A case of Pseudoxanthoma elasticum:

A rare disorder in Korea with few reported cases

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Case

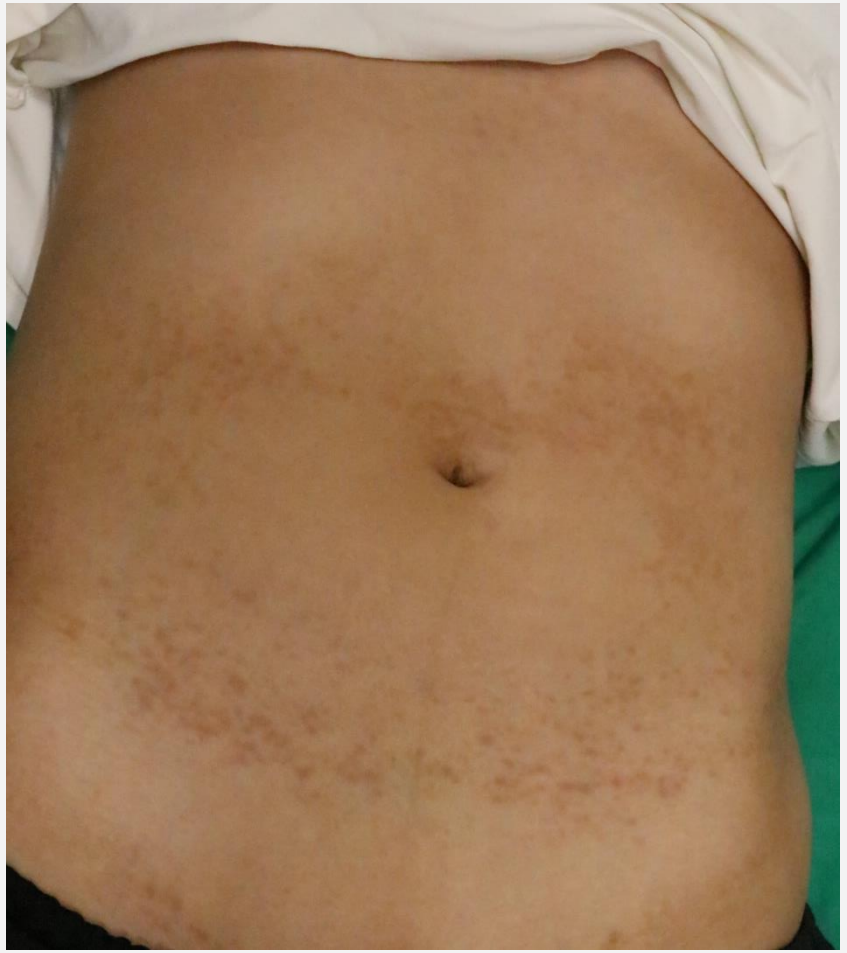
- **Patient**
  - M/14

- **Chief complaint**
  - Skin lesion on trunk, neck
  - Onset : 7-8YA

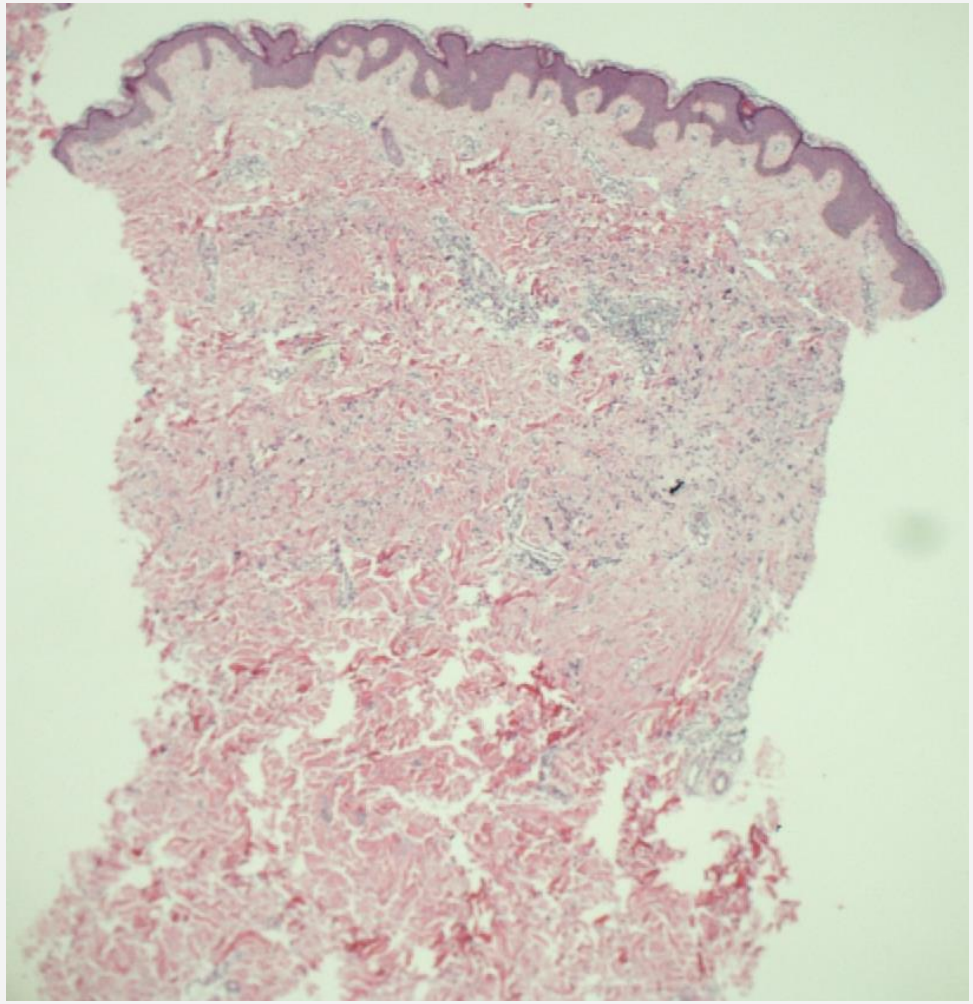
- **Present illness**
  - Erythematous to brownish maculopatches on abdomen, inguinal area
  - Multiple skin colored to yellowish papules on neck

- **Past History** : h/o Tonsillitis

Skin manifestation



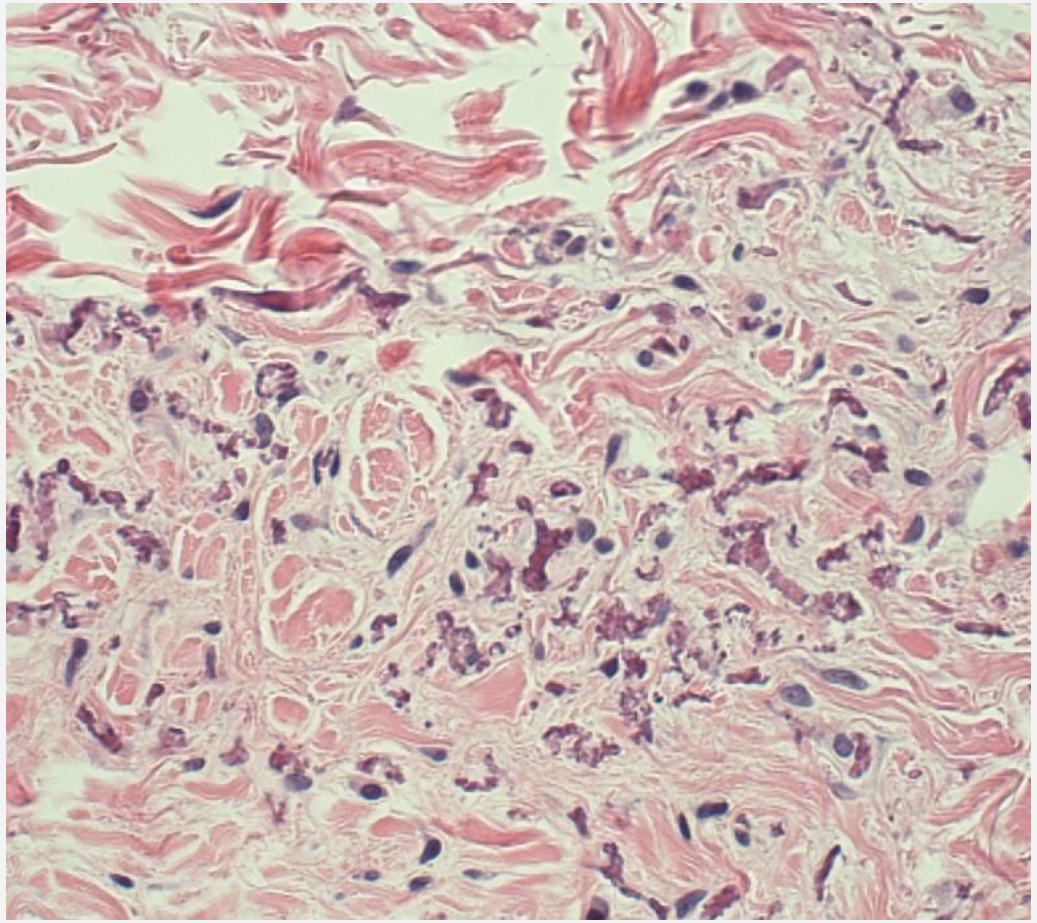
Histopathologic findings



(H&E stain, X40)

- No typical findings on horny layer, epidermis
- Basophilic dermis

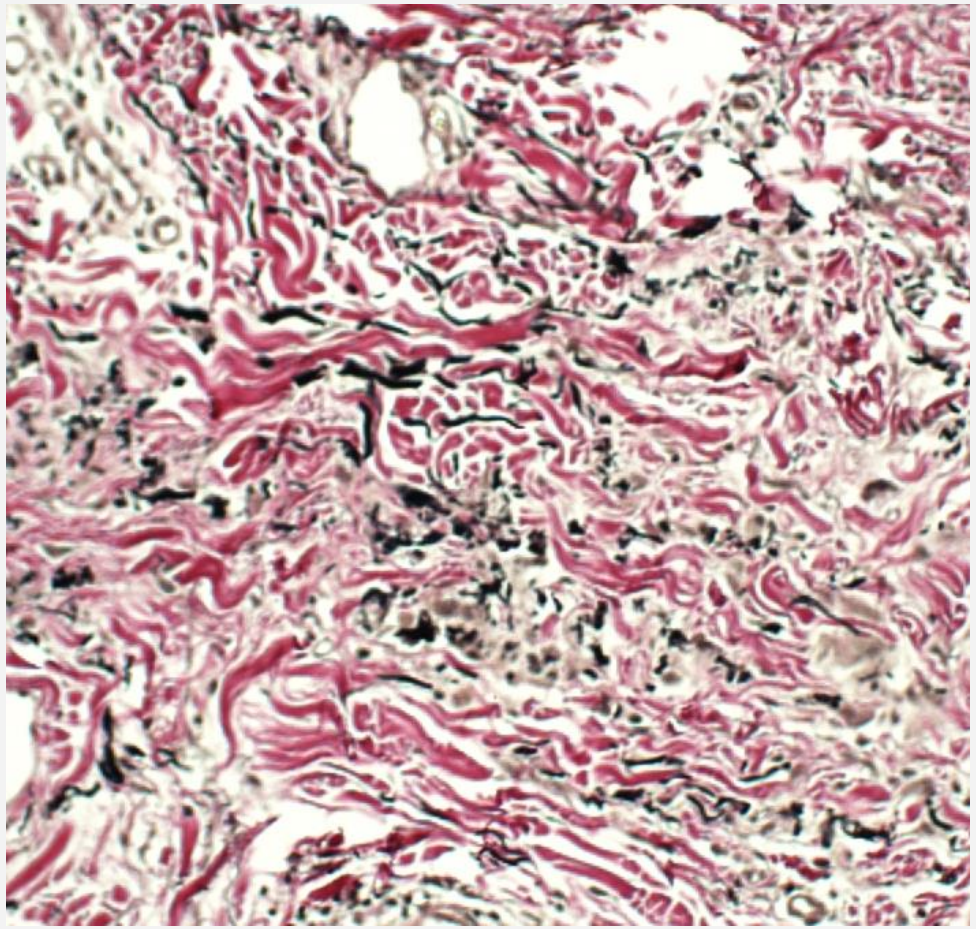
Histopathologic findings



(H&E stain, X400)

- Fragmented, clumped, basophilic elastic fibers
- Granular deposits

Histopathologic findings



(EVG stain, X200)

- Fragmented, clumped, basophilic elastic fibers
- Granular deposits

Diagnosis & Management

- **Diagnosis**

- Intradermal granular deposits

,composed of fragmented clumped basophilic elastic fibers

, partly phagocytized by histiocytes,

consistent with pseudoxanthoma elasticum

- **Management**

- referred to clinical genetics department for genetic counseling

Review

Pseudoxanthoma elasticum (PXE)

Pseudoxanthoma elasticum

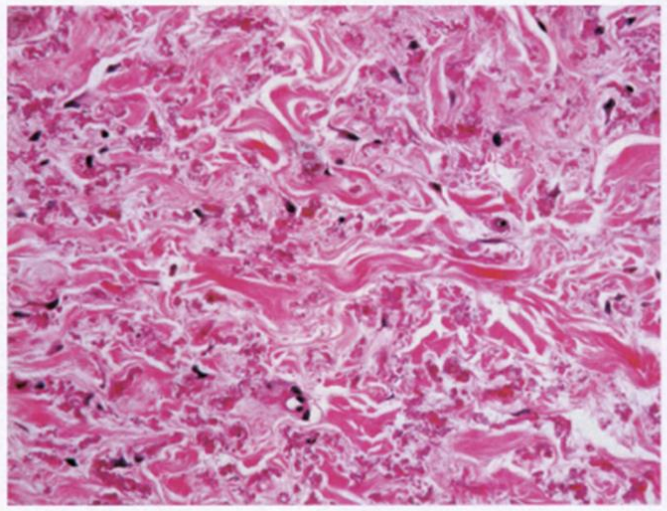
- Abnormal calcification and mineralization of elastic fibers in affected tissues
- Yellow papules and plaques, skin laxity on neck, axillar, arms (bilateral)
- Inguinal area, abdomen, mucosal area (lip, GI)
- Ocular manifestations: incur vision loss at later stages

- Autosomal recessive disease
- Classic: ABCC6 gene mutation (encodes efflux transporter protein)
- Progressive deposition of calcium and phosphorus on elastic fibers → become fragmented

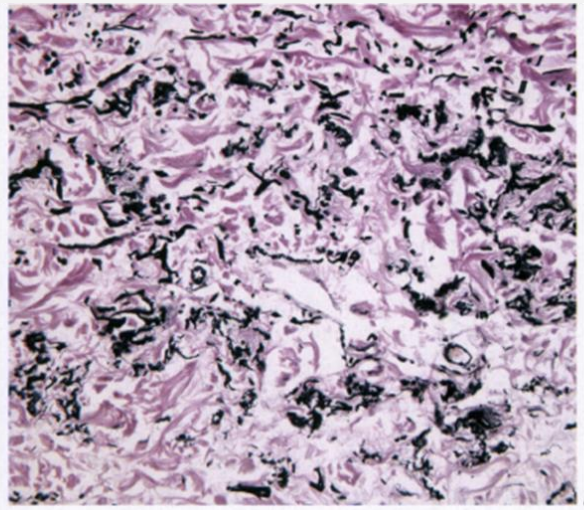
Clinical manifestation

Cutaneous features	Ocular features	Cardiovascular features	Other features
1) 1-5mm yellowish papules, distributed over the lateral neck, flexural areas 2) Possible involvement of oral, rectal, or vaginal mucosa 3) Reticular or linear arrangement of lesions 4) Lesions progressively coalesce into larger plaques with a "cobblestone" appearance 5) Affected skin becomes soft, lax, slightly wrinkled over time 6) Prominent mental fold 7) Lesions are asymptomatic	Retina, angioid streaks, etc -> may lead to vision loss	Hypertension Peripheral artery diseases Aneurysm Ischemic or hemorrhagic stroke Heart attack Sever limb claudication Stenosis, etc	GI, UTI bleeding Calcification Miscarriage in early pregnancy

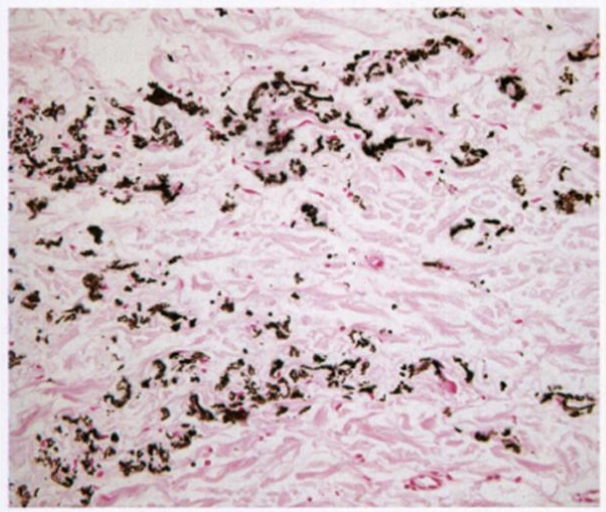
Histophathology



H&E



Elastic tissue stain



Von Kossa

- Elastic fibers of reticular dermis are short and fragmented (faintly basophilic)
- Calcification of the elastic structures
- Von Kossa: staining for Calcium

Differential diagnosis

Differential diagnosis	Differences
Solar elastosis	Abnormal elastic tissue located in the upper third of the dermis Calcium deposition (-) -> need Von Kossa stain
Calcific elastosis (= Perforating pseudoxanthoma elasticum, Localized acquired pseudoxanthoma elasticum)	Perforation (+) If perforation is absence -> clinical data are necessary (usually on periumbilical, related with pregnancy, obesity, operation related trauma)
PXE-like phenotypes	Sickle cell disease, ß-thalassemia (related to HBB mutation) GGCX mutation (related with coagulation deficiency)
Papillary dermal elastolysis	Absence/reduction of elastic fibers in the papillary dermis

Treatment

- No cure for PXE, treatment is supportive
- Avoid contact sports, intense exercise d/t risk of traumatizing already-calcified vessels
- Close f/u with ophthalmology and cardiology is imperative

Conclusion

- This case presented **erythematous to brownish maculopatches on trunk / skin colored to yellowish papules on neck** for several years

- **Pseudoxanthoma elasticum** is an uncommon entity.

- **Close follow up** is imperative for screening complications.

References

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