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Gingival hyperplasia and delayed exfoliation of primary teeth in a patient with Chromosome abnormalities – A case report

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Abstract
Hypertrichosis is characterized by increased growth of the hair, which is beyond the normal variation and in areas that are not predominantly androgen-dependent. Gingival overgrowth, a common gingival disease, can be caused by inflammation, drug use, leukemic infiltration, and malignancies. Tooth eruption disorders can manifest in several clinical conditions, where the oral location, the number of affected teeth, and the etiology of the disorders vary considerably.
This clinical case reports delayed permanent teeth eruption with generalized gingival hyperplasia in a child with a genetic disorder and heredited hypertrichosis. A 10-year-old male patient was referred to the pediatric dentist with the main complaints of delayed eruption of permanent teeth and swollen gums. The child was diagnosed with high-grade hydronephrosis in the right kidney before birth. Genetic tests revealed that the child has a pathological male karyotype 45, XY, with a translocation between chromosomes 13 and 14 inherited from the maternal side. On extraoral examination, dysmorphic facies, a brachyplatycephalic head configuration, coarse facial features, a broad nose, and a short neck were evident. The gingiva was pale pink with a fibrous consistency and extended on all tooth surfaces. The gingival tissue did not bleed on probing. Delayed eruption of permanent teeth was observed, with only twelve primary teeth present.
Rare diseases often show unique dental-craniofacial manifestations, and pediatric dentists, oral maxillofacial surgeons, and orthodontists are responsible for identifying and managing each patient's oral symptoms.

Keywords: Hypertrichosis, gingival hyperplasia, case report, rare disease, delayed tooth eruption

Introduction
Hypertrichosis is characterized by increased growth of the hair, which is beyond the normal variation and in areas that are not predominantly androgen-dependent [1]. These features are usually independent of age, race, or sex [1]. Hypertrichosis is classified according to the degree of distribution (localized or generalized), the age of its beginning (acquired or congenital), and whether the condition is isolated or related to various abnormalities [1]. The use of drugs, infection, neoplasia, genetic diseases, and metabolic nonendocrine disorders have been the main reasons for hypertrichosis.
Gingival overgrowth, a common gingival disease, can be caused by inflammation, drug use (including phenytoin, nifedipine, or cyclosporine), leukemic infiltration, and malignancies [2]. In contrast, Idiopathic Gingival Fibromatosis is a rare disease with an undetermined cause [3]. Known by various terms such as hereditary gingival hyperplasia, elephantiasis gingiva, diffuse fibroma, familial elephantiasis, gingivostomatitis, and congenital familial macro gingiva, IGF presents as a non-hemorrhagic, diffuse increase in bulk of keratinized gingiva [3]. The firm and leathery consistency of the gingiva leads to functional disturbances and compromised esthetic, psychological, and social issues, significantly impacting the lives of those affected [4]. This anomaly typically begins with the eruption of either primary or permanent teeth, resulting in prolonged retention of primary dentition, the late eruption of permanent teeth, and malocclusion [4, 5].
Tooth eruption disorders can manifest in several clinical conditions, where the oral location, the number of affected teeth, and the etiology of the disorders vary considerably [6]. Genetic factors are often the cause of eruption failure [6]. Other systemic conditions associated with impairment of growth, such as anemia and renal failure, have also been correlated with delayed tooth eruption and other abnormalities in dentofacial development [7].
This clinical case reports delayed permanent teeth eruption with generalized gingival hyperplasia in a child with a genetic disorder and heredited hypertrichosis.

Case report
A 10-year-old male patient was referred to pediatric dentist with the main complaints of delayed eruption of permanent teeth and swollen gums.
Based on medical history, the child was born through an expected delivery from the mother's first pregnancy. The mother did not mention any chemical intake or medication during pregnancy. The paternal side of the family has a history of diabetes mellitus. The child was diagnosed with high-grade hydronephrosis in the right kidney before birth, which was confirmed by ultrasound after birth. The child has had generalized hypertrichosis all over his body and face, as well as facial dysmorphism since birth. During the first few months of life, the child had several uro-infections, which led to the removal of the right kidney at the age of 8 months. Genetic tests revealed that the child has a pathological male karyotype 45, XY, with a translocation between chromosomes 13 and 14 inherited from the maternal side. The child also has a heterozygous deletion of the long arm of chromosome 17 - 17α12, which has genes associated with developing renal abnormalities and diabetes mellitus.
On extraoral examination, dysmorphic facies, a brachyplatycephalic head configuration, coarse facial features, a broad nose, and a short neck were evident (figure 1).

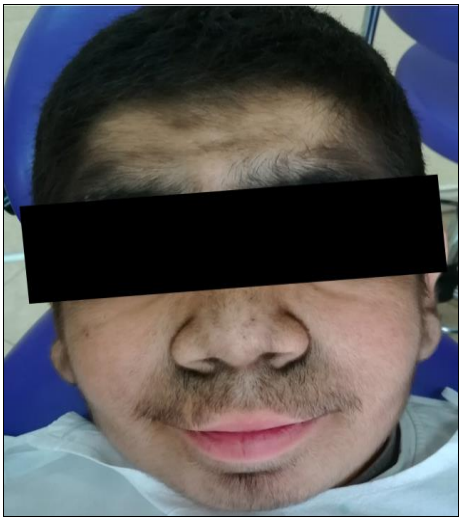

Fig 1: Extraoral status with dismorphic facies, brachyplatycephalic head configuration and broad nose

A bilateral quadrifinger groove is observed on both palms. Intraoral examination revealed diffuse gingival enlargement in the entire dentition (figure 2).

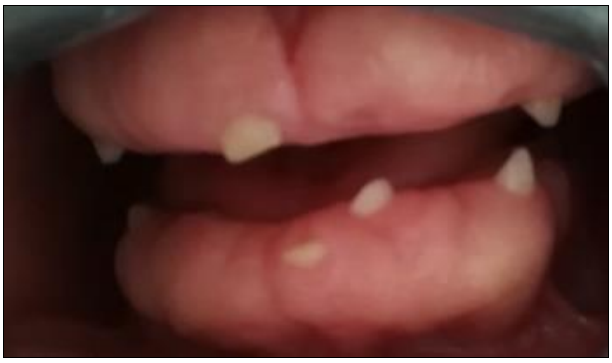

Fig 2: Intraoral examination with diffuse gingival enlargement and persisting primary teeth

The gingiva was pale pink with a fibrous consistency and extended on all tooth surfaces. The gingival tissue did not bleed on probing. Delayed eruption of permanent teeth was observed, with only twelve primary teeth present.

The primary teeth were dislocated but with standard color, structure, and size. No dental or orthodontic treatment has yet been carried out. The radiograph confirms the presence of 12 primary teeth and all permanent teeth, which do not show any features of structural defects (figure 3).

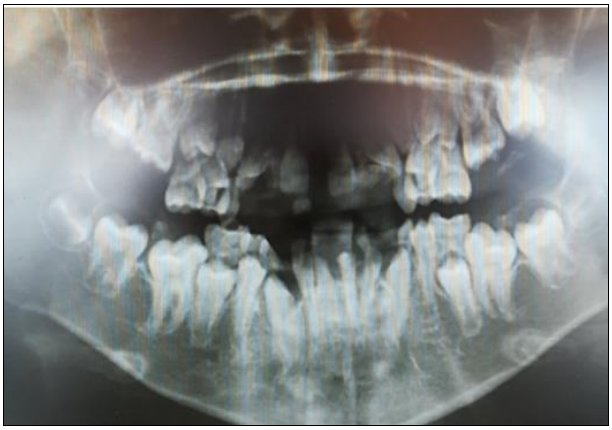

Fig 3: Panoramic X-ray with presence of all permanent teeth

(The patient's parents signed informed consent for publication of the clinical case materials.)

Discussion

Microdeletion and microduplication of chromosome 17q12 are recurrent chromosomal imbalances that originate from non-allelic homologous recombination. 17q12 deletion syndrome is a result of deletion of a fragment of chromosome 17 in each cell. The deletion occurs on the chromosome's long (q) arm at a position designated q12 [9]. The central affected genes, hepatocyte nuclear factor-1-bet and LIM homeobox 1 (*LHX1*), play a crucial role in the development of this syndrome [8]. The symptoms and signs of 17q12 deletion syndrome can be different and vary, even among affected members of the same family [9, 10]. Among the more common features associated with these chromosomal changes are problems with the development or function of the urinary system, specifically the kidneys [9]. These abnormalities range from mild or no problems with kidney and urinary tract function to severe malformations, leading to kidney failure before birth, to mild or no problems with kidney and urinary tract function [9]. Our patient was diagnosed with high-grade hydronephrosis in the right kidney before birth, which caused several uro infections after birth that led to the removal of the right kidney at the age of 8 months.

The dysmorphic features seen in patients with 17q12 deletion syndrome are known to be highly variable, with a prevalence of approximately 67% [11]. The most commonly reported features include a high forehead, frontal bossing, depressed nasal bridge, deep-set eyes, full cheeks, down-slanting palpebral fissures, high palate, and high-arched eyebrows [12]. Other features such as hypoplastic nails, 2-3 finger/toe syndactyly, and clinodactyly of the fifth finger are also frequently observed [12]. Our patient noted specific symptoms such as coarse facial features, a broad nose, and a short neck (figure 1), which are consistent with the diagnosis of 17q12 deletion syndrome.

An intraoral examination of our patient revealed diffuse gingival enlargement in the entire dentition, with pale pink gingiva and fibrous consistency. Delayed eruption of permanent teeth was also observed. Hereditary gingival fibromatosis with hypertrichosis is a rare disease characterized by varied gingival overgrowth and generalized hair growth, impairing function, and unaesthetic appearance [13]. This condition is usually treated by removing the hyperplastic tissue via surgical intervention. Following treatment, regular recall visits with reinforced oral hygiene measures can reduce recurrence rates [13].

Our patient presented with a complex case of generalized delayed teeth eruption, a manifestation of his genetic disorder. Managing such eruption disorders in these conditions is multifaceted, with various methods suggested [7]. These include no treatment (observation), elimination of obstacles to the eruption (e.g., cysts, soft tissue overgrowths), exposure of affected teeth with and without orthodontic traction, autotransplantation, and systemic disease control. The variation in the normal eruption of teeth is a common finding. However, significant deviations from established norms should serve as a call to action for the clinician to delve deeper into the patient's health and development [14].

Conclusion

It is of utmost importance for each clinician to fully comprehend the related clinical features and potential treatments. This understanding is crucial for early diagnosis and management, which can significantly improve the prognosis of patients with rare genetic disorders. Rare diseases often show unique dental-craniofacial manifestations, and it is the responsibility of pediatric dentists, oral maxillofacial surgeons, and orthodontists to identify and manage each patient's oral symptoms.

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