Clinical features of incontinentia pigmenti with emphasis on dermatoglyphic findings

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Incontinentia pigmenti is a syndrome characterized by both systemic and oral manifestations. Presented here are the dental, clinical, radiological, genetic and dermatoglyphic findings of a 6 year old female case and her family members. The following features were apparent: oligodontia in maxillary and mandibular arches in both dentition, peg-shaped incisors and brown lessions on the body surface. No other problems were observed. The case had remarkable dermatoglyphic findings such as hypothenar loops associated with distally displaced axial triradii on both palms, reduced total finger and summed palmar a-b ridge-counts, decreasing plantar pattern intensity on the left sole. The other family members had similar dermatoglyphic characteristics. The paternal grand father, the father and the brother had eye defects .
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INTRODUCTION

Incontinentia pigmenti (IP) (Bloch-Sulzberger syndrome) is an uncommon hereditary disorder and is a disturbance of skin pigmentation sometimes associated with a variety of malformations of the eyes, heart, central nervous system and teeth.[1-3] The main features are vesicular, verraus and pigmented macular lesions of the skin.[4] Large pigmented macules arranged in patches or whorls may be present at birth or appear somewhat later. The macular lesions are brownish-gray to chocolate-brown in color and are distributed over the trunk and extremities.[5]

The condition have been first noticed by Garrod in 1906 but the name of the syndrome was introduced by Bloch Sulzberger in 1926.[6] Although comprehensive reviews arrived at an opinion that the syndrome was an X-linked dominant condition lethal in males the etiology of IP is not clear.[3] Person brought up for discussion the intriguing question of whether IP represents a failure of immune tolerance.

The disease usually appears at birth or shortly thereafter. The initial symptom consists of erythematous eruption with linear vesiculation, followed a few weeks or months later by verrucous growths and spots of pigmentation, mostly on the torso. These spots gradually fade and usually disappear by adulthood.[7] Several other associated deficiencies can be presented, including ocular injuries, cataract, optic atrophy, retinal dysfunction, blue sclera, nystagmus, strabismus and fibroplasia in the rear of lens, involvement of the central nervous system and osseous injuries with malformation of extremities.[2,3,6]

The characteristic dental defects in IP are partial anadontia and the presence of some peg-shaped teeth. The primary and/or the permanent teeth may be involved. Late eruption of teeth is also observed in some cases.[7-9]

Presented here is a patient with IP and her family members and additional compare the dental, clinical, genetic and dermatoglyphic findings with those cited in the literature and give some further evidence concerning with the etiology of the condition.

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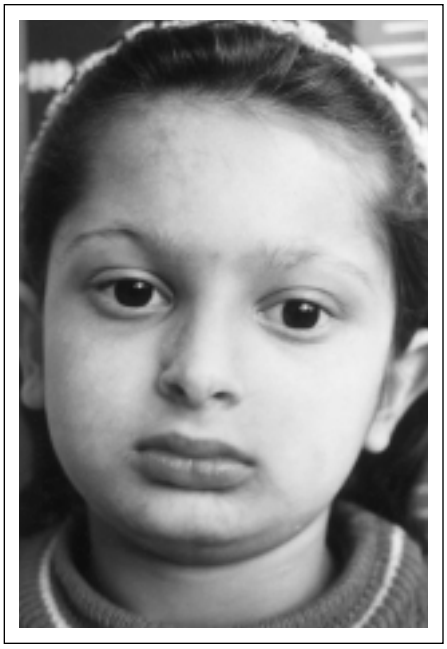

Figure 1. Frontal view of the patient with IP.

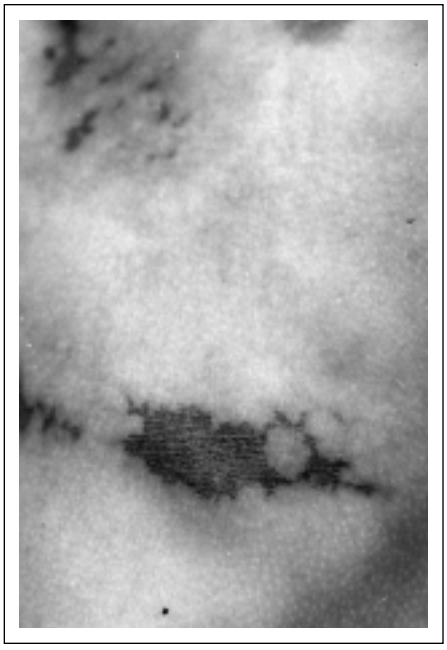

Figure 3. The brown skin lesion seen on the body.

Figure 2. Side view of the patient with IP.

CASE REPORT

A 6 year-old female patient (III-13) with IP attended to the Department of Pedodontics, Faculty of Dentistry, Marmara University for evaluation of partial anadontia. (Figures 1 and 2) A diagnosis of IP had been made at birth. She had cutaneus vesicular lessions. These changed to brown lesions after a few weeks. During physical examination, the presence of brown macular lesions on the body surface and some erythemaous lesions within white distinctive borders on the tongue were observed (Figures 3 and 4). She had developed normally and showed no other abnormalities. Intraoral (Figure 5) and radiographic examination (Figure 6) showed the absence of several teeth in both primary and permanent dentition (Figure 7). There was no active periodontal disease or dental caries.

The mother of the patient (II-14) had two miscarriages. The paternal grandfather (I-1), the father (II-6) and the brother (III-14) had ocular defects such as cataracts, myopia, rents in retina and ptosis, respectively. The ocular defects in question seemed to be not related to the IP. The family members excepting the patient had no cutaneous abnormalities. So, the patient was a sporadic case showing X-linked dominant trait (Figure 8).

The dermatoglyphic findings of the case with IP and her family members are shown in (Table 1) and (Figure 9). The case (III-13) had H and _ loops associated with t″ and t^b triradii on her both hands. She had a T^r configuration on her right hand as well. The total finger ridge-count (TRC) was lower than those of the female control cases. The mean for TRC's of the female control cases is x=133.6±2.89 (n=250). The father (II-6) had less TRC, on his left palm. The mean for TRC's of the male control cases is x=150.79±2.84 (n=250).The pattern intensities were lower on both soles as well. The summed palmar a-b ridge-count of the brother (III-14) decreased. The mean for summed palmar a-b ridge-counts of the control cases is x=80.19±0.54. He had a H^r loop associated with t″ axial triradius on his left palm and had H and _ loops combined with t″, t_ and t^b triradii on the right. The father (II-6) had a lower TRC and had a H loop on his left palm. The summed palmar a-b ridge-count of the mother (II-7) was also lower like her son. She had H, _ loops associated with t_ and t^b triradii on her left hand and a H loop associated with t_ axial triradius on the right. There was a T^r configuration on this palm as well. The hypothenar pattern types on the left palms of the case and her mother were identical. The brother

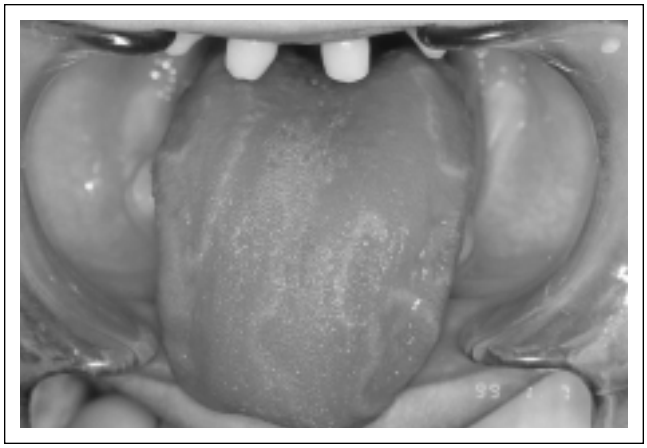

Figure 4. Tongue lesion noted.

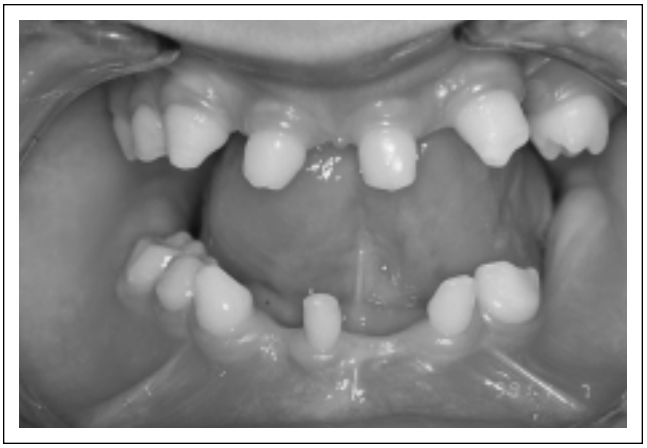

Figure 5. Intra-oral view of the patient.

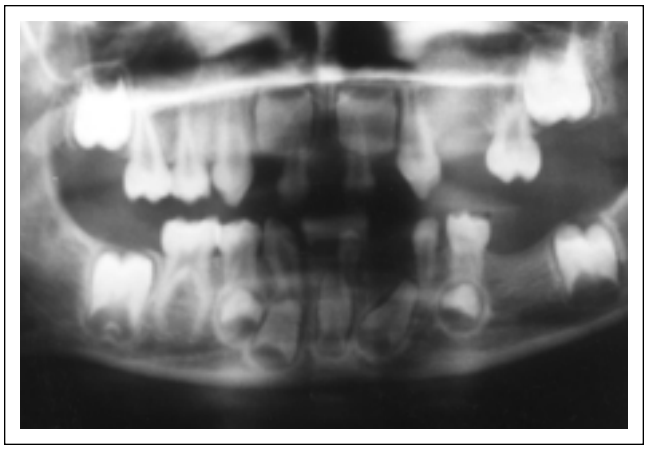

Figure 6. Panoramic radiograph showing absence teeth.

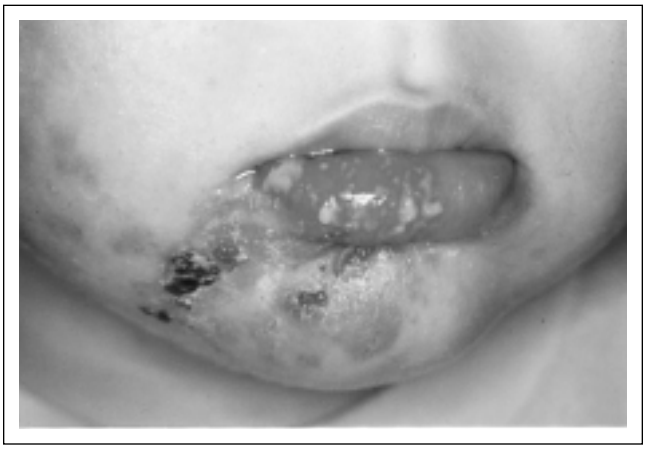

Figure 7. Lower face of the patient with IP.

(III-14) and the father (II-6) had simian lines on their left and both hands, respectively.

There was no need for specific treatment until permanent dentition completed. The patient will be followed-up periodically.

DISCUSSION

IP is basically a syndrome of genetic origin, presenting important buccal manifestations, and is most probably transmitted as a dominant character linked to the sex. It is primarily found in females, who comprise more than 95% of all reported cases.[3, 10]

Although comprehensive reviews conclude that the IP is an X-linked dominant disorder, essentially lethal in males and a family showed an autosomal dominant inheritance pattern of condition, the etiology of IP seemed to be heterogenous.

Lenz showed that affected males may be result of half chromatid mutations, although Hecht *et al*. disagreed.[11, 12] Cannizero and Hecht have mapped the gene for IP to band $X_p11.21$, although this has recently been challenged.[13] Polymorphic microsatellite markers,

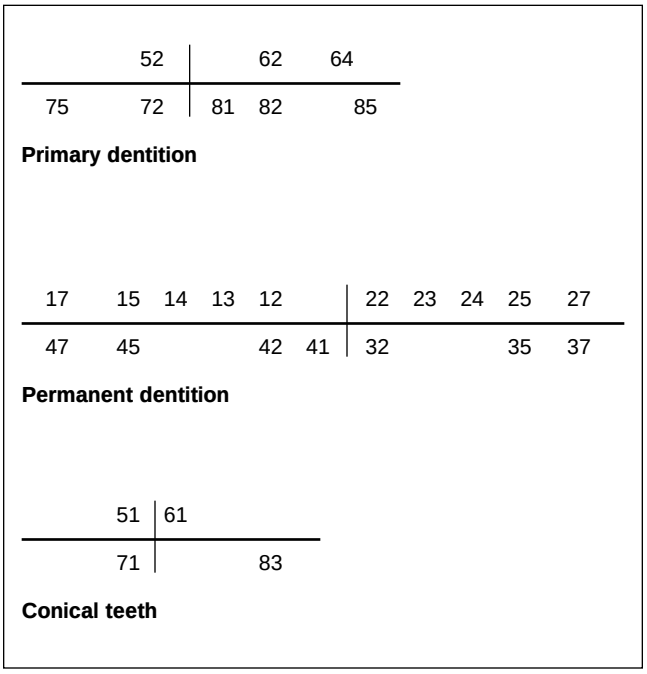

Figure 8. The pedigree of the patient is demonstrated.

Table 1. Digital type, palmar and plantar configurations, total finger and summed palmar a-b ridge-counts of the patient with IP and her family members.

	Fingers V IV III II I	TRC	Palmar formulae	Summed palmar	Plantar formulae a-b ridge-count
The patient III-13	L W W W R W R U U U U U	122	III H _ t" t t^b 4 (5") IV H $\hat{H}$ T^r t" t' t t^b 4 (4)	75	f z 3 I V f h z 3
The brother III-14	L U W W R W R U U W W W	220	I III Hr e t" t^b 4 (4) II III H $\hat{H}$ t' t t^b 5 (5')	57	I III p 4 I III p 4
The father	L U U U T U R U U U U U	96	IV H t" t 4 (1) IV t 4 (5")	89	I f 4 I III f p 4
The mother	L U U U W W R U U U R W	145	III IV H $\hat{H}$ t' t t^b 5 (5') III H T^u t' t t^b 4 (5')	64	I IV IV IV f p z 5 I II IV f p z z" 3

closely linked to the IP gene on the X chromosome, showed that each son inherited a different X chromosome from his mother.[14]

The abortions were observed in the mother of the present case, as well. In addition, another support concerning with affected embryo may come from a dermatoglyphic study. The affected persons had severe congenital anomalies and unusual dermatoglyphics. The patient and her family members in the study presented here showed characteristic dermatoglyphic features. In addition, there were great similarities between the pattern types of the patient and her brother, particularly between the configurational types of the patient and her mother.

Further, although the modes of the inheritance patterns of the dermatoglyphs are not clear yet, most of dermatoglyphic characteristics are hereditary and we might expect unusual dermatoglyphs only in our patient not in other family members, when there was an embryonic damage. So, unusual dermatoglyphic patterns observed in the individuals presented here and similarities between the dermatoglyphics of them seemed to be peculiar to this family.

As a result the patient presented here was a sporadic case of an X-linked dominant trait and the condition had occurred as a result of a *de novo* mutation. Because, both the parents and the brother showed no IP. The abortions most probably were males showing the trait because of the lethality of the condition in males.[3, 10]

Partial anodontia and peg-shaped or otherwise malformed teeth are the most common anomalies associated with IP and they were observed in our patient as well. There was no need for a particular treatment of the teeth of the patient until completion of the permanent dentition. The patient will be followed-up periodically.

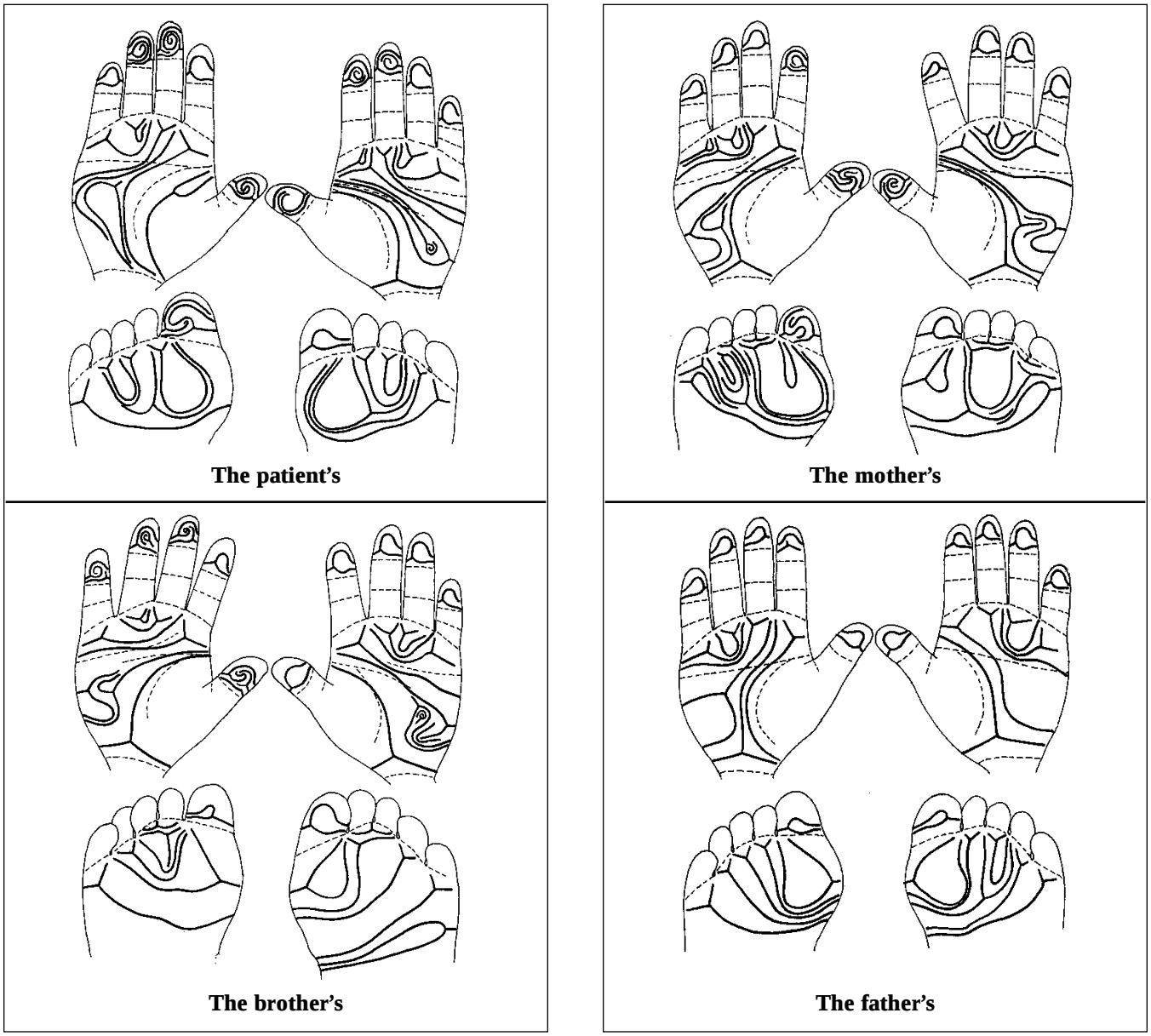

Figure 9. The dermatoglyphs of the patient and her family members is seen.

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