include the zygoma, and the sphenoid part of the lateral orbital wall. Some patients may also exhibit body asymmetry due to scoliosis and rib malformation.[1,7]

Associated skin appendages tend to be periorbital but postauricular, malar and even labial examples have been noted. Three types of focal skin lesions tend to occur; aplasia, hypoplasia, and punch like defects.[2,6]

We report the case of a seven-year-old boy diagnosed with Occulocerebrocutaneous Syndrome with a supernumerary tooth in the primary dentition. This has not been reported previously and may represent part of the phenotype.

CASE REPORT

A male infant was transferred to Great Ormond Street Hospital (GOSH) soon after birth. His parents were both healthy and non consanguineous. A normal prenatal ultrasound had been recorded at 20 weeks during an unremarkable pregnancy. Birth was an uncomplicated vaginal delivery and at this point the infant was noted to have a swollen left eye where the iris and sclera could not be seen. He was also found to have multiple skin tags, pits and healed scars on his left shoulder, chest and genitalia. In particular there was a large pedunculated tissue mass, which was approximately 2cm in diameter and was attached to the anterior chest wall (Fig 1). The skin abnormalities were examined by a dermatologist, who thought that they might be remnants of intrauterine scarring, possibly due to intrauterine infection.

Following his first feed the infant went into respiratory distress and a subsequent chest x-ray showed a typical appearance of a congenital diaphragmatic hernia. Examination of the cardiovascular system and abdomen was found to be unremarkable. Central nervous system examination with exception of the left eye was normal and an MRI taken at the time, showed no brain abnormality. Blood was sent for chromosome analysis and results showed 46XY normal male blood. A torch screen was also performed and this came back negative and therefore the infectious disease team thought that it was unlikely that the child had suffered a congenital infection.

The infant was taken for repair of the left diaphragmatic hernia, inversion appendectomy and excision of the skin lesion over the chest. The diaphragm was repaired with a patch and the histology of the skin lesion was a found to be a benign hamartoma.

Post operatively the baby was transferred to neonatal intensive care. The ophthalmology team examined him and an ultrasound scan was carried out on the left eye. This showed a small cystic lesion filled with a homogenous mass behind the globe. No vision in the left eye was recorded and the right eye appeared to be clinically normal. The child was referred to Moorfields Eye Hospital where they kept him under observation until the age of four. It was decided to wait for a few years before operating on the orbital cyst because its development was helping the left eye socket to develop normally. This was important to create space behind the left eyelid, allowing the placement of an artificial eye.

Due to the clinical features of micropthalmus, orbital cysts and the characteristic skin lesions described previously the child was diagnosed with Occulocerebrocutaneous syndrome and his care was taken over by the Neurologists and Geneticists at Great Ormond Street Hospital.

Initially the child was referred by his geneticist to the Maxillofacial and Dental Department at four years of age for assessment of his dentition. He had no complaints and no history of dental treatment. Extra oral examination was unremarkable apart from the obvious facial asymmetry. The child's left eye was lower than the right (Fig 2).

Intra oral examination found the patient to be caries free with good

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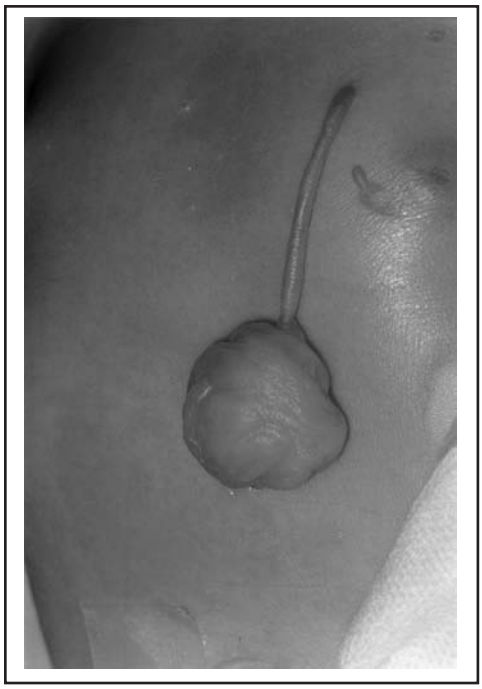

Figure 1: Photograph showing a pedunculated skin tag, attached to the anterior chest wall.

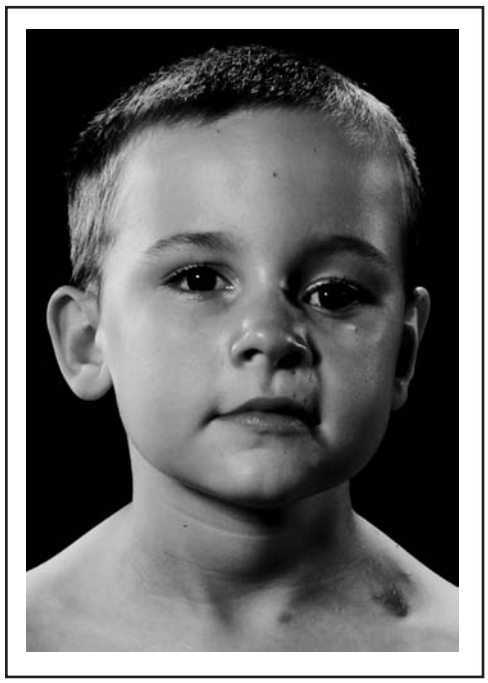

Figure 2: Full face photograph showing facial asymmetry, in particular left orbital dystopia.

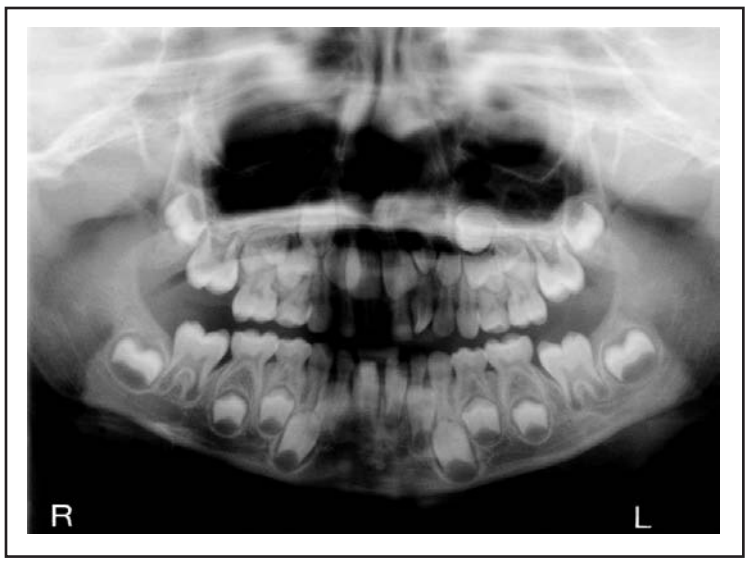

Figure 3: Fig 3: OPG showing presence of supernumerary teeth.

oral hygiene. The enamel was of normal quality and quantity. All primary teeth were present and a supernumerary tooth was noted between the upper left central and lateral incisors. Preventative advice was given and the patient kept under six monthly reviews.

At one of these early visits an OPG was taken to assess the developing dentition, it was then that the existence of a second supernumerary in the permanent dentition was noted (fig 3). In subsequent visits it was noted that the lower left first permanent molar had hypoplastic enamel. This was subsequently restored with composite resin.

At the last appointment the patient, now aged seven years, was in mixed dentition. He had no complaints and the medical history was described as before, with no significant change. Dental history showed the patient had been brushing his teeth twice a day with a children's toothpaste but his parents were trying to wean him off this and get him to use adult toothpaste. His diet had improved considerably and the patient was consuming very few sweets and chewing sugar free gum instead.

The primary supernumerary was still present in the upper left anterior region. The restoration on the lower left first permanent molar was sound and no further restorations were required. A repeat OPG was taken so that the position of the unerupted teeth including the supernumerary could be assessed. The positions of both supernumerary teeth were the same. Further review appointments were arranged to monitor the developing dentition. It was decided that should the supernumerary teeth impede the eruption of the permanent teeth then they should be extracted.

DISCUSSION

This is the first recorded case of a primary supernumerary tooth being found in a child diagnosed with Occulocerebrocutaneous Syndrome.

According to most accounts, supernumerary teeth occur more frequently in the permanent dentition than in the primary dentition.[8] Studies have found that the range in which supernumeraries occur in the primary dentition varies considerably from 0.03 to 1.9%.[9,10]

The low incidence of primary supernumerary teeth can be attributed to two factors, the first being that they are often normally shaped, and frequently appear to be in normal alignment, therefore are often overlooked. The second is that they tend to exfoliate. [9,11,12]

Studies have shown primary supernumerary teeth occur most frequently in the lateral incisor region whereas permanent supernumeraries occur in the central incisor region. Also unlike their permanent counterparts they tend to erupt and their occurrence is not sex related.[12-15] Supernumerary teeth have been associated with a number of syndromes including Cleidocranial Dysplasia and Gardner's syndrome among many others. In this case it may be pert of the phenotype or an incidental finding.

It is likely that the enamel hypoplasia on the first permanent molar is environmental rather than genetic in view of the medical history. As further permanent teeth erupt the enamel will be examined for defects to aid diagnosis. Should the composite restoration fail an occlusal onlay may be required.

The exact effects of the supernumerary teeth in this case have not yet developed. If the primary supernumerary does not exfoliate removal will be required. The unerupted supernumerary has a favourable position and may erupt. Future treatment could involve simple extraction, which will hopefully allow the permanent incisors to erupt into a good position. The main complication is interference with the eruption of permanent dentition, including malpositioning, delayed eruption or non-eruption. Future management will involve correcting any such anomalies.

The presence of supernumerary teeth in dentition of someone with Occulocerebrocutaneous Syndrome has never been reported. The case highlights the importance of dental assessment in patients with syndromes so that the phenotype can be accurately recorded.

REFERENCES

1. Delleman JW, Oorthuys JWE. Orbital cyst in addition to congenital cerebral and focal dermal malformations. A new entity. Clinical Genetics. 19: 191-198, 1981.
2. Delleman JW. Orbital cyst in addition to congenital cerebral and focal dermal malformations. A new entity. Clinical Genetics. 25: 470-472, 1984.
3. Moog U, Die Smulders C, Systermans JM, Cobben JM. Oculocerebrocutaneous syndrome: report of three additional cases and aetiological considerations. Clinical Genetics. 52 (4): 219-225, 1997.
4. Moog U, Kruger G, Stengel B, Smulders C, Dykstra S, Wagemakers E. Oculocerebrocutaneous Syndrome: a case report a follow up, and differential diagnostic considerations. Genetic Counselling. 7(4): 257-265, 1996.
5. Baraitser M, Winter RM. Colour Atlas Of Congenital Malformations Syndromes. Mobsy Wolfe. 188, 1996.
6. Gorlin RJ, Cohen M, Stefan Levin JRL Syndromes of the head and neck 3rd Ed. Oxford University Press. 511-512, 1990.
7. Giorgi PL. Oculocerebrocutaneous syndrome. Description of a new case. Eu J Paediat.148: 325-326, 1989.
8. Angle B, Hersh JH. Anopthalmia, Intracerebral Cysts and cleft Lip/Palate: Expansion of the Phenotype in Oculocerebrocutaneous Syndrome? Am J Med Gen.68: 39-42, 1997.
9. Pinborg, JJ. Pathology of the dental hard tissues. Copenhagen; Munksgaard 28, 1970.
10. Bodin I, Julin P, Thomson M. Hyperdontia I. Frequency and distribution of supernumerary teeth among 21,609 patients. Dento maxillofacial Radiology 7: 15-17, 1978.
11. Luten JR. The prevalence of supernumerary teeth in primary and mixed dentitions. J Dent Child. 34: 346-353, 1967.
12. Ravn JJ. Aplasia, supernumerary teeth and fused teeth in the primary dentition. Scand J Dent Res. 79: 1-6. 1971.
13. Jariven S, Lehtinen L. Supernumerary and congenitally missing primary teeth in Finnish children. Acta Odontologica Scandinavica. 39: 83 -86, 1981.
14. Stafne EC. Supernumerary teeth. Dental Cosm. 74: 653-659, 1932.
15. Magnusson B. Tandovertal I den den temporara dentitionen. Svensk Tandlak Tid. 52: 357-372, 1959.