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**Case Report** **Pediatric Dentistry**

# Dental Management in Children with Incontinencia Pigmenti (Bloch-Sulzberger Syndrome): Case Reports and Literature Review

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## Abstract

***Introduction*:** Incontinentia Pigmenti (IP) is a multisystem genodermatosis characterized by cutaneous, neurologic, ophthalmologic, and dental abnormalities. Skin alterations are the main characterestic of IP. They can change and even disappear over time, in contrast, dental ones are permanent and considered as the most frequent extra cutaneous signs. The aim of this work is to present two rare clinical reports of Incontinentia Pigmenti and to adress its frequent orofacial manifestations in the dental litterature. ***Case Reports***: In this paper, we report two clinical cases of incontinencia pigmenti in two female patients, five and nine years old, who were first examined by a dermatologist due to skin changes and then referred to our pediatric dentistry department in la Rabta hospital for oral examination. We managed these cases in collaboration with the pediatric department. We also conducted a research in the following electronic databases: PubMed and google scholar using the following keywords: ("Incontinentia Pigmenti" AND "Child" AND "Tooth Abnormalities"), from 2011 to 2021, to compare dental findings in our patients with literature. ***Discussion*:** The most representative dental features of IP are: delayed tooth eruption, agenesis, high arched palate, peg like and malformed teeth, affecting both primary and permanent teeth. Therapeutic management of our patients consisted in the replacement of absent teeth with an aesthetic and functional space maintainer. The assessment of orofacial manifestations in Children with Bloch Sulzberger syndrome indicates that it appears almost exclusively in females and is usually lethal in males. The clinical expression of this disease found in our patients reflects the Data reported in the literature. The latter showed that the presence of symptoms other than skin changes is important if dermatological signs are subtle, which is in accordance with both our patients' clinical expression. The most relevant finding of this review is the observation that most IP patients with odontological findings had the congenital absence of six or more teeth opposed to our second patient who had only one absent tooth. ***Conclusions*:** Incontinencia pigmenti is of a good prognosis but it may affect patients' quality of life hence the importance of knowing this syndrom and its oro-facial characteristics in order to obtain a correct diagnosis and apply an appropriate approach treatment.

**Keywords:** Bloch-sulzberger, rare syndrom, dermatologic, oral manifestations, dental, management.



## INTRODUCTION

Incontinentia Pigmenti (IP), also known as « Bloch-sulzberger » disease, is an X-linked dominant syndrom predominantly affecting females and lethal in males because of x-chromosome dizygosity and negative selection of cells carrying the mutant x chromosome.

It's a rare genodermatosis with an incidence of 0.7 per 100.000 births [1, 2].

This multisystem disease is characterized by cutaneous, neurologic, ophthalmologic and dental abnormalities. Skin alterations are the main characterestic of IP. These manifestations can change

and even disappear over time evolving through four stages: visiculor, hyperkeratotic, hyperpigmented and hypopigmented [3, 4].

In contrast, dental abnormalities are permanent and considered as the most frequent extra cutaneous signs [3, 6].

Diagnosis usually is made from the clinical presentation and occasionally skin biopsy which is associated with genetic examination [2, 6].

In this work, we report two clinical cases of incontinencia pigmenti managed in the department of pediatric dentistry at the University Hospital Rabta, in Tunisia, North Africa, in collaboration with pediatric one and we also conducted research in different electronic databases to compare dental findings in our patients with literature.

## CASE REPORTS: CASE ONE

A nine-year-old girl diagnosed with Bloch-Sulzberger syndrome was referred to the Pediatric Dentistry Department at the university hospital Rabta for a dental rehabilitation. The general physical examination revealed no skin changes with normal facial features.

The child was born by full term and normal delivery. Her mother reported that she was born with some skin lesions on the upper lumbs treated with topical corticoid. She also has ocular disorders but no central nervous system abnormalities. The diagnosis of the IP has been established by clinical examination associated with anatomopathological exam (Fig. 1).

**Fig. 1: Facial characteristic features of an IP patient**

Clinical and radiological oral check-up revealed multiple dental agenesis, more than six primary and permanent teeth. As we can notice the #12, #22, # 31, # 41, # 36, #46 and the germ of #14, #15, #24, #25, #34, #35, #44, #45 were absent (Fig 2a, 2d). We also noted conical crowns and dysmorphic molars #16 and #26 (Fig 2b). The patient worried about an early dental loss. Indeed, flat palate and resorbed alveolar ridges were mentioned (Fig 2c).

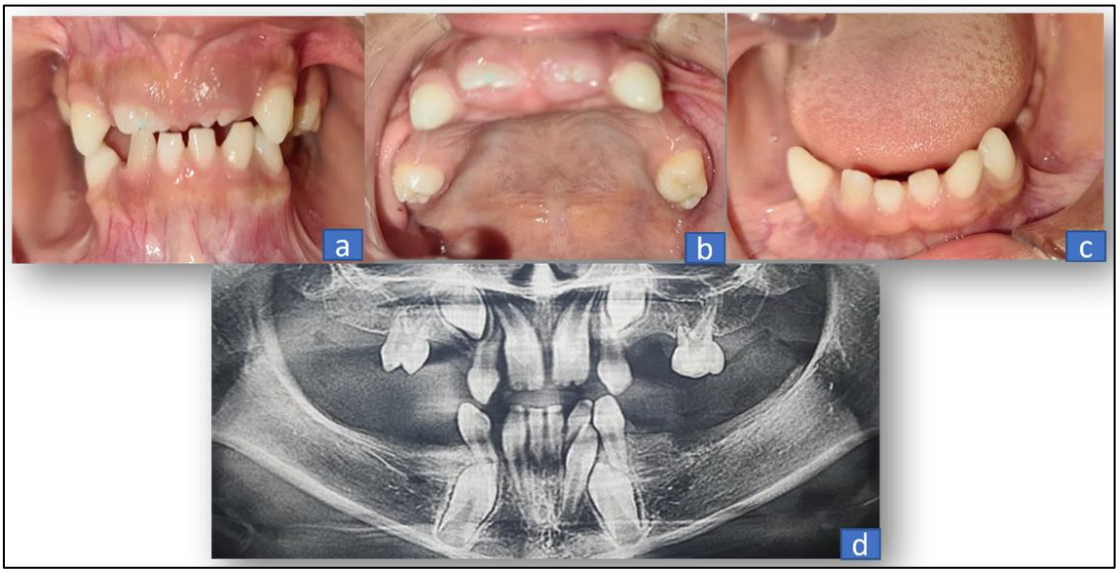


**Fig. 2: Dental abnormalities in a child with IP**

The therapeutic management consisted in the replacement of absent teeth with an aesthetic and functional space maintainer (Fig. 3).

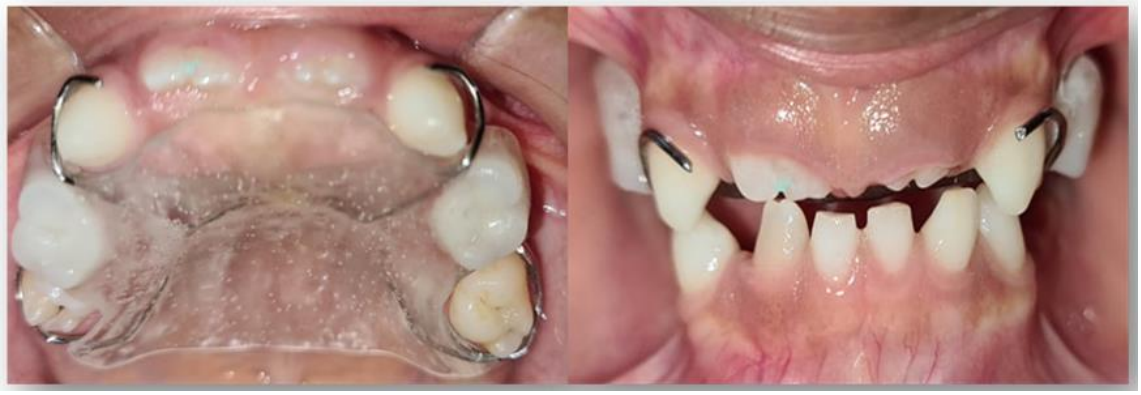

**Fig. 3: Aesthetic and functional space maintainer**

## CASE TWO

A 5-year-old girl diagnosed with IP disease, presented to the paediatric dentistry department ever cited for a dental check-up. She had no history of consanguineous marriage, and she was born at term. No ophthalmological anomalies, non-neurological signs were duty to report (Fig. 4).

**Fig. 4: A 5-year-old girl diagnosed with IP disease**

The general physical examination revealed asymptomatic skin changes on the trunk and the extremities. These linear hypochromatic lesions on the locations previously affected by vesiculobullous lesions as also known as “lines of Bloch Bulzburger” are corresponding to the fourth stage of Bloch Sulzberger (Fig. 5).

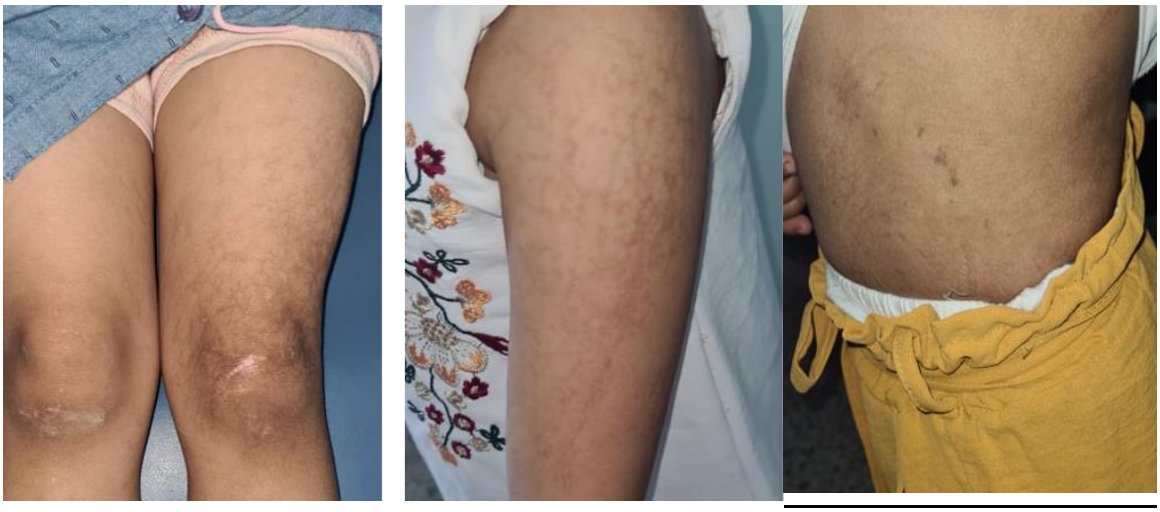

**Fig. 5: Bloch-Sulzberger lines on the trunk and extremities**

Intra oral examination showed subtitle signs; normal arch shape, most deciduous teeth were present with only one absent teeth# 85 (Fig. 6a, b). The# 51 and the #61 had superficial caries lesions. A retro-alveolar x-ray showed that the germ of the 45 was present (Fig. 6c).

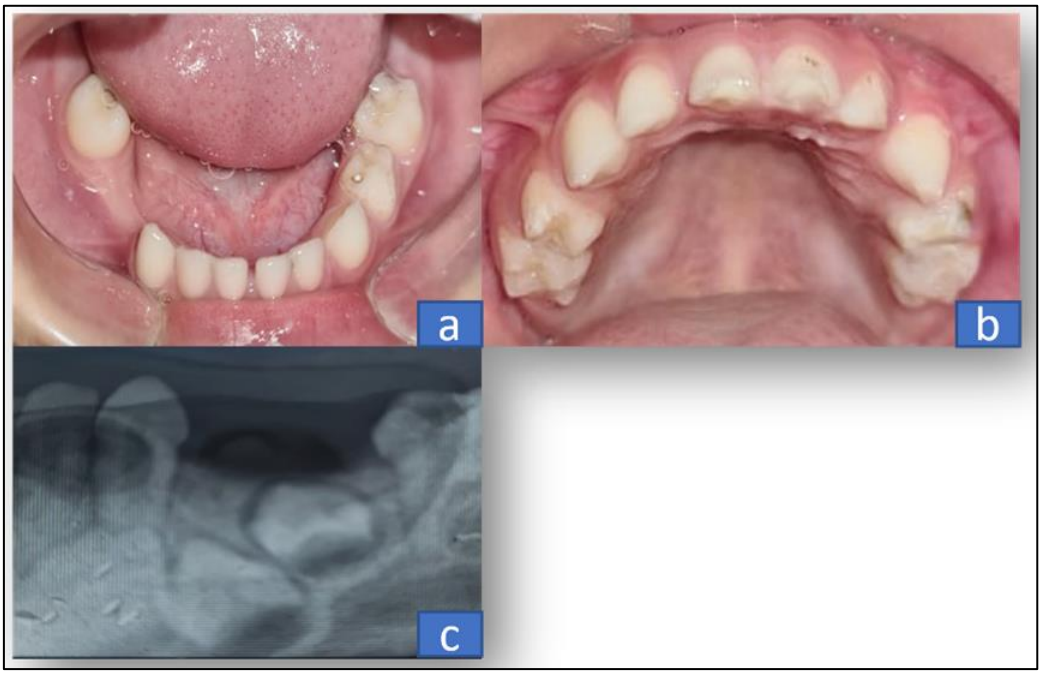


**Fig. 6: Dental features**

Our treatment plan was maintaining the space left by the 85 thus allowing the subsequent eruption of the #45. Caries curettage was done and check-up appointments have been set (Fig. 7).

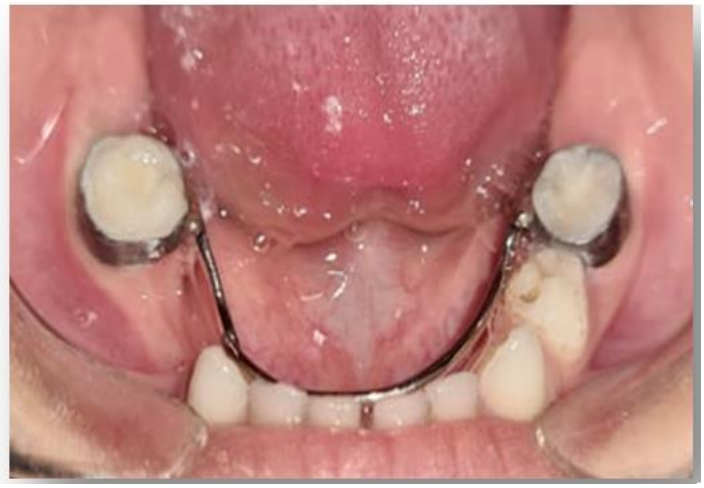

**Fig. 7: Bilateral space maintainer**

## DISCUSSION

Incontinencia Pigmenti is a rare syndrome that belongs to genodermatosis diseases with different other changes. It is caused by a mutation on the NEMO gene, located on the q28 portion of X chromosome [1].

Skin alterations are the main characteristic of IP. According to the literature, these manifestations can change and even disappear over time, they evolve through four stages: The first stage is named vesicular or vesicular-bullous, appear at birth or during the first two months and can last from weeks to months. The second one is when linear verrucous hyperkeratotic plaques appear. Next the hyperpigmentation stage occurs with brown blashko lines which are slowly fading. The last stage is the hypopigmentation [3, 4, 12].

In up to 80% of the cases of incontinentia pigmenti there are associated extracutaneous manifestations [4, 5].

Dental, eye, central nervous system and bone may be affected. Oral abnormalities are permanent and considered as the most frequent extra cutaneous signs. The second most frequent affected area is ophthalmological [3, 6, 12].

Thus, oro-facial findings are sufficient for detecting the presence of IP gene and identify other affected individuals [2, 6].

In some published results in the literature of IP patient series with dental anomalies, the proportion of investigated IP patients with dental and oral anomalies was high: between 80% and 90% [7, 8].

We illustrate the results of a brief literature review, conducted in the following Databases "Pubmed" and "Google Scholar", for the period 2011to 2021 (table 1).

The most relevant dental findings associated to the IP disease through this review were the congenital absence of six or more teeth, early dental loss, and malformed teeth [10]. These findings were in accordance with the first case report but were absent in our second case that had only one absent primary tooth and no crown abnormalities (table2).

More than 50% of clinical cases described in the literature review represented high arch palate and cleft palate opposed to our two case reports with no oral manifestations but only dental abnormalities [9].

The literature showed that the presence of symptoms other than skin changes, and especially dental disturbances, is important if dermatological signs are subtle and vice versa, which is in accordance with both our patients clinical expression [2] (table2).

Bearing in mind that there is correspondence between IKBKG mutations, that produce severe cellular disorders in skin and other affected tissues, mostly of ectodermal origin, and mutations of genes known to cause oral and dental anomalies [11].

Dental and oral anomalies are seldom life-threatening, but they may influence the quality of life of these patients. Some of these anomalies may cause feeding problems, and others could cause serious psychological problems for IP patients [11].

The currently available evidence in the literature regarding the dental findings in patients with Incontinencia Pigmenti is very limited. This study may help for the diagnosis of this disease through oro-facial signs and reinforce the recommendation that a child with IP should have a dental examination as soon as possible.

**Table 1: References that is eligible for the final phase of our selection process**

| Article author | Article type | Electronic database | Year of publicattion | Main findings |
|---|---|---|---|---|
| Poziomczyk CS and all | Clinical study | Pubmed ( the international society of dermatology ) | 2015 | ✓Dental findings in 70% of Patients: Conical teeth , Hypondontia , |
| Yaga YS and all | Case report | Pubmed ( departement of oral medecine and radiology ) | 2018 | ✓Hypodontia ( molars are missing ) ✓Peg shaped teeth ( anterior ) |
| Fernand DSM and all | Case report | Pubmed ( departement of oral medecine and radiology ) | 2017 | ✓High arch palate 50% of cases ✓Malocclusion ( crossbite ) (rare) ✓Agenesis 90% ✓Conical crowns 71% of cases ✓Enamel hypomineralisation (rare) |
| Senezana Menic and all | Systematic review | Google scholar ( clinical oral invest ) Added manually | 2012 | ✓Oro-facial findings in 54% of IP patients ✓Dental abnormalities: Hypodontia , delayed dentition , dental Shape anomalies ✓Oral manifestations: High arch palate (frequent), cleft palate (rare) |
| Senezana Menic and all | Clinical study | Pubmed ( clinical oral invest) Added manually | 2016 | ✓Oro-facial abnormalities in 80% of IP cases : teeth-shape anomalies, early dental loss , delayed eruption, anodontia, gothic palate |

**Table 2: Patients' Oro-facial findings**

| Patient | Age | Dermatologic findings | Oro-facial manifestations |
|---|---|---|---|
| Case 1 | 9 years old | Subtle signs | Flat palate, Conical teeth, Early dental loss, Oligodontia |
| Case 2 | 5 years old | Lines of Bloch Sulzberger | Suble signs: Only one absent tooth |

## CONCLUSION

Dermatologists are ideally placed to identify individuals with Bloch Sulzberger disease in early childhood because skin changes start nearly immediately after birth.

Dental and oral alterations, especially agenesis and conic teeth, are frequent elements in the manifestation of this syndrome.

As they are the second most frequent finding in IP, especially in cases without skin changes, the role of dentists is very important, not only in the early diagnosis of the syndrome but specially to improve the life quality of these patients.

Through a correct and accurate diagnosis, the need for multidisciplinary therapy can be established to promote the physical, social and emotional well-being of the patients.

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