OCULO-DENTO-DIGITAL DYSPLASIA

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A 21-year-old male was seen with typical oculo-dento-digital dysplasia. However, photophobia and corneal turbidity were additional features in our patient. These have not been reported earlier.

Key words : Oculo-dento-digital dysplasia, Photophobia, Syndactyly.

First described by Meyer-Scwickerath et al in 1957,[1] oculo-dento-digital (ODD) dysplasia is a syndrome consisting of : (1) characteristic facies exhibiting a thin nose with narrow nostrils and hypoplastic alae, (2) micro-ophthalmia with anomalies of the iris, (3) syndactyly of the fourth and fifth fingers, and (4) hypoplasia of the enamel of teeth. The syndrome is rare and has infrequently been reported in the literature.[2,3] The purpose of this communication is to report a case of ODD in a Libyan male, the first from this country.

Case Report

A 21-year-old male patient was seen for dryness of skin over the face and lustreless dry hair, but physical examination revealed several abnormalities. The patient was the first of 6 siblings, 4 males and 2 females. None of the family members had any skeletal or facial abnormalities or any other significant disease. There was no history of consanguinity among parents. The antenatal period and the delivery of the patient had been uneventful but at birth he was noted to have bilateral webbing of the second and third fingers of both the hands and toes. The milestones of development had been delayed but the physical growth and mental development appeared to be normal. Correction of the syndactyly on the left hand was carried out 8 years ago.

The height of the patient was 150 cm and weight 52.5 kg. He appeared to be thin built and underweight for his age. The head was narrow and elongated in the vertical direction, and mandible seemed to be receding. He had typical facies with hypoplasia of the alae nasi. The ears were lower than normal and exhibited a prominent bulge in the upper portion of the pinnae. The eyes were small and closely set, with prominent epicanthal folds. There was bilateral turbidity of the corneas with impaired visual acuity. The teeth were yellowish, carious and loosely set with hypoplasia of the enamel. The skin of the face was slightly scaly. Hair were sparse and lusterless but there was no abnormal pigmentation. Corrected syndactyly was noted in the left hand between the middle and the index fingers which was rudimentary. Right hand showed syndactyly between 2nd and 3rd fingers and camptodactyly of the 2nd finger. In the left foot there was syndactyly between the 3rd and 4th toes. Neurological examination was unremarkable. Hearing in both the ears was normal. Review of the other systems did not reveal any abnormality.

Routine laboratory investigations and an electroencephalogram were normal. Skiagram of the skull revealed a minor bulging of the front of head and the incisors were incomplete.

Comments

The clinical findings in the patient were typical for ODD. Paucity of case reports in the literature suggests that the syndrome is rare.[4]

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It appears to be inherited as an autosomal dominant trait.[2,3] The clinical findings in the reported cases have been rather consistent apart from some variations recorded in individual cases. The characteristic facies, hypoplasia of the nasal alae, eye and dental abnormalities and syndactyly of fingers and toes are diagnostic. Since the mental development is normal,[3] usually the life span is not affected and these patients lead a normal life. However, the eye abnormalities can at times cause problems. The most common eye change is microcornea[3] and often there is associated micro-ophthalmos with small orbits. Secondary glaucoma, congenital cataracts, reduced lip apperture, persistence of pupillary membranes and optic atrophy have also been reported.[2] In most cases vision has been essentially normal. In our patient turbidity of cornea on both sides reduced the visual acuity for both eyes to 6/18. In addition the patient had photophobia which has not been described earlier in this syndrome.

The changes in the hands in the form of bilateral syndactyly of the fourth and fifth fingers with camptodactyly and ulnar clinodactyly have been typical in most cases. In our patient there was syndactyly of fingers and toes and camptodactyly of right index finger. Clinodactyly was not observed. Hair in our patient were dry and lustreless. This has also been a consistent feature of this syndrome.[5,6]

References

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