Incontinentia Pigmenti: A Case Report

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Abstract

Incontinentia pigmenti (IP) is a genodermatosis with an X-linked dominant mode of inheritance, characterized by ectodermal, mesodermal, neurological, ocular, and dental manifestations. The purpose of this case study was to report the oral and dental manifestations of an IP case in a Venezuelan pediatric patient. A 9 year-old Venezuelan girl was evaluated. She showed macular pigmented lesions in her face, trunk, back, legs, and fingers as well as abnormal hair distribution, alopecia on the vertex, and hypoplasia of eyebrows. During the dental examination, conical shaped-teeth and delayed dental eruption was evidenced. The microanalytical examination showed dentin without significant alterations in the mineralization except for hypermineralization in focal areas. In addition, a decrease in the enamel mineralization was observed. (Pediatr Dent 2006;28:54-57)

KEYWORDS: INCONTINENTIA, PIGMENTI, DENTAL, ORAL, CASE, REPORT, CHILD

Received June 16, 2005 Revision Accepted December 7, 2005

Incontinentia pigmenti (IP), also known as Bloch-Sulzberger syndrome, was first described by Garrod in 1906 and subsequently reported by Bloch in 1926 and Sulzberger in 1927. IP is a rare, X-linked dominant genodermatosis affecting 1 in 40,000 girls, being lethal in males. Mutations in the IKK-gamma gene—also called NEMO, which maps to Xq28—are involved in IP's pathogenesis.[1,2] This disorder is characterized by an erythematous eruption with linear vesiculation, most often localized on the back, skin, or extremities, and is present at birth or soon after. Subsequently, months after the disease's onset, verrucous growths are noted, predominantly on the extremities, resolving spontaneously or leaving an atrophic or a depigmented area. This may be limited to the extremities or to the trunk and head, but with a predominance of lesions on the arms and legs.

The final stage is characterized by macules, streaks, and splashes of brown to slate-gray pigmentation on the torso and less often on extremities. The pigmentation and other residues of skin manifestation gradually fade and are usually absent by adulthood.[3,4] Other features include central nervous system disorders, scalp abnormalities, nail dystrophy, and ocular and dental anomalies. Infants affected are generally not systemically compromised, despite eosinophilia and leukocytosis in the blood count.[3,4] An increased susceptibility to infections has also been reported.[5] There is no ethnic or geographic predominance, although most cases have been reported in Caucasians.[5] IP patients have a characteristic dentition and experience changes varying from marked hypodontia to delayed eruption and conical crowns on both dentitions.[6]

The purpose of this case study was to report the clinical, radiological, and microanalytical findings of a patient with incontinentia pigmenti.

Case description

A 9-year-old Venezuelan girl was seen at the Faculty of Dentistry, Central University of Venezuela, Caracas, Venezuela, for evaluation of her dental condition. The patient weighed 5.40 lbs and measured 12 inches at birth and gained weight slowly. A provisional clinical diagnosis of IP was made when the patient was 3 days old and confirmed histopathologically from a biopsy taken from her back. There was no family history of IP. The vesicular lesions over her extremities and body cleared up at 6 months of age. The patient had an abnormal hair distribution, "woolly hair," and normal nails. She was allergic to formula and was placed on soybean milk during infancy. She was hospitalized 3 times due to recurrent gastrointestinal infections.

During the physical examination at age 9, the patient showed pigmented macular lesions on her face, trunk, back, fingers and legs and her nipples were absent. The patient had alopecia on the crown of the head (Figure 1) with a scarred patch on the skin. Scant eyebrows and eyelashes

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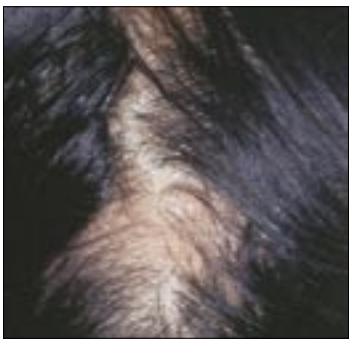

Figure 1. Alopecia located at apex of head of 9-year-old female with incontinentia pigmenti.

were noted (Figure 2). A follow-up examination was performed at age 9, at which time she weighed 45 lbs, measured 36 inches, and showed no other abnormalities.

At the dental examination, marked conical-shaped teeth were observed as well as delayed eruption of primary and permanent teeth. Multiple diastemas in all quadrants were noted. Mild chronic localized gingivitis at the permanent maxillary left central incisor was noticed, and there was no evidence of dental caries (Figure 3).

The radiographic examination showed that 14 permanent teeth were missing. Taurodontic pulps were also observed. In addition, both maxillary central incisors had enamel defects and were notched on their incisal edges. On the cephalometric radiograph, the patient presented a skeletal Class III jaw relationship and anterior crossbite. There was no need for dental treatment until the eruption of the permanent dentition started, although the anterior crossbite had been treated with functional appliances (Figure 4).

The patient's primary maxillary left central incisor was available for histological examination, and scanning electron microscopy (SEM) microanalysis was conducted. Five primary incisors were used as control teeth after extraction for orthodontic purposes. The affected and control teeth were sectioned longitudinally, parallel to the long axis, in 330-µm thick sections.

The tooth fragment was immersed in liquid nitrogen, freeze-dried, carbon-coated, and examined using a Philips XL30 scanning electron microscope (Philips, Eindhoven, Netherlands) equipped with an energy dispersive X-ray detector (EDAX International, Philips, Eindhoven, The Netherlands) equipped with an EDAX DX-4 microanalytical system. A total of 20 analyses each were conducted on enamel and dentin. Spectra were collected by pinpoint electron beam at X40,000. The peak-to-background (P/B) ratio method was used to measure the concentration.

Standard calcium and phosphorus salts were processed in an identical manner and used for quantitative analysis.[9,10] Morphological study specimens were gold coated after electron probe microanalysis (EPMA) and examined via SEM.[7,8]

The SEM microanalysis of enamel showed parallel prisms—with a variable diameter and irregularly rounded elements of variable sizes arranged, in some cases, in a beaded or ribbon-like pattern—disposed in the interprismatic areas. Some prisms revealed a filamentous configuration. (Figures 5 and 6). SEM images of dentin in teeth with IP showed dentin tubules with narrow lumens. Peritubular dentin was clearly visible. No sclerotic casts were seen in the tubular lumina.

In the present case, the concentrations were expressed as weight fractions: calcium=23.35±3.35 in enamel and 27.98±3.95 in dentin. The results for phosphorus were 14.21±1.94 in enamel and 15.05±1.77 in dentine. The control teeth weight fractions showed: (1) calcium=32.02 in enamel, 28.09 in dentin; and (2) phosphorus=18.90 in enamel, 16.73 in dentin.

Discussion

The clinical findings in this patient are consistent with those reported by others.[3,4,6] In the present case study, the child gained weight slowly, in contrast with a previous report from the literature.[6] Interestingly, the patient developed several infections that required hospital care, in accordance with previous studies by Gorlin and Anderson.[6]

The patient presented several dental manifestations including oligodontia, changes in tooth shape—mainly conical-shaped incisors, canines, and bicuspids—and delayed eruption affecting primary and permanent teeth in accordance with previous studies.[6] These reports also concluded that there is a characteristic dentition in IP, with a triad of delayed eruption, marked hypodontia, and conical crowns in both primary and permanent dentitions.[6,11]

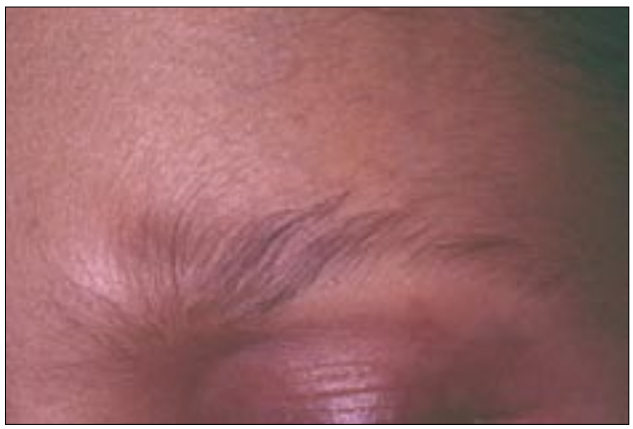

Figure 2. Hypoplasia of eyebrows and eyelashes.

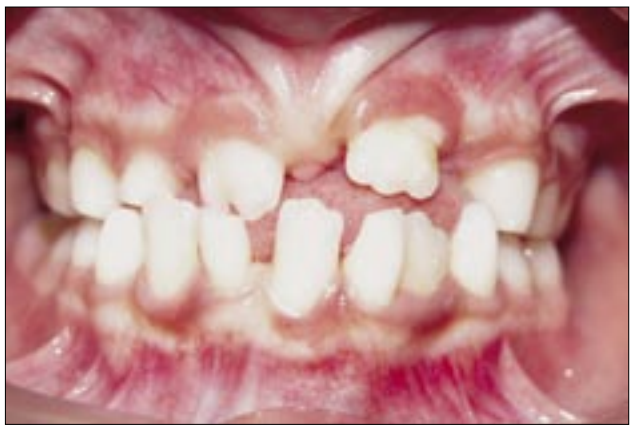

Figure 3. Dentition of 9-year-old female with incontinentia pigmenti.

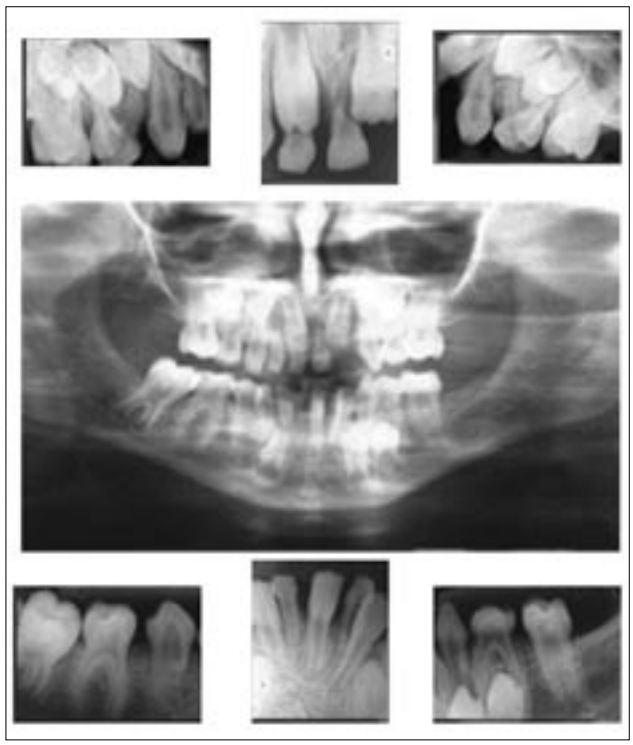

Figure 4. Panoramic and dental roentgenograms view showing multiple missing teeth in a patient with incontinentia pigmenti.

The radiographic examination showed numerous missing teeth, conical-shaped teeth, and taurodontism, similar to other authors' observations.[3,6,12,13] To the best of the authors' knowledge, no previous reports have emphasized the histological examination of IP-affected teeth. In the present case, SEM was conducted to describe the morphology and the microanalytical findings of a primary left central incisor available.

The prism structure's pattern revealed alteration in the nature and rate of ameloblast secretory activity during the development of the affected enamel.[14] These features have been previously described in normally developed human enamel as well as in amelogenesis imperfecta.[10,15] An immature morphological pattern was noted to persist in the enamel IP-affected patients.

Other features observed in IP teeth included the presence of noncrystalline-appearing, irregular, rounded formations located in the interprismatic areas. These formations with organic matter not removed during maturation might be normal unprocessed or structurally abnormal enamel protein.[16]

The microanalysis results showed a decrease in the enamel mineralization in IP when compared with the control group. The dentin, however, did not show significant alterations in the mineralization, similar to the dentinal changes observed in amelogenesis imperfecta.[10]

The differential diagnosis of IP includes patients affected by Franceschetti-Jadassohn syndrome, which is synonymous with Naegeli syndrome.[6] IP's dental features may also be confused with other congenital disorders such as syphilis and ectodermal dysplasia.[12] The relationship between eosinophils and skin blisters may be applied to dermatitis herpetiformis, erythema multiforme, erythema bullosum, pemphigus, and other bulluous diseases.[3]

References

1. Smahi A, Courtois G, Vabres P, et al. Genomic rearrangement in NEMO impairs NF-kappaB activation and is a cause of incontinentia pigmenti. The International Incontinentia Pigmenti (IP) Consortium. Nature 2000;25;405:466-472.
2. Scheuerle A. Male cases of incontinentia pigmenti: Incontinentia pigmenti: A case report and review. Am J Med Genet 1998; 77:201-218.
3. Carney RG, Carney RG JR. Incontinentia pigmenti. Arch Dermatol 1970;102:157-162.
4. Carney RG Jr. Incontinentia pigmenti A world statistical analysis. Arch Dermatol 1976;112:535-542.
5. Roberts JL, Morrow B, Vega-Rich CM, et al. Incontinentia pigmenti in a newborn male infant with DNA confirmation. Am J Med Genet 1998;75:159-163.

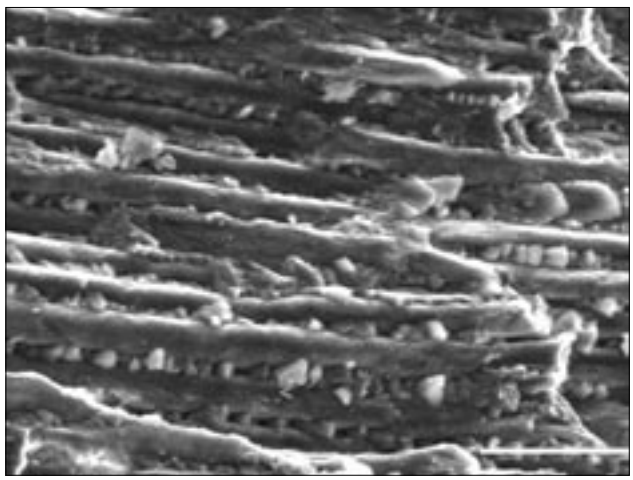

Figure 5. Scanning electron microscopy microanalysis of enamel (magnification ×1,500).

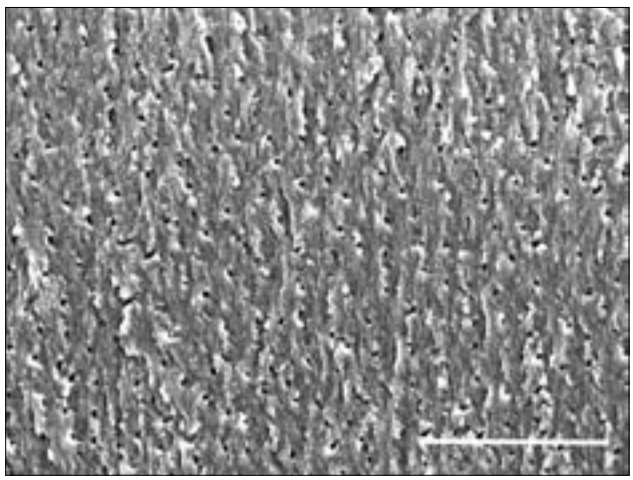

Figure 6. Scanning electron microscopy microanalysis of the dentine (magnification ×750).

6. Gorlin RJ, Anderson JA. The characteristic dentition of incontinentia pigmenti. J Pediatr 1960;57:78-85.
7. Statham PJ, Pawley JB. A new method for particle X-ray microanalysis on peak to background measurements. Scanning Electron Microsc 1978;1:469-478.
8. Small JA, Heinrich KFJ, Newbury DE, et al. Progress in the development of the peak-to-background method for the quantitative analysis of single particles with the electron probe. Scanning Electron Microsc 1979;II:807-816.
9. López-Escámez JA, Campos A. Standards for X-ray microanalysis of calcified structures. Scanning Electron Microsc 1994;8:171-185.
10. Sanchez-Quevedo MC, Ceballos G, Garcia JM, et al. Dentine structure and mineralization in hypocalcified amelogenesis imperfecta: A quantitative X-ray histochemical study. Oral Dis 2004;10:94-98.
11. Cho SY, Lee AC, Drummond BK. Surviving male with incontinentia pigmenti: A Incontinentia pigmenti: A case report. Int J Paediatr Dent 2004;14:69-72.
12. Macey-Dare LV, Goodman JR. Incontinentia pigmenti: Seven cases with dental manifestations. Int J Paediatr Dent 1999;9:293-297.
13. Russel D, Finn S. Incontinentia pigmenti (Bloch-Sulzberguer syndrome): A Incontinentia pigmenti: A case report with emphasis on dental manifestations. J. Dent Child 1967; 34:494-500.
14. Wright JT, Robinson C, Shore R. Characterization of the enamel ultrastructure and mineral content in hypoplastic amelogenesis imperfecta. Oral Surg Oral Med Oral Pathol 1991;72:594-601.
15. Risnes S, Radlanski RJ, Renz H. A scanning electron microscopic study of developing human deciduous enamel on the dependence of the outline of surface pits on the angle of observation. Arch Oral Biol 1998;43:111-115.
16. Wright JT, Butler WT. Alteration of enamel proteins in hypomaturation amelogenesis imperfecta. J Dent Res 1989;68:1328-1330.

Abstract of the Scientific Literature

The Effects of Fluoride Varnish on Demineralized Enamel

Fluoride varnish has been proven to inhibit dental caries, but little has been done to investigate its remineralization effects. The purpose of this study was to evaluate in vitro this property of a topical fluoride varnish when applied directly to a carious lesion vs being applied to the tooth surface surrounding the lesion.

Fifteen extracted human molar teeth, which had been kept hydrated, were sectioned mesiodistally—with one half serving as a control. The buccal surfaces were coated with a protective nail varnish, intentionally leaving a window of enamel 1x5 mm in size. Lesions were created by suspending the teeth in an artificial caries-producing medium. After sectioning through the area of each lesion, a quantitative analysis was performed. All teeth were then painted with a 5% NaF varnish: half of the sample size had the entire tooth painted, including the lesion; the other half had the tooth up to the lesion surface painted. All teeth were stored in an artificial saliva solution for 30 days and then requantitated. The results showed that remineralization did occur, with no significant differences noted between the 2 different application techniques. Both techniques showed effective remineralization.

Comments: This study showed that fluoride varnish remineralizes substrate enamel lesions. This is of considerable value to practitioners, who frequently see "white spot" lesions. The application of a fluoride varnish is a conservative, simple, cost-effective, and easy treatment approach to stop the demineralization process and initiate remineralization. We can easily examine patients on a frequent recall basis to check on the progress of these "white spots" and take appropriate action, if warranted. **GM**

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Casellano JB, Donly KJ. Potential remineralization of demineralized enamel after application of fluoride varnish. Am J Dent 2004;17:462-464.

39 references