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INCONTINENTIA PIGMENTI STAGE II

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Eight weeks old female child with linear verrucous lesions on extremities, eosinophilia and characteristic calcified dyskeratotic cells on histopathological examination confirming the diagnosis of second stage of incontinentia pigmenti is reported. Brief account of genetic counselling is given.

Key words: Incontinentia pigmenti.

Incontinentia pigmenti (IP) is rare. Only a few cases have been reported from India.[1] Although first described by Bloch and Sulzberger in 1926 and 1927 respectively, in a review of world literature Carney[2] found only 653 cases. The inheritance is sex linked dominant. IP is prenatally lethal to males, giving a sex ratio of 97:3 in favour of females. Most of the male cases are thought to be due to mutations.[2]

Clinically, the patient passes through three stages. The first stage is characterised by vesiculo-bullous lesions, commonly on the extremities, while the second stage also seen on the extremities is characterised by verrucous lesions. Following the verrucous stage, the highly characteristic pigmentary stage begins. The pigmentation is characterised by a bizarre distribution over the central portion of the trunk. a fourth stage has also been recently recorded. In this stage, hypopigmentation and atrophy occur in the same areas which were affected previously by hyperpigmentation.[2] Their presence may serve as a clue to the hereditary pattern and be significant for genetic counselling.[3]

Systemic manifestations are seen in a large number of patients. These include abnormalities of the hairs, eyes, central nervous system and the skeleton, making genetic counselling an important aspect of disease management.

We are reporting a case of IP with verrucous lesions on the extremities, this stage of the disease being the least commonly reported.

Case Report

A 1½ month-old female infant was brought for hyperpigmented verrucous lesions on her extremities which had appeared during the first week of life. There were no lesions at birth. These appeared first on the right lower extremity and gradually increased in size and number to involve all the extremities. Both the parents were normal. The patient was their first off-spring, delivered normally at full term. There was no history of consanguinity. In addition to the linear verrucous lesions on the extremities, a few macular pigmented lesions were also seen on the chest and abdomen.

Routine investigations revealed eosinophilia of 14% with an absolute eosinophil count of 1400/cmm. The total leucocyte count was 10000/cmm with neutrophils 28%, lymphocytes 56% and monocytes 2%. Other investigations were within normal limits. The mother's absolute eosinophil count was 1600/cmm.

Histopathological study revealed hyperkeratosis and irregular lobulated acanthosis. There were a large number of dyskeratotic cells. Some of the dyskeratotic cells were calcified. A few melanophages were seen in the dermis.

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Comments

In IP, the disease process starts in utero. The first stage, as in our case, may be totally absent. In such a case, the second stage may be mistaken for other linear verrucous lesions such as the verrucous epidermal nevus.

The eosinophilia, both in the patient and the mother as well as a few pigmented macules on the patient's chest were clues for the diagnosis of IP. The diagnosis was confirmed by histopathological studies. a large number of dyskeratotic cells present throughout the acanthotic lobulated epidermis with several calcified cells are diagnostic and distinctive features of IP.[4] Calcified individual dyskeratotic cells are probably seen only in IP.

Since more than 50% of patients develop serious ocular and central nervous system complications, a few guidelines as enumerated by Carney[2] are worth keeping in mind. They are: (1) For a woman with IP, at least one pregnancy in four will result in miscarriage. (2) Statistics indicate that half of her female offsprings will have IP and the daughter is likely to be more severely affected than her mother. (3) In a woman without IP, who has a child with IP, the likelihood of that condition being due to a spontaneous mutation is probably high and the risk of miscarriage or of bearing another daughter with IP is a little lower.

The advice given by us to the parents regarding the future pregnancy was as follows. In the future pregnancy, amniocentesis to determine the sex of the fetus should be done. If the sex is male, there is 50% chance of spontaneous abortion, while if the pregnancy continues, a healthy child is expected. If the sex is female, therapeutic abortion is indicated to avoid ocular and nervous system complications which are expected in 50% of the affected female off-springs.

References

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