F022 - Virtual Dermatopathology Self Assessment:

Case 04

Answers, Information and References

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I have no financial conflicts of interest and nothing to disclose
Case History

48 year old woman with yellowish papules in the flexural areas of her extremities and visual problems.
The correct diagnosis is:

A. Elastosis perforans serpiginosa
B. Mid dermal elastolysis
C. Cutis laxa
D. Pseudoxanthoma elasticum

A. None of the above
Clinical Features

Characterized by elastic tissue damage in 3 compartments:

Skin: Yellowish linear and reticular papules and «cobblestone» plaques particularly on lateral neck, axillary, cubital and popliteal folds, groins and periumbilical area; redundant folds and oral mucosa

Eye: Angioid streaks in Bruch’s membrane leading to vision loss, hemorrhages and chorioiditis

Vessels: Arterial obstruction, aneurysms and hemorrhages notably in the brain, heart and periphery
Discussion:

- **Differential Diagnosis**: acquired pseudoxanthoma elasticum in
  - Skin: solar elastosis + D-penicillamin (do not calcify), chronic kidney disease, niter (saltpeter) exposure, eosinophilia-myalgia syndrome
  - Skin, vessels and eyes: beta-thalassemia & sickle cell anemia, primary systemic amyloidosis, hemochromatosis

- **Genetics**: recessive ABCC6 mutations ATP Binding Cassette transmembrane transporter family C, member 6
  - 1503 aa and 165 kDa, expressed in kidney, liver >> skin & vessels > retina & placenta

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Histopathology

- Pseudoxanthoma elasticum is marked by
  - Short, curled and distorted, **fragmented basophilic elastic fibers** in the mid and upper reticular dermis. Occasionally, there may be perforating transepidermal elimination.
  - **Calcium deposits on elastic fibers** appear as purple clumps in HE stains in advances disease.
  - However, elastin (Verhoeff) and calcium (von Kossa) stains are helpful to recognize the characteristic « sprouting » elastic fibers.
  - Histologic severity correlates well with clinical changes.
Histopathology

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References

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