SELF - ASSESSMENT PROGRAMME

A 10 day old female neonate reported with a few scattered bizarre scaly macules on the limbs, verrucous papules on the trunk and vesicular lesions arranged in linear streaks on the limbs, present since birth. The child was otherwise well and feeding normally. She was the second born to her nonconsanguinous parents after an uneventful gestation. The elder sibling — a 3 year old male — was healthy.

1. The most likely diagnosis in this case would be :

   a. Epidermolytic hyperkeratosis.
   b. Congenital syphilis.
   c. Incontinentia pigmenti.
   d. Pemphigus neonatorum.
   e. Urticaria pigmentosa.

2. What investigations would be of help ?

   a. Blood S.T.S.
   b. Skin biopsy.
   c. Culture of blister fluid.
   d. Tzanck smear.

   The VDRL was negative and blister fluid was sterile.

   The histological features were suggestive of incontinentia pigmenti.

3. What would these features be ?

   a. Intraepidermal vesicles filled with eosinophils.
   b. Sub-epidermal bulla with eosniophils.
   c. Whorling of prickle cells with central dyskeratotic cells.
   d. Epidermis practically nonpigmented with melanophages in upper dermis.
   e. Extensive Basal cell degeneration with pigment incontinence.

4. What other features would you look for ?

   a. Ocular changes.
   b. Dental defects.

c. Central nervous system disorders.

d. Skeletal abnormalities.

5. The genetic defect in this condition is an :

a. Autosomal dominant trait.

b. Autosomal recessive trait.

c. X-linked dominant trait.

d. X-linked recessive trait.

6. What changes are likely to follow in this patient ?

a. Pigmentation will gradually fade in adolescence and may completely disappear by 20th year.

b. May develop depigmentation.

c. May show mental retardation or spastic paralysis.

d. May develop blindness.

ANSWER

1 (c) Vesicular lesions are possible in all the five conditions. In pemphigus neonatorum (bullous impetigo) they soon become pustules and are accompanied by rather severe constitutional symptoms. In urticaria pigmentosa pigmented macules could certainly occur but they are often oval and discrete rather than in linear whorls and the papules would be pale and smooth rather than verrucous. In epidermolytic hyperkeratosis the background would have been erythematous. The presence of verrucous papules and a bizarre pattern of macules, together with vesicular lesions would strongly favour a clinical diagnosis of incontinentia pigmenti.

2 (a) & (b) Biopsy would be the obvious choice. Some of us feel VDRL should be done as a routine in all neonates with bullous lesions.

3 (a) (c) & (d) The bullous lesions would show accumulation of eosinophils; in the verrucous stage many large dyskeratotic cells scattered throughout a psoriasiform epidermis is diagnostic. Pigmented macules would show dermal melanophages and a practically nonpigmented epidermis.

4 (a) (b) (c) & (d) All these changes should be looked for, particularly ocular and neurological changes.

5 (c) The defect is thought to be on X-linked dominant trait which is lethal in males; 97% of reported cases are females.

6 (a) (b) (c) & (d) Pigmentation generally subsides as the child grows, but associated developmental anomalies are often serious.

Discussion

Incontinentia pigmenti (Block-Sulzberger syndrome) is an uncommon genodermatosis that usually affects female infants. The disease is most often characterized by an erythematous eruption with linear vesiculation that is often present at birth, followed by verrucous growths on the extremities. The final stage is characterized by irregular macules, streaks and splashes of brown to slate-grey pigmentation usually distributed asymmetrically on the torso and, less often, on the extremities. The pigmentation and other skin manifestations gradually resolve so that they are usually absent by adulthood. However, it is a potentially serious disease because of the developmental abnormalities that affect the hair, eyes, teeth, central nervous system and skeletal system, with one or more systemic manifestation in 79.8% of patients. One fourth of the patients have a major CNS anomaly (e.g. mental retardation, microcephalus, motor retardation, paralysis, convulsive disorders) and ocular changes occur in almost 20% of patients, sometimes leading to frank blindness. Genetic counselling is mandatory.

References

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