Incontinentia Pigmenti in Identical Twins with Separate Skin and Neurological Disorders

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Incontinentia pigmenti in female identical twins is reported. The first baby showed the typical pigmentation of incontinentia pigmenti, while the second baby had hydrocephalus (colpocephaly) without pigmentation. They were identical, with a rate of 99.9% in 18 blood-type studies. Virus was not detected and cytogenetic studies proved normal. Both showed peripheral eosinophilia. The individual expressions of Incontinentia pigmenti in these identical twins were separated into cutaneous lesions and lesion of the central nervous system (intra-uterine hydrocephalus). Cutaneous lesions developed after birth. Twins with Incontinentia pigmenti are extemely rare and in this family showed different expressions of this disease in space and time. ***Key words: Bloch-Sulzberger syndrome.***

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Incontinentia pigmenti is an X-linked dominantly inherited disorder. It usually shows typical pigmentation, and in about 80% of cases it is associated with various congenital abnormalities such as neurological and ocular disorders and anomalies of dentition and bone (1). The first case of incontinentia pigmenti was reported by Garrod (2) in 1906. Bloch (3) and Sulzberger (4) subsequently described the clinical syndrome. There are few published reports of Incontinentia pigmenti in twins, in which both twins showed typical pigmentation (5). In our case, individual expression was separated in each twin.

CASE REPORT

Female twins were born following cesarean section at 40 weeks of gestation because their mother showed hydroamnios and dyspnea. The mother was 24 years old and had been healthy until delivery. The twins did not suffer from perinatal asphyxia and there was no particular family history. The first baby weighed 2.7 kg at birth and had a normal head circumference. She showed blister formation on arms and legs one day after birth. On arms and legs, chest and abdomen, erythema and bullae tended to arrange in lines, and after a while, crusts and brown pigmentation appeared (Fig. 1). Apart from pigmentation, no abnormal signs were found. Analysis of her peripheral white cells showed 14% eosinophilia.

Skin biopsy on day 62 revealed parakeratosis, eosinophilic necrosis, and liquefaction in the epidermis, and lymphocytic infiltration and incontinence of pigments in the dermis. This was considered histopathologically to represent the second stage of incontinentia pigmenti.

The second of the twins babies weighed 3.3 kg at birth. She had a larger head, 38.5 cm (+3.4 SD) in circumference. Computerized tomography revealed colpocephaly (hydrocephalus with disproportional dilatation of the posterior horns) (Fig. 2). Cerebrospinal fluid examination was normal. She had no skin pigmentation. Apart from colpocephaly, she had no other minor or major anomalies,

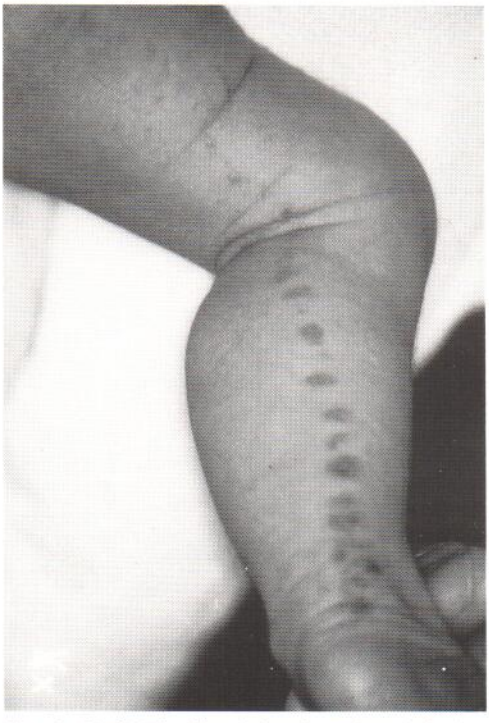

Fig. 1. (*Left*) The first baby showed typical pigmentation of incontinentia pigmenti on day 182.

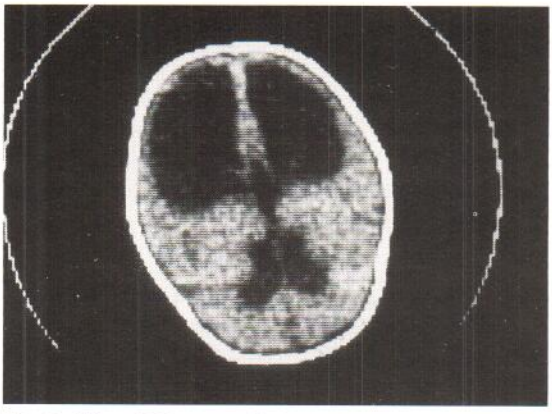

Fig. 2. (*Above*) The second baby had a larger head, without blister formation. Computerized tomography showed hydrocephalus.

and no abnormal neurological signs. She too showed eosinophilia (10%).

Viral examinations of both twins for Herpes simplex, Herpes zoster, Cytomegalovirus and Rubella proved negative. Cytogenetic studies showed 46,XX, normal chromosomes.

Their placenta was monochorionic and diamniotic at birth. Twin diagnosis was carried out with 18 blood types, ABO, MNSs, Rh, P, Duffy, Kidd, Diego, Xg, Hp, Tf, Gc, Pi, AcP, EsD, GPT, PGM, 6PGD, and GLO. The probability of monozygosity was 99.965%, and probability of dizygosity, 0.035%.

We observed these identical twins for 7 years from birth. Their physical and neurological development was normal. Pigmentation in the first baby had started to fade by 2 years of age.

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