Incontinentia Pigmenti

Report of a Family


Incontinentia pigmenti usually begins in early childhood as a series of linear and grouped vesicles on an erythematous base, which, when ruptured, tend to become impetiginized. These may disappear and recur for weeks or months, gradually giving way to an intermediate temporary stage of linear verrucose lesions, or the vesicles may lead directly to the third or pigment stage, characterized by a reticulated pattern of pigmented macules, whorls, lines, and patches. Many years later, in some patients, a final stage may occur in which the lesions fade, leaving either no blemishes or slightly atrophic, slightly depigmented areas to mark the sites. Many cases are associated with mild to severe ectodermal and mesodermal defects.2,5,6,11

Since the literature, pathogenesis, and characteristics of the disease have been surveyed in detail by previous papers on the subject,2,5,6,9,11 only some of the more pertinent cases will be reviewed here.

Under the diagnosis of systematized nevus, the first two cases (identical twins) were presented by Bardach 1 in 1925. Two years later Naegeli 7 reported three cases under the name of chromatophore nevus; the lesions were found in a father and his two daughters. A third sibling was diagnosed as having hydroa vacciniforme.

In 1938 Sulzberger 11 reported a typical case in a 19-year-old woman whose family presented three generations of developmental defects in the female members only. A family tree was included, and attention was focused on the familial and hereditary aspects of the disease. Only one member had skin lesions.

Two cases, a father and a daughter, both with typical skin lesions, were described by Sobel 10 in 1948. This was the first report since Naegeli’s 7 cases in which cutaneous lesions appeared in more than one generation.

In 1952 Findlay 4 reported a typical case in a young girl whose mother had “a band of reticulated pigmentation over the right shoulder and right side of the neck coming over the middle of the clavicle to the sternum, diagnosed as systematized ephelis.” A maternal aunt had similar freckles. This constituted the third report of cutaneous lesions in more than one generation and the first report of a mother-daughter relationship.

Finally, in 1955, Philpott, Woodburne, and Philpott 8 described four cases, two pairs each of mother and daughter, with typical lesions.

The purpose of this paper is to present the case histories of what we believe to be a family with typical skin lesions in the female members for three generations.

Reports of Family Members *

CASE 1A.—J. S. T., 54-year-old man, no known lesions. He was not available for examination. History from G. M. T.

CASE 1B.—G. M. T., 42-year-old woman. She gave a vague history of skin lesions in infancy. Unfortunately, her parents are dead, and hospital records do not extend back into her childhood. At present she has two small, irregular, reticulated spots.

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* Numbers refer to the generation, and letters, to the members of each generation.
pigmented, 2×2 cm. macules on the left lower back.

Case 2A.—J. L. T., aged 24, daughter of G. M. T. She was first seen at the Indianapolis General Hospital Pediatrics Clinic in December, 1934, with a complaint of “broken out.” At that time she was 11 months old and had presented a history of vesicles and verrucous lesions for some time. Subsequently the lesions disappeared, only to be replaced by reticulated hyperpigmented lesions on both sides of her body. At present she resides in Michigan and is not available for reexamination. History is from Indianapolis General Hospital records.

Case 2B.—E. T., aged 21, son. No lesions reported.

Case 2C.—C. T., aged 19, son. No lesions reported. Subjects 2B and 2C are in the armed forces and are not available for examination. History is from G. M. T.

Case 2D.—M. T., aged 14, son. There is no history of vesicles or verrucous lesions, but he presents two lightly pigmented macules on the left chest. The patient has had Jacksonian seizures since birth and a right rotoscoliosis.

Case 2E.—D. T., aged 8, daughter. The patient was born at home in January, 1950, after an uneventful gestation and delivery. The child appeared normal until the fourth day, when the mother noted numerous vesicles on the arms, trunk, and legs. Seen by the Indianapolis General Hospital Pediatric Clinic, she was treated with penicillin ointment for six to eight weeks, whereupon the vesicles subsided, leaving diffuse pigmented lesions. She was first seen by the Dermatology Clinic in April, 1950, at which time she presented slightly raised, hyperpigmented, linear and reticuliform lesions of the arms, legs, chest, and abdomen. She also had flat slate-gray lesions on the back. Histopathological sections showed only several foci of heavily laden melanophores in the upper corium and papillae. She was presented to several authorities at a special clinic in May, 1950, and a diagnosis of “linear generalized nevus” was made. After the discovery of incontinentia pigmenti in two other members (Cases 2F and 3A) of her family, the patient was reexamined in the Dermatology Clinic in March, 1957. At this time she presented reticulated, lightly pigmented, and atrophic lesions across both shoulders and descending down onto...
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her arms (Fig. 1). Her mother states that these lesions have been steadily fading since the age of 2. No developmental defects have been found. Following review of the 1950 biopsy specimen, a diagnosis of incontinentia pigmenti was made.

Case 2F.—M. E. T., aged 17, daughter. The patient was first seen by the dermatology department on Nov. 15, 1956, four days after the delivery of her first child. The department was asked to see the neonate because of vesicular and crusted lesions on the back and extremities. In the course of examining the infant, the mother was also examined, revealing pigmented lesions which had been present all of her life.

The patient was born on May 15, 1940. On the following day redness of the skin was noted, and on May 17 a vesicle was found on the left leg and another on the right arm. By the following day, the patient had a generalized vesicular eruption, except for the scalp, palms, and soles. The lesions were described as "small, superficial vesicles, which form in groups and close together, many of which are confluent. These vesicles frequently occur in linear groups, especially on the extremities." Impressions were bullous impetigo and generalized dermatophytosis. After local therapy with aluminum acetate solution (Burrow's solution) and thimerosal (Merthiolate) tincture, many of the vesicles cleared or dried up "leaving pigmented areas." There was no real improvement, however, and the vesicles continued to appear. Finally, since the patient was afebrile and steadily gaining weight, in spite of the cutaneous lesions, she was discharged to the pediatric clinic in August, 1940. Readmitted one week later, she was treated with 1% methyrosaniline chloride (gentian violet) for two weeks, without improvement, and was again discharged unchanged. In May, 1941, the patient was admitted for marasmus and measles. Though the hospital record does not mention a vesicular eruption, it was noted that "there are elevated, indurated areas of skin over the chest which appear to be keloids of rather massive extent." The patient did not return to Indianapolis General Hospital again until the birth of her first child. She has no recollection of any type of rash except the present one, which consists of a reticulated brown patch of pigment on the left side of her trunk, extending from axilla to hip (Fig. 2). Several small irregular patches dot both hips; these are clinically identical with the two macules found on her mother (Case 1B). Examination was otherwise negative.

Case 3A.—D. D. T., aged 15 months, grandchild of G. M. T. (Case 1B). The department first saw this patient on the fourth day of her life because of bullous and crusted lesions on the body since birth. Newer lesions on the abdomen and extremities were vesicular and on a slightly erythematous base. Crusted lesions were present on all parts of the body, and on the flanks, arms, and legs were linear and reticulated pigmented lesions. A clinical diagnosis of incontinentia pigmenti was made. An antibiotic

Fig. 2.—Case 2F.

Fig. 3.—Case 3A.
ointment was used to control superficial infection. When reexamined six weeks later, the infant presented reticulated, hyperpigmented lesions coursing in a linear arrangement on all four extremities (Fig. 3). A few vesicles were present at the wrists and knees, and minimal reticular pigmentation could be found in the flanks. Biopsy of a vesicle showed dermoepidermal junction separation, with an eosinophilic and polymorphonuclear leukocytic band-like infiltrate beneath. Interspersed in the infiltrate were several melanin-laden macrophages. In addition, extracellular melanin was scattered throughout the area, and there appeared to be a partial loss of pigment in the basilar layer of the involved site. Two weeks later, all lesions seemed to be fading, and by the time the child was 3 months of age no vesicular or verrucous lesions were found, but reticular pigmentation persisted on the trunk and extremities. The skin was otherwise smooth and soft. X-ray survey indicated that the spine, long bones, and jaw were normal. Dentition and growth of the child have been normal, and by the age of one year the leg lesions had become lighter in color, depressed, and somewhat atrophic in appearance.

CASE 4A.—Father of D. D. T. (Case 3A). Nothing is known about this person.

Summary and Comment

Incontinentia pigmenti is briefly described, and the literature pertaining to the cutaneous lesions in more than one generation is briefly reviewed.

A family tree is herein reported in which all the female members for three generations displayed the cutaneous lesions of incontinentia pigmenti. These cases tend to support the sex-limited mode of inheritance.

One male member of the second generation presented developmental defects without skin lesions. Since no defects were found or reported in any other members, it remains conjecture whether these defects are associated with incontinentia pigmenti or are mere coincidence.

The fourth stage of incontinentia pigmenti, that in which the lesions fade and perhaps disappear, is frequently overlooked. Two of the cases in this paper (Cases 2E and 3A) are at present in this stage, and two other patients (Cases 1B and 2F) have passed through this stage and their conditions have stabilized with minimal pigmentation. This fourth stage makes the acquisition of detailed family histories difficult, as patients tend to forget "skin blemishes" of their childhood.

This family tree points out the need for accurate description of skin lesions in hospital and clinic records. This entire family history was uncovered by backtracking after the birth of Patient 3A.

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REFERENCES

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