Congenital Diseases (Genodermatoses)

Paul K. Shitabata, M.D.
Director of Dermatopathology
Harbor-UCLA Dermatology

Which is (are) the incorrect association(s) for CHILD Syndrome:

- C-Congenital nevi
- H-Hemidysplasia
- I-Ichthyosiform erythroderma
- L-Lentigo
- D-Defects of the limb

Answer

- C-Congenital hemidysplasia
- L-Limb Defects

Which has increased epithelial proliferation?

- Ichthyosis vulgaris
- Epidermolytic hyperkeratosis
- Erythrokeratodermia variabilis

Answer

- Epidermolytic hyperkeratosis

Which association is incorrect?

- Harlequin ichthyosis-increased delivery of desmosomal proteases to stratum corneum
- Refsum's syndrome-cerebellar ataxia
- Pachyonychia congenita-Autosomal dominant
- Dyskeratosis congenita-X-linked recessive

Answer

- Harlequin ichthyosis-decreased delivery of desmosomal proteases to stratum corneum
  - This leads to failure to degrade corneodsemosomes and massive hyperkeratosis

Pertinent histopathological differential diagnoses for Rothmund-Thomson syndrome include all of the following except:

- Dermatomyositis
- Mycosis fungoides
- Discoid lupus erythematosus
- Lupus vulgaris
- Phototoxic dermatitis

Answer

- Lupus vulgaris
  - The predominant finding is that of a cell-poor interface dermatatis with melanophages-so called poikiloderma changes
  - Occasional variants of mycosis fungoides may present with these changes and can be considered if cytologically atypical T-cells are present

Common histopathologic patterns associated with porokeratosis include the following except:

- Lichenoid dermatitis
- Acantholysis
- Parakeratosis
- Dyskeratotic keratinocytes
- Keratinocytic atypia

Answer

- Acantholysis

Remember that porokeratosis may have a diffuse lichenoid infiltrate in the center of the lesion, away from the cornoid lamella.

Incorrect association?

- Darier's disease-Acantholysis
- Hailey-Hailey disease-Corp ronds
- Acrokeratosis of Hopf-Acantholysis
- Bullous congenital ichthyosiform erythroderma-epidermolytic hyperkeratosis

Answer

- Hailey-Hailey disease-Corp ronds

  Only a crumbling dilapidated brick pattern is present in this disease, lacking corp ronds or corp grains.

Histopathologic tissue reaction patterns of Incontinentia pigmenti include all except:

- Eosinophilic spongiosis
- Neutrophilic spongiosis
- Melanophages
- Hemosiderin laden macrophages
- Papillomatosis

May have more than one answer

Answer

- Neutrophilic spongiosis
- Hemosiderin laden macrophages

The classic findings begin with an eosinophilic spongiosis, followed by epidermal hyperplasia, and finally melanophages.

Elastic fibers associations?

Disease	Elastic Fibers (+,-, altered)
Cutis Laxa	
Ehlers-Danlos syndrome	
Connective tissue nevus	
Acrokeratoelastoidosis	

Answers

Disease	Elastic Fibers (+,-, altered)
Cutis Laxa	Decreased
Ehlers-Danlos syndrome	No change to slight increased type I>>II, III
Connective tissue nevus	Increase
Acrokeratoelastoidosis	Altered-Diminished and fragmented

Review of Epidermolysis Bullosa

Type	Name	Inheritance
Epidermal	Simplex Cockayne Dowling-Meara (herpetiform)	AD AD AD
Junctional	Letalis Benign	AR AR
Dermal	Dystr-dom Dystr-rec (generalized) Dystr-rec (localized) Dystr-inversa Acquisita	AD AR AR AR -