

## Abnormal Dentition in a Boy with Incontinentia Pigmenti:

### Case Report

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#### Abstract

Incontinentia pigmenti (IP) is an X-linked dominant genodermatosis characterized by typical skin lesions along Blaschko's lines and associated with ocular, dental, nails, hair, skeletal, central nervous system and cardiovascular anomalies. We report a 5-year-old boy with cutaneous hyperpigmentation along Blaschko's lines, atrophic streaks, strabismus and mental retardation. He showed the characteristic abnormal dentition seen in IP as partial hypodontia, peg-shaped anterior teeth and un-erupted teeth. The expression of IP in boys is exceptional as the disease is lethal in males.

**Key Words:** Incontinentia Pigment; Dentition; Male

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#### INTRODUCTION

Incontinentia pigmenti (IP, Online Mendelian Inheritance of Man [OMIM] No. 308300) is a rare X-linked dominant genodermatosis characterized by typical skin lesions along Blaschko's lines and associated with ocular, dental, nails, hair, skeletal, central nervous system and cardiovascular anomalies [1].

Dental abnormalities are most frequent and present in 60% of patients older than 1 year [2,3]. Partial anodontia of deciduous or permanent teeth and peg-shaped or conical teeth are the most frequent dental problems [1-4]. Eruption of teeth may be incomplete or delayed [2-4]. Some patients were reported to have multiple caries and "crumbly" teeth [3].

In patients with incontinentia pigmentia and dental abnormalities, significant alterations in the mineralization of the teeth have also been observed. Early caries and general poor dental quality may be specific for both first and second dentitions.

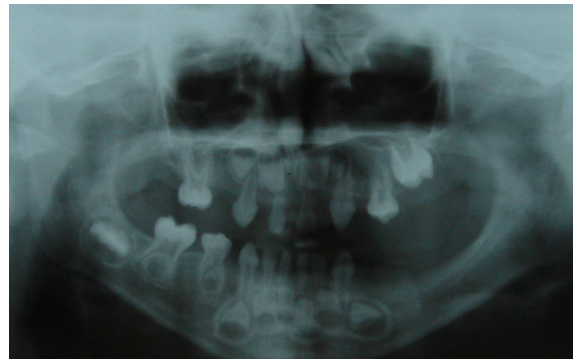
The diagnosis of dental changes can not be given earlier than the age of one year [2]. We report a boy with IP showing characteristic dental findings.

#### CASE REPORT

A 5-year-old boy presented to the dentist for abnormal dentition. He was born to a 40-year-old mother, G2P2, at full term after an uneventful pregnancy.



**Fig1.** Hyperpigmentation along Blaschko's line on the trunk



**Fig 2.** Radiographic view of the Patient

His skin had been fiery red at birth and vesicles had developed shortly afterwards. They had been replaced by verrucous lesions after a few weeks. The lesions then had cleared gradually leaving linear pigmentation. In physical examination, he was overweight and mentally retarded. Hyperpigmented lines were noted along Blaschko's lines in his trunk and limbs (Figure 1). Hypopigmented atrophic streaks were also seen on his upper limbs.

Alopecia and strabismus were other positive physical findings. His parents were not consanguineous and his older brother was healthy. He was 25 years old. History of similar disease was not present in the family. The diagnosis of IP was made. His genetic counseling was otherwise normal.

An oral examination, including panoramic radiograph was completed (Figures 2-5). The soft tissue was normal and only 14 primary teeth were present. First and second mandibular left primary molar teeth (K,L) were absent and in the maxilla, right first primary molar, right primary central incisor, left primary lateral incisor and left second primary molar (B,E,G,J) were missing.

On oral examination of the patient, some of both mandibular and maxillary anterior teeth were conical and in the radiograph, the primary teeth roots were normal. The first right mandibular permanent molar (30) showed a delayed development compared to their maxillary counterparts.

Regarding susceptibility to caries, the teeth were normal and there was only occlusal caries in the first right primary mandibular molar tooth (S) which was treated by amalgam. The child's dental health was normal and with good occlusion. The interdental spaces were visible due to some missing teeth, sub-mucosal cleft palate and the conical condition of some other teeth. No hypoplasia or hypocalcification were seen on the teeth.

Radiography revealed 13 un-erupted permanent teeth, including six mandibular anterior teeth (22,23,24,25,26 and 27), first right premolar (28) and molar (30), four maxillary incisors and first left molar (6,7,8,9,10,11 and 14).

## DISCUSSION

IP is an X linked dominantly inherited disorder reported primarily in females and believed to be embryonically lethal in the majority of males. In the majority of cases IP is due to a mutation in a gene called NEMO (NF- $\kappa$ B essential modulator) which normally protects against TNF-induced apoptosis.

It is characterized by typical skin lesions along Blaschko's lines usually evolving through four stages: 1-Vesicular (from birth or shortly thereafter); 2-Verrucous (between 2 and 8 weeks of age); 3-Hyperpigmented (several months of age into adulthood) and 4-Hypopigmented (from infancy through adulthood) [5].

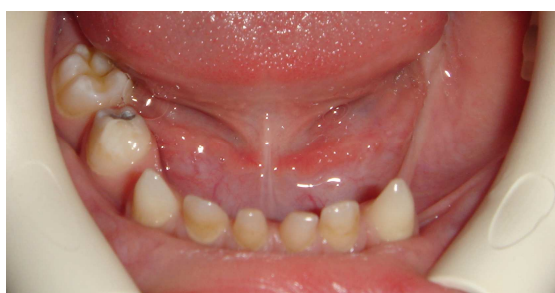
The inflammatory sequence sometimes recurs within the pigmented area during the first few



**Fig3.** Clinical view of the Patient in centric occlusion



**Fig 4.** Clinical view of the Maxilla



**Fig 5.** Clinical view of the Mandibular

months of life, particularly at the time of febrile illness.

The early inflammatory phase of IP reveals eosinophilic spongiosis and scattered dyskeratotic keratinocytes. The epidermis of verrucous lesions is acanthotic with dyskeratosis and foci of dyskeratosis. In stage 3, there is pigmentary incontinence; whereas, stage 4 is characterized by a thinned epidermis and dermis devoid of appendages [6]. Associated cutaneous findings include patchy scarring alopecia, woolly hair nevi, nail dystrophy and anhydrosis in atrophic streaks. Extracutaneous features include dental abnormalities (partial anodontia and conical or peg-shaped teeth), CNS abnormalities (seizure, delayed psychomotor development, MR, spastic hemiplegia) and ocular disease (retinal vascular abnormalities, strabismus and blindness) [1]. Therefore, all affected infants should be referred for a baseline ophthalmologic examination and periodic neurodevelopmental and dental evaluation.

No therapeutic measures influence the natural evolution of IP, treatment being symptomatic [7]. Our patient showed the classical sequential cutaneous findings of IP (vesicular, verrucous lesions and pigmentation along Blaschko's lines as well as linear hypopigmented atrophic hairless lesions).

Mental retardation, strabismus and anomalous dentition were other supportive features.

Although IP is mainly seen in females and generally lethal in males in utero, surviving males are exceptional. Some male cases were due to concomitant Klinefelter syndrome (XXY), but in the majority, no such association was present.

Postzygotic mutation and hypomorphic alleles may explain IP in such otherwise genetically normal males [8].

Our patient had neither a family history of IP nor an abnormal karyotype. An outstanding feature in our patient was his characteristic abnormal dentition, for which the parents sought dentist consultation.

In our case, dental examination showed partial hypodontia, peg-shaped teeth and delayed teeth eruption, typically seen in IP [1-4]. Partial hypodontia is the most common dental anomaly with a prevalence of 43%. There were no signs of other abnormal findings including gothic plate that were reported by Minić [2] and Himelhoch et al. [3]. Unlike studies reporting hypocalcifications and prevalent caries [3], our patient had no enamel de-

fects or susceptibility to caries and only minimal caries on the occlusal surface of the first primary right mandibular molar was detected. This patient was interesting for the presentation of IP in a boy as well as his characteristic dental findings.

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