Continuing Medical Education

Clinical findings in genodermatoses based on Schahner LA & Hansen RC: Pediatric dermatology, New York: Churchill Livingstone, 1995.

Chose the one best answer to the following questions:

1. Patients with the Richner-Hanhart Syndrome have the following skin signs:

A. Palms and soles: Hyperhidrosis, bullae, painful hyperkeratotic erosions.
B. Eyelids and ears: Teleangiectasia and dermatitis.
C. Face: Milia, hypertrichosis and erosions.
D. Body: Hypertrichosis and hyperhidrosis.

2. Café-au-lait macules are seen in the following diseases:

A. Pachyonychia congenita
B. Albrights's syndrome
C. Refsum's disease
D. Conradi-Hunerman's disease

3. Rombo's disease is characterised by the following skin abnormalities:

A. Thin scalp hair, thin eyelashes, thin nails, dry skin.
B. Pachyonychia congenita, palmoplantar keratosis, hyperhidrosis, follicular keratosis.
C. Vermiculate athropoderma, milia, hypothrichosis, trichoepitheliomas, basal cell carciniomas, peripheral vasodilation, cyanosis.
D. Milia, hypertrichosis and erosions.

4. Pili torti, seizures, hypothermia, drowsiness, hypotonia, blindness progressive neurological deterioration. Bone and urinary tract abnormalities, and emphysema, occur in the following disease:

A. Lindgren's disease
B. Mauritzen's disease
C. Minkies disease
D. Menkes disease

In the following you must chose the best combination of answers.

A. Answers 1 and 4 are correct.
B. Answers 1, 2 and 4 are correct.
C. Answers 2 and 4 are correct.
D. Answers 3 is correct.

5. Christ-Siemens-Touraine syndrome (anhidrotic ectodermal dysplasia) may include the following signs:

1. Thin scalp hair
2. Thin nails
3. Subungual hyperkeratosis
4. Dry mucosa e.g. atrophic rhinitis,

6. Jadassohn-Lewandowsky syndrome (pachyonychia congenita) may include the following signs:

1. Focal dermal hypoplasia
2. Palmoplantar keratosis
3. Eczematous dermatitis and peculiar smell.
4. Hyperhidrosis

Suggested answers:

1. A: Richner-Hanhart Syndrome (tyrosinemia II):
Skin: Palms and soles: Hyperhidrosis, bullae, painful hyperkeratotic erosions.
Other organs: Mental retardation, ocular changes: corneal clouding, a.o.
2. B: Albright Syndrome (polyostotic fibrous dysplasia):
Skin: Café-au-lait macules,
Other organs: Polyostotic fibrous dysplasia of bone, precocious puberty, endocrinopathies.
3. C: Rombo syndrome:
Skin: vermiculate athropoderma, milia, hypothrichosis, trichoepitheliomas, basal cell carciniomas, peripheral vasodilation, cyanosis.
4. D: Menkes disease:
Skin: Coarse, lusterless, hypopigmented, twisted hair (pili torti).
Other organs: Seizures, hypothermia, drowsiness, hypotonia, blindness progressive neurological deterioration. Bona and urinary tract abnormalities, and emphysema.
5. B: Christ-Siemens-Touraine syndrome (anhidrotic ectodermal dysplasia):
Skin: Lack of sweating, thin scalp hair, thin eyelashes, thin nails, dry skin.
Other organs: Dental abnormalities, dry mucosa, e.g. atrophic rhinitis, corneal dystrophy, uveitis,
6. C: Jadassohn-Lewandowsky syndrome (pachyonychia congenita):
Skin: Pachyonychia congenita, palmoplantar keratosis, hyperhidrosis, follicular keratosis.