# Incontinentia Pigmenti in 22-month Old Afro-Caribbean Fraternal Twin Girls

## A Case Report

T Percival, K Smith

***ABSTRACT***

*Incontinentia pigmenti is a rare X-linked dominant condition characterized by cutaneous, neural, ocular and dental manifestations. The condition has mainly been reported in Caucasian females. The aim of this case report is to highlight the clinical presentation in Afro-Caribbean twin girls. The girls demonstrated abnormal hair distribution, pigmented limbs and torso, small conical or missing teeth with delayed dental eruption.*

**Keywords:** Caribbean, dental, incontinentia pigmenti

# Incontinencia Pigmentaria en Gemelas Afrocaribeñas de 22 Meses

## Un Reporte de Caso

T Percival, K Smith

**RESUMEN**

*La incontinencia pigmentaria es una rara condición dominante ligada al cromosoma X, caracterizada por manifestaciones cutáneas, nerviosas, oculares y dentales. La condición ha sido reportada principalmente en mujeres caucásicas. El objetivo de este reporte de caso es resaltar su presentación clínica en gemelas afrocaribeñas. Las chicas mostraron una distribución anormal del cabello, extremidades y torso pigmentados, pequeños dientes cónicos o ausentes con retraso en el brote dental.*

**Palabras claves:** Caribe, dental, incontinencia pigmentaria

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## INTRODUCTION

Incontinentia pigmenti is a rare X-linked disorder considered to be a syndrome of multisystem polydysplasias, commonly affecting the skin, eyes, central nervous system and teeth. It is caused by mutations in the NEMO (NF-κB essential modulator) gene, the protein product of which protects against tumour necrosis factor-α-induced apoptosis (1).

The prevalence of incontinentia pigmenti is unknown (2) but the frequency is estimated at 1:40 000 girls (3). The condition is usually lethal *in utero* for males. The majority of the reported cases worldwide occur in Caucasian females. More than half the documented cases had a positive family history for incontinentia pigmenti (2, 3).

Clinical manifestations of the disease vary widely, even among family members (4, 5). Dermatologic findings are most often the first observed sign of incontinentia pigmenti. The natural history of the disease describes cutaneous involvement as following four stages: stage 1 can be seen from birth to four months and often presents with erythema, vesicles and blisters in a linear pattern on the extremities. Stage 2 can occur from two weeks of life to six months and can manifest as hyperkeratotic papulae/plaques and verrucous lesions also on the extremities. Stage 3 is visible from 12 to 26 weeks and is characterized by hyperpigmentation. Stage 4 is entered thereafter and usually persists into adulthood as streaky hypopigmented or atrophic areas on the lower limbs that usually fade with time (2–6). All cutaneous lesions follow the Lines of Blaschko and may be 'S', 'V' or 'U' shaped patterns on the skin of the trunk and extremities.

Other findings of incontinentia pigmenti include an eosinophilia which is a common accompanying haematological finding in stage 1 of the condition; sparse/fine hair and in some cases vertex alopecia, finger or toe nail dystrophy *eg* pitting and ridging, nystagmus, strabismus or cataracts affecting the eyes and convulsive disorders, mental and/or

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motor retardation that arise from central nervous system involvement.

In more than 80% of patients, dental anomalies represent the most common non-cutaneous manifestation of incontinentia pigmenti. Hypodontia, microdontia (as pegged and conically shape teeth), delayed eruption and accessory cusps are frequently reported and both primary and permanent dentitions may be affected.

To the authors' knowledge, this is the first documented case of incontinentia pigmenti in Trinidad and Tobago and indeed the Caribbean. This case reports on 22-month old fraternal female twin girls of Afro-Caribbean descent presenting for dental consultation to a dental hospital in Trinidad.

Fig. 1: Vertex alopecia and visible scalp striations.

## CASE REPORT

Twenty-two-month old twins A and B presented to the Child Dental Health Clinic of the University of the West Indies Dental Hospital with their mother and grandmother who were concerned about the relatively few teeth present in the children's mouths at that age and also the abnormal shape of the existing teeth.

A comprehensive medical history revealed that the twin girls were diagnosed with incontinentia pigmenti soon after birth. The mother reported that pregnancy was uneventful and without complications. The girls were up-to-date with immunizations. There was no history of central nervous system problem, however, recurrent eye infection and epiphora (watering of eyes) were the ocular symptoms in twin B. The girls are still undergoing investigations on the cardiovascular system, as preliminary reports indicate some cardiac rhythmic anomalies (Table). There is no positive family history of this condition.

Table: Summary of twin's systemic involvement

| | | |
|---|---|---|
| Skin | Yes | Striated pigmentation noted on arms, back and perioral region; significant decrease in distribution and intensity reported over the past four months – Stage 3 and Stage 4 |
| Eyes | Yes | In Twin B, recurrent eye infections started at 10 months and epiphora was noted |
| Hair | Yes | Vertex alopecia. Significant growth of hair on head with time; sparse hair on arms and face |
| Nails | No | Appeared normal |
| Central nervous system | No | None reported |
| Teeth | Yes | Hypodontia and microdontia |
| Other | Under investigation for cardiovascular involvement | |

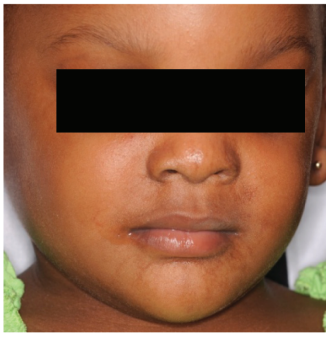

Fig. 2: Hyperpigmented striations on the perioral region and sparse eyebrows.

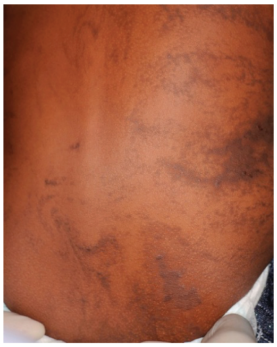

Fig. 3: Striations on back in linear whorls.

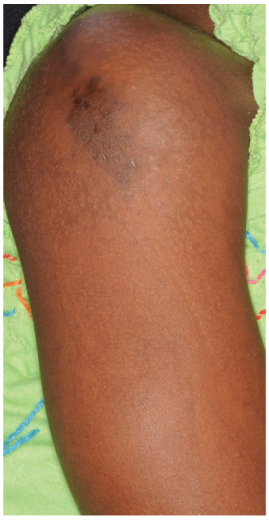

Fig. 4: Lesions on arm.

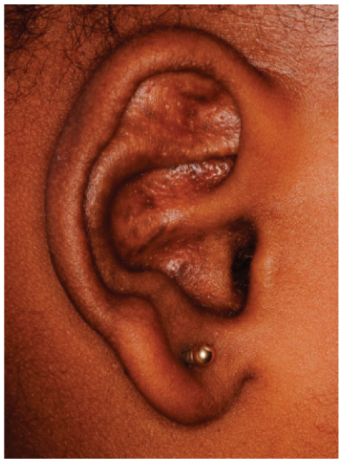

Fig. 5: Lesions on ear.

Upon examination, striated pigmentation was noted on the perioral region, the ears, hands, arms and the torso and legs. The girls had very thin eyebrows and sparse eyelashes. Vertex alopecia was noticeable (Figs. 1–6). The parent reported that there had been a significant (subjective) improvement in the hair growth and skin lesions over the past six months.

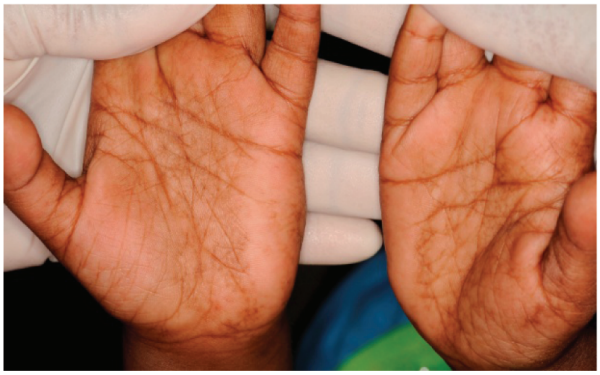

Fig. 6: Lesions on palms of hands.

Intraoral examination revealed notably thin alveolar ridges, especially in the posterior upper and lower regions. The teeth present are represented below:

Twin A:

| | |
|---|---|
| A | A |
| BA | A |

Twin A had a mesial incisal enamel defect on the upper left A.

Clinical photographs taken of Twin A are shown in Figs. 7 and 8.

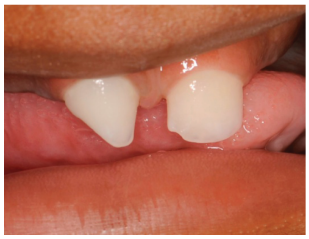

Fig. 7: Maxillary teeth – conical shaped upper right A and upper left A with mesial enamel defect.

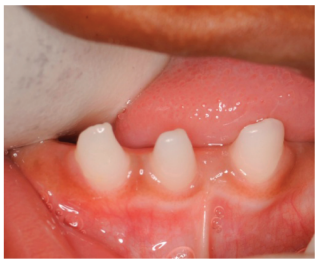

Fig. 8: Mandibular incisors, all conical in shape.

Twin B:

| | |
|---|---|
| A | A |
| A | |

Clinical photographs taken of Twin B are shown in Figs. 9 and 10.

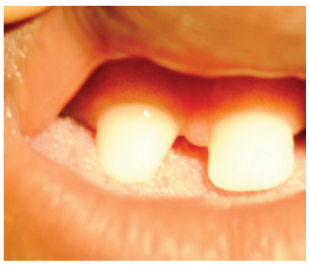

Fig. 9: Maxillary teeth – conical shaped upper right A and normal upper left A.

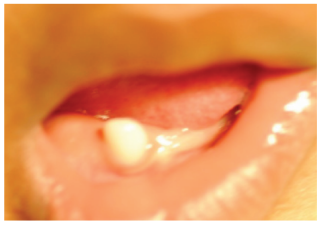

Fig. 10: Mandibular incisor.

No radiographic examination was undertaken. It has been well documented that there is an increased incidence of missing teeth in this condition. There was no justification for radiographic exposure of the twins at such a young age to confirm the absence of teeth.

## DISCUSSION

The diagnosis of incontinentia pigmenti relies heavily on the identification of the early cutaneous signs and an ability to distinguish the condition from other very similar conditions such as syphilis, ectodermal dysplasia, herpes simplex, impetigo, Langerhans cell histiocytosis, verrucous vulgaris and Goltz syndrome. (4, 6–8) Observation of the subtle differences in clinical and dental manifestations along with genetic testing can lead to a definitive diagnosis (6). However, in the local setting, ready access to this valuable tool may be a limiting factor. Thus, many cases may go undiagnosed or misdiagnosed.

The early diagnosis of incontinentia pigmenti and subsequent assessment of medical factors can impact on the long-term prognosis of each case. In the absence of ophthalmic and central nervous system involvement, the onus is now to monitor and manage the cutaneous and dental manifestations as may be required.

The significant absence and malformation of teeth can affect proper facial development, mastication, speech development, appearance and self-esteem in the growing child (4). Parents should be made aware of the common occurrence of late eruption and missing teeth and the effects on the developing dentition. It is also important to ensure that the nutritional needs are being met (4, 6). The practice of thorough oral hygiene methods and healthy eating habits should be encouraged to help preserve the existing dentition for as long as possible.

Although parents may have high expectations with respect to the provision of dentures so that appearance and esteem are improved, the various difficulties and challenges encountered in the provision of prostheses for such cases should be discussed. With parental support and an integrated and multidisciplinary approach, optimal results in aesthetics and function may be achieved (5, 6). It is proposed that review be undertaken every six months or sooner, if required. Any siblings should also be examined for dental anomalies. It may be prudent that genetic counselling be offered. Psychological support should also be available for the growing girls and their parents (6).

Incontinentia pigmenti has significant dental manifestations requiring long-term follow-up and a multi-disciplinary approach to the management of afflicted patients to provide a functional and aesthetic dentition. We acknowledge that the availability of genetic testing to confirm the diagnoses of incontinentia pigmenti may lead to significant under-reporting of the condition in Trinidad and Tobago.

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