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Oculo-Dento-Digital Dysplasia (Oddd), Report of a Case

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Abstract

Oculo-dento-digital dysplasia (ODDD) is a congenital disorder manifesting with multiple phenotypic abnormalities involving the face, eyes, teeth, and limbs in addition to neurologic symptomatology. We report on a case of oculodentodigital syndrome with peculiar dental and multysistemic findings suggesting that a multidisciplinary managment is mandatory along with an appropriate genetic counseling once the causative mutation is detected.

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Key word: Oculo-dento-digital dysplasia, hypoplastic enamel, cutaneous syndactyly

Introduction

Oculodentodigital syndrome (ODDD, MIM # 164200) is characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding (1)

Neurologic abnormalities are sometimes associated (2) and lymphedema has been reported in some patients with ODDD (3,4,5)

Clinical Report

We report on a 14-year-old female born at term from non consanguineous parents; her birth weight was 3310 gr, lenght 50 cm and cranical circumference 34cm. At

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birth, maternal-fetal isoimmunization was found, for which phototherapy was performed for few weeks
She was diagnosed with cutaneous syndactyly between the fourth and fifth fingers, bilaterally, subjected to surgical correction at the age of 3-year old.

In a regular period, the girl reached the first stages of motor and relational development. From the age of 27 months he has control, even at night, of the sphincters.
Subsequently presents microcornea and astigmatism. Systemic manifestations suggest the supect of oculodentodigital syndrome.
After DNA sequencing a mutation in GJA1 in heterozygosis was detected (3).

She came to our attention at the age of 4-year-old presenting scarce oral hygiene, multiple caries in both permanent and primary dentitions, enamel hypoplasia resembling amelogenesis imperfecta, wide spaced teeth; under deep sedazion with midazolam she underwent to several dental treatments such es extraction of 75, 16, removal of placque and calc accumulation; she came then in 2021 and she showed deep caries of 46, 26, 27 and 25; the collaboration was improved hence the dental treatments could be carry out in the dental clinic without the use particular behavioral managment. Again the enamel was hypoplastic, severe open bite, on skeletal basis thus destinated to maxillo facial intervention at the end of growth. Hypoplastic enamel and scarce oral hygiene are completely against any orthodontic treatment although in presence of indications.

INHERITANCE
- Autosomal dominant
HEAD & NECK
Head
- Microcephaly
Ears
- Dysplastic ears (in some patients) - Hearing loss, conductive
Eyes
- Microcornea - Microphthalmia - Short palpebral fissures - Epicanthal folds - Glaucoma - Cataract - Iris anomalies
Nose
- Small nares - Thin hypoplastic alae nasi - Narrow nasal bridge - Thin anteverted nares - Prominent columella
Mouth
- Cleft lip - Cleft palate - Broad alveolar ridges
Teeth
- Enamel hypoplasia - Selective tooth agenesis - Microdontia - Premature loss of teeth - Dental caries - Taurodontism (reported in 1 patient)
CARDIOVASCULAR
Heart
- Endocardial cushion defects (uncommon) - Atrial septal defect (uncommon) - Ventral septal defect (uncommon) - Cardiac conduction defects (uncommon)
ABDOMEN
Gastrointestinal
- Bowel dysfunction (in some cases)
GENITOURINARY
Bladder
- Neurogenic bladder (in some patients)

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SKELETAL
Skull
- Skull hyperostosis
Spine
- Vertebral hyperostosis
Pelvis
- Hip dislocation
Limbs
- Broad tubular bones - Cubitus valgus
Hands
- Syndactyly of 4th - 5th fingers - Short middle phalanx of the 5th finger - Fifth finger camptodactyly - Midphalangeal hypoplasia - Clinodactyly
Feet
- Syndactyly of 3rd - 4th toes
SKIN, NAILS, & HAIR
Skin
- Diffuse yellow-orange non-epidermolytic hyperkeratosis on palms and soles (palmoplantar keratoderma)
Nails
- Brittle nails
Hair
- Fine, dry hair - Sparse, slow-growing hair
MUSCLE, SOFT TISSUES
- Lymphedema of lower limbs (in some patients)
NEUROLOGIC
Central Nervous System
- Mental retardation (rare) - Hyperactive deep tendon reflexes - Paraparesis - Quadriparesis - Ataxia - Spasticity - Dysarthria - Seizures - Neurogenic bladder - Basal ganglia calcification - Cerebral white matter abnormalities
MISCELLANEOUS
- Variable phenotype - Cardiac features are observed in ~3% of cases - Neurologic features have been diagnosed in ~30% of cases - 50% of cases represent new mutations associated with advanced paternal age

Table 1 summarise clinical features and inheritance pattern of oculodentodigital sybdrome.

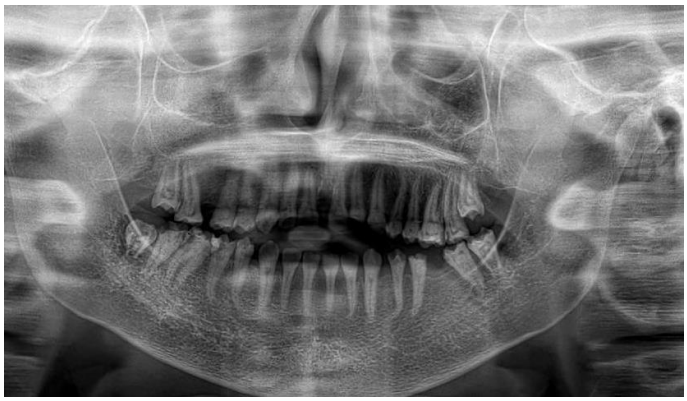

Figure 1. Orthopantomography showing signs of amelogenesis imperfecta , microdontia, severe dental decay in all present tetth.

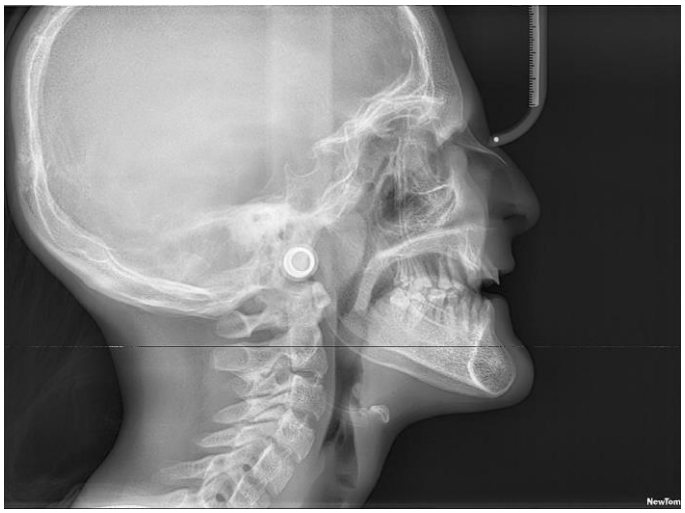

Figure 2. Teleradiography showing severe open bite.

Discussion and Conclusions

Oculo-dento-digital dysplasia (ODDD) is a congenital disorder manifesting with multiple phenotypic abnormalities involving the face, eyes, teeth, and limbs in addition to neurologic symptomatology. Dental features such as enamel hypoplasia, dental decay, premature loss of primary teeth due to their fragility, cleft lip are reported in the literature associated with ODDD (4-12). This report aims to present a female patient with ODDD who was referred due to extensive dental needs. The presence of hypoplastic enamel and scarce oral hygiene

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triggered further orthodontic evaluation evaluation. Characteristic facies with hypoplastic alae nasi and syndactyly offered greater insight into the phenotype of the syndrome. It is important to be aware of genetic disorders associated with characteristic dental malformations to offer appropriate counseling and treatment and to refer these patients to a pediatric dentist in order to prevent possible complications connected to hypomineralized enamel.

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