

Bloch Sulzberger Syndrome - Rare Case Report in an 11 Year Male Child

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ABSTRACT

Introduction: Incontinentia pigmenti (IP) or Bloch–Sulzberger syndrome is a rare X-linked disorder that involves ectodermal tissues. Disease mainly affects the skin, eyes, hair, and central nervous system. Oral involvement is characterized by hypodontia of deciduous and permanent teeth.

Case presentation: We are reporting a very rare case of IP in an 11 year old boy having hyperpigmented lesions on head, neck and trunk. Intraoral examination revealed partially erupted tooth, multiple missing teeth and crowding of lower anterior teeth.

Management and prognosis: The patient was later referred to department of pedodontia for full mouth rehabilitation.

Conclusion: The information from this case report increase awareness among health care providers like general dentist, oral physicians, pediatricians and dermatologists.

Keywords: Bloch-sulzberger syndrome, Nemo gene, Hyperpigmented, Eosinophilia, Blaschko lines.

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INTRODUCTION

Incontinentia pigmenti (IP) is a rare familial disorder affecting the ectodermal syndrome was first described by Garrod et al., in the year 1906. It is an x-linked inherited genodermatosis caused by mutation in the NEMO gene (IKK-gamma), (NF-kappa B), genetic factor located in the q28 portion of this chromosome.^{1,2,3,4,5}

This disorder is known to be present in all races with a 37:1 female to male ratio. Inheritance may be dominant with lethality in males.^{1,3,4} Estimated prevalence is 0.2/100,000 in which changes of skin and skin appendages are usually combined with anomalies of other organs, teeth, eyes and central nervous system.⁷ The skin lesion usually has four stages: vesicubulous, verrucous, linear hyperpigmentation and atrophic lesions. The cutaneous lesions usually shows Blaschko lines.^{3,4,5,6,7} The inflammatory and hyperpigmented stages are the most consistent of these cutaneous lesions.^{4,5} The skin pigmentation gradually fades away leaving no traces in adulthood.^{6,8} Here we are reporting a rare case of IP in a 11 year old male patient.

CASE REPORT

An 11 year old male patient reported to the department of Oral Medicine and Radiology, Hitkarni Dental College and Hospital, with a chief complaint of unerupted maxillary anterior teeth (Figure 1). History of presenting illness revealed that patient was apparently alright one year ago and then noticed an unerupted teeth in his anterior teeth region. His medical history revealed that he had Tuberculosis disease 5 years ago and received the treatment for the same. Patient reported that he used to visit der-

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matologist regularly for his skin problems. He was diagnosed with seborrheic dermatitis with scalp abscess. He was on medication for the same. Past dental history revealed extraction of retained deciduous tooth (65) six months ago. Family history revealed that his mother too suffered from similar type of skin lesions.

On General physical examination hyperpigmentation with Blaschko lines was observed on his forehead, neck, upper trunk of his body (Figure 2, 3) right and left forearm. All his finger nails (figure 4) appeared brittle. Intraoral examination revealed par-

tially erupted 11, clinically missing 12,17,22,27,33,43 and crowding of lower anterior teeth. His mother reported presence of small vesicles in his early childhood, which were small initially and slowly increased in size, later they crusted and became hyperpigmented. Panoramic radiograph revealed impacted 12,13,23,24, right lateral incisor appeared to be placed distal to 14 (figure 5). Blood examination revealed eosinophilia (31%). With all these findings, we arrived at diagnosis of Incontinentia Pigmenti. The patient was later referred to department of pedodontia for full mouth rehabilitation.

DISCUSSION

Incontinentia Pigmenti (IP) or Bloch Sulzberger syndrome is a multisystem disease, also known as Occulo-Dento-Cerbro-Cutaneous syndrome or an Ecto and Mesodermal dysplastic syndrome.^{1,10} The term Incontinentia Pigmenti originates from the microscopic appearance of the lesion, i.e the presence of loose pigment in the basal layer of the epidermis.^{3,5} In 1954 Franceschetti and Jadassohn divided this disorder into two type: the Classic Incontinentia Pigmenti or Bloch Sulzberger variety which occurs almost exclusively in females and the Nageli type (or reticular) with a dominant mode of transmission which involves both sex without ocular malformations.¹⁰ It is an x-linked inherited genodermatosis caused by mutation in the NEMO gene (IKK-gamma)2, NF-kappa B genetic factor

(nuclear kappa B essential modulator), located in the q28 portion of this chromosome.^{1,2,3,4} However failure to identify a NEMO MUTATION does not rule out the diagnosis of Incontinentia Pigmenti. Affected females survive because of X chromosome dizygosity and negative selection of cells carrying the mutant X chromosome.^{9,10}

The skin lesions are the first clinical manifestations to be appeared and they are divided into 4 stages.²

Stage 1 Inflammatory or vesicular stage

It is characterized by the development of papules, vesicles and pustules on an erythematous base. The size of vesicle may vary from 1mm to 1cm or more. They are mainly seen on extremities but may also occur on the trunk, head and neck.^{2,3}

Stage 2 Verrucous Stage

Linearly arranged plaques and warty papules are seen on an erythematous base which follows the line of Blakschko. They are seen on extremities and trunk, but can also be observed on head and neck. Warty papules are seen in 70% of cases. Warty lesions are seen in the form of linear verrucous striae. They have predilection for palms and soles.^{2,3}

Stage 3 Hyperpigmented stage

Linear or whorled lesion along with pigmented lesions are seen .These lesions are seen in 90- 95 % of patients with IP. Commonly



Figure 1: Intraoral picture showing missing teeth



Figure 2: Blakschko lines was observed on forehead



Figure 3: Blakschko lines was observed on his neck, upper trunk



Figure 4: Brittle nails



Figure 5: Panoramic radiograph revealing impacted 12,13,23,24, right lateral incisor appeared to be placed distally

involves trunk and extremities, may also be seen in skin folds on the head and neck region. Hyper pigmentation may be seen on nipples, axillae and groins.^{2,3}

Stage 4 Atrophic or Hypopigmented Stage

Hypopigmentation, atrophy and absence of hair is most frequently seen on lower extremities. They develop during adolescence and persist into adulthood and may be permanent.^{2,3}

Extracutaneous manifestations include ocular lesions neurological manifestations and dental findings. The most frequent ocular abnormalities reported are blindness, strabismus, vitreous lens anomalies and optical nerve atrophy.² Neurological manifestations include seizure, mental retardation, intellectual impairment, hemiplegia, epilepsy, cerebellar ataxia, microcephaly, neonatal encephalopathy, encephalitis, neonatal and childhood stroke.^{2,7}

Dental findings like missing teeth, conoid teeth, additional cusps and delayed tooth eruption and hypodontia are frequently seen. Dental anomalies affect both primary as well as permanent dentition.² Histopathologic features varies according to the phases in which lesion are. Diagnosis was based on history, clinical examination, lab investigation, radiographic examination and histologic findings. Our case reported here matched the major and minor criteria proposed by Landy and Donnai (1993)⁶. However child's parents didn't agree for the biopsy of hyperpigmented lesions. Dermatologist opinion was also obtained before concluding the diagnosis of IP.

CONCLUSION

IP involves many organ systems, it needs a multidisciplinary approach. Genetic counseling of affected females is also important so that the probabilities are understood that offspring may be affected. This case is reported here for its rarity in a male child and classical appearance of hyperpigmented stage of Incontinentia Pigmenti.

The aim of this article was to increase awareness among health care providers like general dentist, oral physicians, pediatricians and dermatologists.

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