G24 The Impact of Dermatologic Consultation in Autopsy Examination: A Case of Pseudoxanthoma Elasticum

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After attending this presentation, attendees will understand and realize the usefulness of dermatologic consultation in autopsy examinations through a case presentation of pseudoxanthoma elasticum.

This presentation will impact the forensic community and/or humanity by demonstrating that a dermatology consultation may be a very useful addition to forensic and hospital autopsies. We present an example of how this consultation may lead to the diagnosis of systemic disease and possible cause of death.

Dermatologic consultation in the autopsy examination may be very useful in many cases. We present a case of pseudoxanthoma elasticum, a genetic disease, that was diagnosed by a thorough external examination and histologic evaluation of the skin.

A forty-nine-year old white female was found unresponsive in an apartment fire and was taken to the local area hospital. On arrival, the patient was in asystole and had a carbon monoxide concentration of sixty percent. She was also found to have evidence of smoke inhalation and burns over approximately fourteen percent of the total body surface area. The patient was pronounced dead and was transferred to the Dallas County Medical Examiner's Office.

On skin examination, the patient had a large leathery, firm, dark brown plaque with surrounding erythema on her back and right flank. On the leading edge of the plaque, there were multiple bullae and skin sloughing. The leathery plaque was clinically consistent with the most severe burn site and was likely the area closest to the heat source given the intense drying of the skin. The bullae and skin sloughing was clinically consistent with a third degree, full thickness, burn. On the skin of her neck, anticubital fossa, and inguinal area, the patient had small yellow papules and a wrinkly appearance that resembled "plucked chicken skin." A biopsy was taken of the bullae on the right flank and the skin the inguinal area.

The biopsy of the bullae was consistent with a full thickness burn and showed a necrotic epidermis and polarization of the nuclei around the hair follicles. The biopsy of the skin taken from the inguinal area was characteristic of pseudoxanthoma elasticum and showed degeneration of the elastic fibers in the middle and lower dermis. On hematoxylin and eosin staining, the elastic fibers were basophilic, irregular, and widely dispersed among the collagen bundles. On von Kossa staining, these elastic fibers were highlighted in a dark brown color.

Pseudoxanthoma elasticum is an autosomal recessive disorder due to a mutation in the ABCC6 gene on chromosome 16. The patients have clumped, distorted, calcified elastic fibers which manifest as disease in many organ systems. The patients typically have flat, yellowish papules on the skin of flexural areas that sometimes coalesce to resemble "plucked chicken skin." Most patients also develop angioid streaks in the eye that may lead to blindness, and many develop progressive calcification of the medium sized arteries which leads to hypertension and myocardial infarctions at a much younger age. Patients may also have calcification of the cerebral and gastric vessels.

Upon further investigation into this patient's history, she was found to be blind and have severe hypertension. These findings, along with the characteristic histologic findings on skin biopsy, lead to a definitive diagnosis of pseudoxanthoma elasticum. This disease has tremendous implications for the family given that it is inheritable and may cause significant morbidity and mortality.

Dermatologic consultation was extremely useful in this case, given that the skin findings, combined with the knowledge of the patient's medical history, provided the diagnosis of a rare, genetic, and life threatening disease.

Dermatology, Consultation, Autopsy

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