INCONTINENTIA PIGMENTI: A REPORT AND REVIEW

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Incontinentia pigmenti is a rare cutaneous disease, probably developmental in origin, characterized by defects of the skin and its accessory structures, ocular and central nervous defects, osseous deformities, and other mesodermal abnormalities.[1,2,7,17,18] Its occurrence is sufficiently rare to warrant a report of a case seen recently.

CASE REPORT

The patient was a 4-weeks-old Coloured female. The mother insists that the baby's skin had been abnormally pigmented in various patterns on the trunk and limbs and under the chin since birth, in association with blisters on the hands and feet during the first 3 weeks of life. The blisters had dried up, leaving hard warty patches on the backs of the hands and the

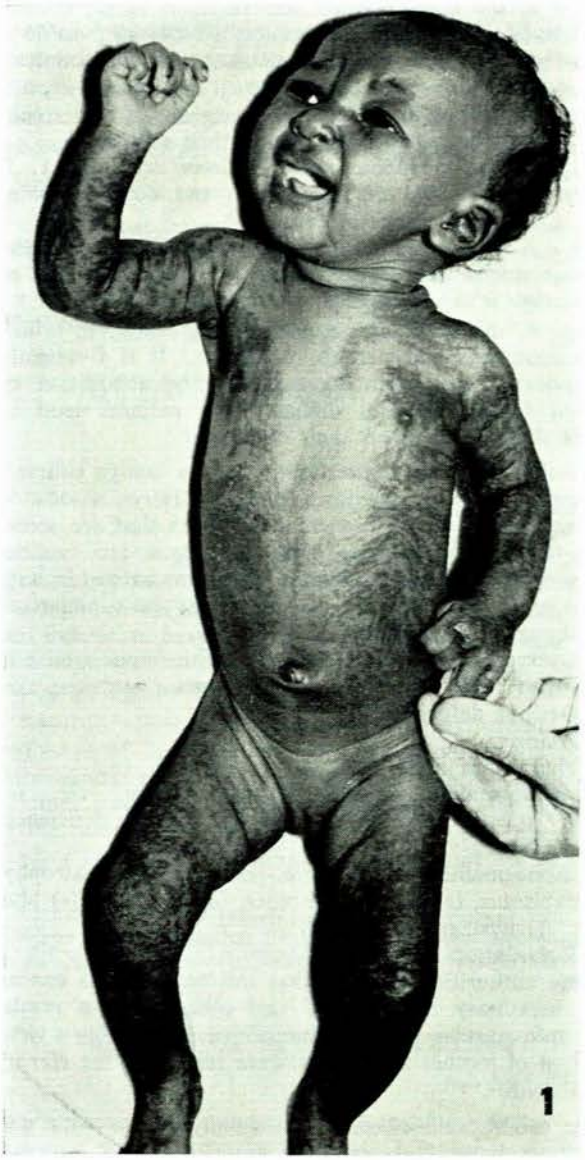

Fig. 1. Showing general distribution of skin pigmentation plus verrucous lesions over knuckles.

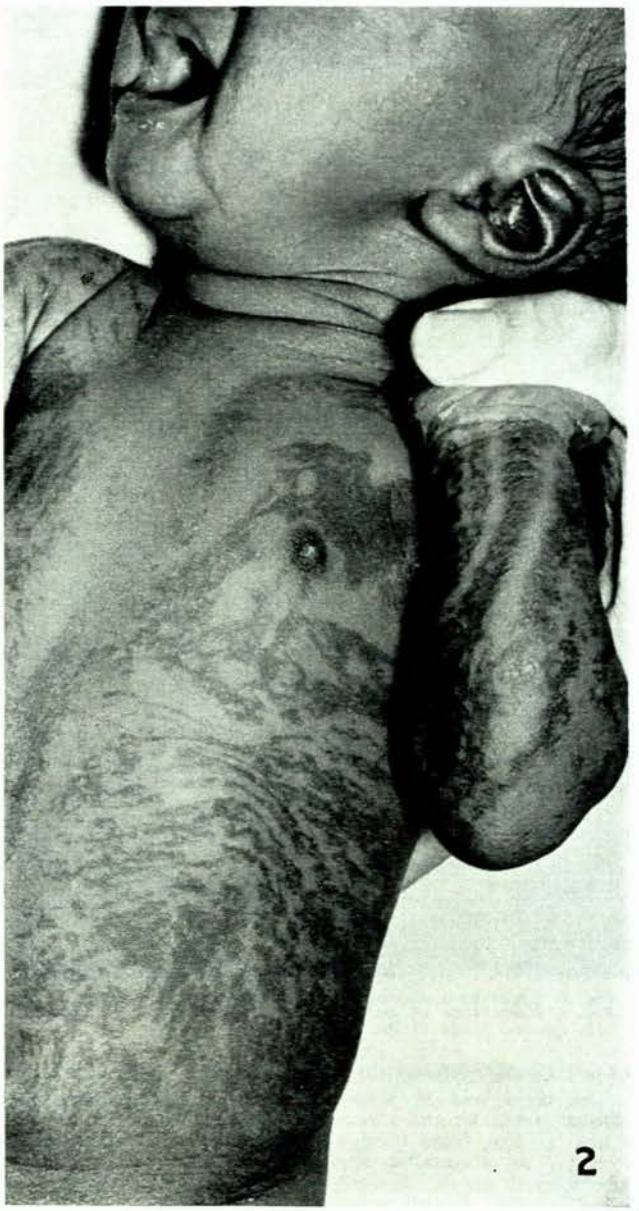

Fig. 2. A close-up view of the patient.

soles of the feet. Examination revealed a skin pigmentation (Fig. 1 and 2) on the limbs, hands and feet, abdomen and neck, and under the chin. This can best be described in Andrews' words[1] as follows: '. . . bizzare pigmented macules of greyish, chocolate-brown colour. They are irregularly shaped splashes, veins, striae, whorls, polyangular flecks, spidery and fountain-spray patterns, without systemization or order. The designs do not follow the lines of cleavage or distribution of nerves, nor the limitation of the median line'. There were verrucous lesions over the knuckles of both hands and under the soles of the feet. The skin over the legs and arms was thickened and not pliable. There were no areas of alopecia, defects of nails, oral leukoplakia, or blockage of the naso-lachrymal ducts. The result of ophthalmological examination was as follows: No nystagmus, eyes white and quiet, cornea clear, pupil reactions full, media clear, fundi appeared normal. On lumbar puncture no abnormality detected.

Family History

According to the grandmother, the mother of this child was born with a lot of blisters and 'sores' all over the body, which when they healed left a similar pigmentation of the skin. This was not as marked as this baby's and disappeared about the age of 7 years.

On examination of the mother, no areas of pigmentation were found. However, the teeth showed a state of hypodontia. Altogether 9 upper teeth and 9 lower teeth are present, and the mother and grandmother assure me that only one permanent tooth has been extracted (Figs. 3 and 4). There is no evidence of ocular changes, pseudopelade, disturbed sweating pattern or palmar or plantar hyperkeratosis in the mother.

The mother has had no abortions, nor any illness while pregnant with this child.

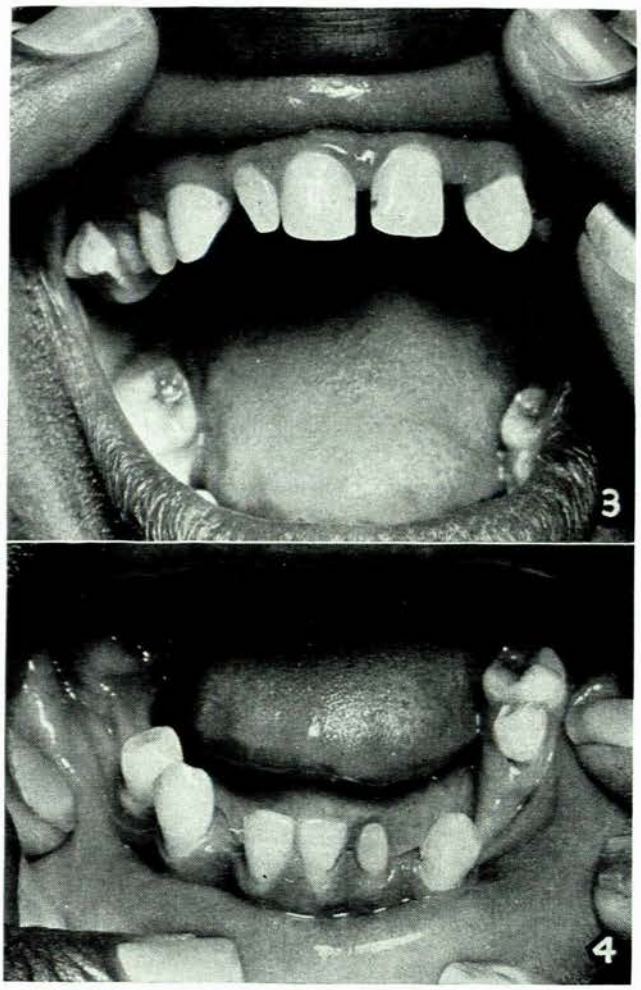

Fig. 3. Upper teeth of mother. She has had no extractions.
Fig. 4. Lower teeth of mother. Only one tooth has been extracted. Note peculiar shape of the incisor.

Progress and Postmortem findings

The child was in hospital for lobar pneumonia, which resisted treatment and was complicated by the development of a lung abscess, from which she died. Postmortem examination revealed no congenital abnormalities apart from the skin. The adrenal glands were normal and microscopically did not show any evidence of aplasia.

The skin on histological examination shows pigment in the superficial keratin layer (which seems somewhat thickened), in the basal layer, and in corial melanophores. No inflammatory features or irregularities of pigmentation in the basal layer are observed, and a granular layer is present. Occasional cells in the basal layer have pyknotic nuclei and pink cytoplasm, but their significance is not clear. More striking but also unexplained is the density of corial and dermal collagenous tissues. (Report by Dr. J. A. H. Campbell, of the Department of Pathology.)

DISCUSSION

This condition was first described by Bloch in 1926, and by Sulzberger in 1928.[10] (The name incontinentia pigmenti has also been applied to a condition that was described by Naegeli, but is entirely unrelated.) Incontinentia pigmenti of Bloch and Sulzberger[6] is usually familial and occurs almost entirely in females.[7,10] The cutaneous signs are present at birth[2,15,16] or appear soon afterwards and progress through four overlapping stages:[6,17]

Stage 1, the erythematous and vesicular stage, usually begins soon after birth.[2,10] It occurs in from 30% to 50% of the patients, and may be mistaken for epidermolysis bullosa. The lesions are accentuated on the flexural aspects of the extremities and the lateral aspects of the trunk. They persist for several months and then subside.

Stage 2, the verrucous stage, follows on stage 1. It occurs in about 30% of the patients and slowly subsides after a period of weeks or months.

Stage 3, the pigmentary stage, usually follows on the previous stages and begins during the first few years of life, though it is sometimes present from birth.

Stage 4, the stage of pigmentary regression, in which the lesions fade and perhaps disappear.[5] It is frequently overlooked. This fourth stage makes the acquisition of detailed family histories difficult, for patients tend to 'forget skin blemishes of their childhood'.[7]

The skin lesions, then, ordinarily take a benign course.[13] However, they are sometimes, but not always, associated with ectodermal and mesenchymal defects that are sometimes familial in incidence, although only a few families are known in which the complete syndrome existed in both parent and child. The abnormalities may be summarized as follows (Mendelsohn[8]):

1. Delayed or imperfect dentition, with hypodontia and malformed chisel-shaped or conic incisors.
2. Osseous deformities.
3. Alopecia.
4. Nail dystrophy.
5. Abnormalities of the nervous system,[3] viz.: (*a*) spastic paresis, (*b*) convulsions, (*c*) mental deficiency, (*d*) microcephaly.
6. Abnormalities of the eye, viz.: (*a*) optic atrophy, (*b*) strabismus, (*c*) corneal opacities, (*d*) cataracts, (*e*) blue sclera, (*f*) nystagmus.
7. Retardation of growth.

Some authorities consider that the syndrome is caused by a hereditary disturbance,[7] and others that it results from unfavourable intra-uterine influences, possibly a virus infection of mother and foetus, even related to the *Herpes simplex* virus.[4,9,12]

One might well imagine that children born with pigmentation have had some prenatal infection. Judging from the literature, it appears that the associated malformations are less common in those in whom an early inflammatory stage has occurred.

Seidlmayer,[15] who found adrenal aplasia at necropsy, mentioned the possibility of an androgen pigment disorder, possibly as part of general ectodermal dysplasia.[12] Whatever the aetiology, a striking characteristic is the multiplicity of the organs involved.[14] Histochemical studies in various stages of the skin manifestations reveal alterations in tyrosinase activity.

The condition was given the name incontinentia pigmenti because the histological picture was conceived as a disorder of the basal-cell layer, with leakage of melanin down to the cutis,[5,11,12] where it is taken up by connective-tissue cells (melanophores) and extracellularly. Secondary incontinentia pigmenti is seen in a number of skin diseases, notably in lichen planus and lupus erythematosus.

SUMMARY

A case of incontinentia pigmenti is described. The notable features are the early development of the pigmentation, and the absence of any obvious associated abnormalities, clinical or postmortem; and the probable occurrence of the condition in the mother, in whom it is associated with hypodontia.

I am grateful to Dr. J. G. Burger, Superintendent of Groote Schuur Hospital, for permission to publish this report, to Dr. R. Lang, head of the Department of Dermatology, for helpful criticism, and to Mr. B. Todt for the clinical photographs.

REFERENCES

1. Andrews, G. C. (1947): *Diseases of the Skin*, p. 586. Philadelphia: Saunders.
2. Carney, R. (1951): Arch. Derm. Syph. (Chic.), **64**, 126.
3. Dock, L. *et al.* (1956): Rev. neurol., **95**, 48.
4. Haber, H. (1952): Brit. J. Derm., **64**, 129.
5. Loveman, A. B. *et al.* (1952): J. Pediat., **40**, 442.
6. Marshall, J. (1960): *Diseases of the Skin*, p. 823. Edinburgh: Livingstone.
7. Marty, S. D. *et al.* (1958): Arch. Derm., **78**, 607.
8. Mendelsohn, R. *et al.* (1959): J. Dis. Child., **97**, 348.
9. Miller, J. L. (1950): Arch. Derm. Syph. (Chic.), **62**, 477.
10. Ormsby, O. S. and Montgomery, H. (1951): *Diseases of the Skin*, 7th ed., p. 708. Philadelphia: Lea & Febiger.
11. Oldfeldt, V. (1959): J. Pediat., **54**, 446.
12. Palmgren, B. (1962): Pediatrics, **29**, 295.
13. Roberts, A. (1958): Brit. Med. J., **1**, 1106.
14. Rubin, L. and Becker, S. W. (1956): Arch. Derm., **74**, 263.
15. Seidlmayer, H. (1962): *Op. cit.*[12]
16. Sobel, N. (1948): Arch. Derm. Syph. (Chic.), **58**, 467.
17. Ramos, J. and Silva, E. (1957): Dermatologica, **115**, 624.
18. Sulzberger, M. B. (1938): Arch. Derm. Syph. (Chic.), **38**, 57.
19. *Idem* (1928): Arch. Derm. et Syph., **18**, 891.