CASE REPORT

Rare case of orofaciadigital syndrome type I

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drabhishek_kgmu@yahoo.co.in**SUMMARY**

Orofaciodigital syndrome (OFDS) is a group of congenital anomalies which affects the face, oral structures and digits. There are nine subtypes with different modes of inheritance. OFDS type I is an X-linked dominant trait with lethality in the vast majority of affected males. We report a case of OFDS type I in an Indian girl at the age of seven who had most of the typical features of OFDS type I and nephrocalcinosis.

BACKGROUND

Orofaciodigital syndrome (OFDS) is a group of congenital anomalies which affects the face, oral structures and digits. There are nine subtypes (not without overlapping features) with different modes of inheritance.¹ The OFDS type I is a multiple congenital anomaly syndrome, first described by Papillon-Léage and Psaume.² This rare syndrome presents with anomalies of the oral cavity, digits and face and causes variable mental deficiency in more than half the number of the patients.^{3,4}

CASE PRESENTATION

We report a case of a 7-year-old girl, who had dolicocephalic skull, flattened facies, low set ears with notching of both ear lobules (figure 1). In the oral cavity multiple anomalies were seen—flat and thin lower lip, bilateral upper lateral frenulae, complete cleft palate, multiple lobulations of the anterior part of the tongue, a soft rounded swelling on the

dorsum of the tongue on the left side. The teeth were badly carious (figures 2 and 3). There was brachydactyly type A1 of the hands and feet (figure 4). Partial alopecia was present but milia were absent. There was developmental delay (more of motor milestones) and the IQ was 66%. Rest of systemic examination was normal.

The skeletal radiographs were normal. Echocardiography and ECG were normal. An ultrasound revealed bilateral nephrocalcinosis (figure 5). No cyst found in the kidney on ultrasonography. CT cranium was normal. Fine needle aspiration cytology of the swelling on the tongue revealed choristoma with lipomatous element. Hearing assessment was carried out which showed moderate conductive deafness along with bilateral B type of tympanogram suggestive of bilateral serous otitis media. Blood and urine investigations performed in view of nephrocalcinosis (blood urea, serum creatinine, serum parathyroid hormone and thyroid stimulating hormone, 24 h urine creatinine, calcium, oxalate and citrate) were all normal.

INVESTIGATIONS

The skeletal radiographs revealed spina bifida at S₁S₂S₃. Echocardiography and ECG were normal. An ultrasound revealed bilateral nephrocalcinosis. CT cranium was normal. Fine needle aspiration cytology of the swelling on the tongue revealed choristoma with lipomatous element. In view of

Figure 1 Right and left lateral view of the face.

To cite: Singh AB, Girhotra M, Goel M, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2012-007733

Figure 2 Gingiva and teeth.

Figure 3 Oral cavity.

the difficulty in hearing, hearing assessment was carried out which showed moderate conductive deafness along with bilateral B type of tympanogram suggestive of bilateral serous otitis media. Blood and urine investigations performed in view of nephrocalcinosis were all normal. Genetic/molecular analysis was not performed due to financial prospects.

DISCUSSION

OFDS type I is associated with dysfunction of primary cilia.⁵ The phenotype includes the following abnormalities: oral (lobated tongue, hamartomas of the tongue, cleft of the hard or soft palate, accessory gingival frenulae, dental caries and anomalous anterior teeth), facial (ocular hypertelorism, hypoplasia of the alae nasi, median cleft or pseudocleft upper lip and micrognathia), digital (brachydactyly, syndactyly, clinodactyly, preaxial or postaxial polydactyly of the hands and duplicated hallux), brain (intracerebral cysts and cerebellar agenesis) and kidney (adult polycystic kidney disease).^{4 5} Mild degrees of intellectual disability may be present in about half the patients.^{4 5} The features of our patient were consistent with the phenotype of OFDS I. She also had nephrocalcinosis, not previously described.

The condition is inherited in an X-linked dominant pattern with lethality in the vast majority of affected males.⁶ Mutations in OFD I gene have been identified to be the genesis of this condition. Individual variation in the X-inactivation of the pattern of this gene may also explain the clinical variability observed in OFDS I.⁷ Approximately 75% of affected individuals are simplex cases (ie, with no family history of OFDS I). The risk that the unaffected mother of an affected simplex case will give birth to another female with OFDS I is less than 1%. At conception the risk to the offspring of females with OFDS I of inheriting the disease causing allele is 50%. Since affected males miscarry the expected sex ratio of offspring is 33% unaffected females, 33% affected females and 33% unaffected males.⁵

Learning point

- A greater understanding of the syndrome would assist in earlier diagnosis. Management involves managing the individual defects surgically and genetic counselling prior to conception. Reports of OFD I in Asians are extremely rare and we did not come across any published case from our country.

Figure 4 Hand and feet.

Figure 5 Right and left kidney in ultrasonography.

Acknowledgements We acknowledge Mr Vivek Tiwari Central Library, King George Medical University, Lucknow who helped us in typing.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Commissioned; internally peer reviewed.

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