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EC PAEDIATRICS
Case Report

A 9 Year Old Boy with Hypodontia

Jorge Sales Marques*

Pediatric Department, Centro Hospitalar Vila Nova de Gaia, Portugal

***Corresponding Author**: Jorge Sales Marques, Pediatric Department, Centro Hospitalar Vila Nova de Gaia, Portugal.

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Abstract

The Ectodermal dysplasia (ED) is a rare hereditary disorder, inherited by x linked recessive in 95% of cases and dominant in 5% of situations.

The disorder can involve the teeth, skin, nails and sweat glands.

We report a 9 years old boy, that went to the stomatology because of dental caries at 8 years of age.

The dental x-ray showed absent of 21 teeth and the superior incisive with abnormal shape.

The boy don't have problem of sweat or intolerance to heat, hyperpyrexia after only mild exertion or a meal.

The physical examination revealed a normal phenotype, with pointed incisive teeth and hypodontia.

The father has congenital absent of 2 superior canine teeth without any other symptoms.

The ED was confirmed by gene study.

The risk of recurrence of the next pregnancy is 50%, because of the autosomal inheritance.

Keywords: *Pointed Incisive; Hypodontia; Ectodermal Dysplasia*

Background

The Ectodermal dysplasia (ED) is a rare hereditary disorder, inherited by x linked recessive in 95% of cases and dominant in 5% of situations.

The disorder can involve the teeth, skin, nails and sweat glands.

Not always all the symptoms appeared at the same time in the patient. The child can show only hypodontia or absent of sweating, abnormal nails or keratosis. All depends on the ectoderm involvement. In addition to the previous symptoms, central nervous system and adrenal medulla can be affected, pharyngeal and laryngeal mucosa may be atrophic, causing dysphonia and hoarseness of voice.

Hypohidrotic ectodermal dysplasia (HED) is the most common form and is transmitted as an X-linked recessive disorder. The mother will have hypodontia or conical teeth and reduced sweating.

The incidence is between 1:10,000 to 1:100,000 male live births.

EDA is associated with X-linked HED. Ninety-five percent of individuals with HED have the X-linked form. EDAR and EDARADD are inherited as autosomal dominant and autosomal recessive forms of HED. Both are associated with 5% of HED.

Different subgroups are created according to the presence or absence of the four ED defects:

- ED1: Trichodysplasia (hair dysplasia)
- ED2: Dental dysplasia
- ED3: Onychodysplasia (nail dysplasia)
- ED4: Dyshidrosis (sweat gland dysplasia).

If the disorder appears before the 6th week of embryonic life, the dentition will be the main noted. After 8th week, the other ectodermal structures will be affected.

Case Report

A 9 years old boy, with past history of snoring at 2 years of age cause by adenoids hypertrophy but without any later surgery, hands erythema and impetigo at 7 years old treated with fusidic acid, went to the stomatology because of dental caries at 8 years of age.

The dental x-ray showed absent of 21 teeth and the superior incisive with abnormal shape.

The boy don't have problem of sweat or intolerance to heat, hyperpyrexia after only mild exertion or a meal.

The physical examination revealed a normal phenotype, with pointed incisive teeth (Figure 1) and hypodontia.

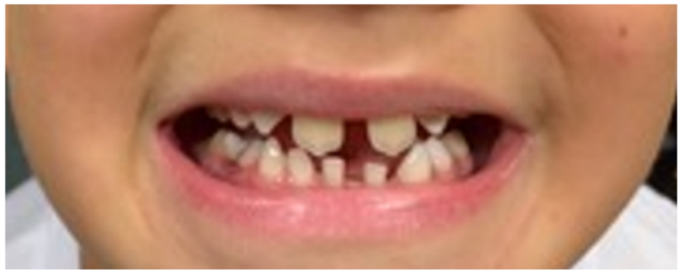

Figure 1: *Pointed incisive teeth and hypodontia.*

No any signs of skin or nails abnormalities. No hoarseness. No heart murmurs or lungs with wheezing or reduced sounds. No hepato-splenomegaly. Eyes and ears are normal at observation.

Normal motor skills and no learning disabilities.

The parents are unrelated. The father has congenital absent of 2 superior canine teeth without any other symptoms.

Discussion

This case is a typical finding of an ED, with dental dysplasia 5 % of all cases. The genetic counseling in this case is a risk of 50% for next pregnancy, since the father is affected.

According to the subgroup classification, this is an ED2, that cause hypodontia or absent teeth.

Our case, although the patient has past history of skin involvement, the lesions at that moment and later no more evolution to keratosis or nails dysplasia, seems that the only ED affected are the teeth like his father.

The gene study was focus not on x linked recessive inheritance but in dominant transmission. The EDARADD was negative but this result cannot rule out the diagnosis.

The prognosis of the disorder depends on the dental correction and prosthesis.

Conclusion

When we observe a patient with abnormal shape of the incisive with absent teeth, is important to consider Ectodermal dysplasia as first diagnosis and check for others signs that can or not appear in the skin or nails, without any history of sweating during fever or exercise.

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