H66 Pulmonary Alveolar Microlithiasis: A Case of Sudden Death

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Learning Overview: After attending this presentation, attendees will better understand Pulmonary Alveolar Microlithiasis (PAM), a rare hereditary lung disease, and its histology.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by increasing attendees' understanding of PAM and the role of histology in cases of sudden death with non-specific autopsy findings.

Introduction: PAM is a rare autosomal recessive disorder characterized by intra-alveolar accumulations of calcium phosphate that eventually impedes gas exchange. It is caused by mutations to the type IIb sodium-phosphate gene cotransporter (SLC34A2) responsible for phosphate homeostasis found on type II pneumocytes.[1] The inability to transport the phosphorus ions out of the alveoli leads to the microlith formation. Most people with PAM have a sibling with the disease. Symptoms usually appear in the third or fourth decade. Patients with PAM are usually asymptomatic in early stages. As the disease progresses, symptoms include dyspnea, dry cough, chest pain, and cor pulmonale. Diagnosis is usually made by imaging or lung biopsy. Chest radiograph would have the "sandstorm" appearance of the diffuse opacifications of both lungs.[2] Histologically, these intra-alveolar accumulations are round to oval concentric laminated microliths. The only effective treatment is a lung transplant.[3]

Materials and Method: The decedent was a 32-year-old woman with a history of diabetes and alcohol abuse. She was not taking any medications. She had an episode of vomiting the morning of her death but was feeling better around dinnertime the same day. She was found unresponsive shortly thereafter and was taken to the hospital and was not revived.

Results: At autopsy, the lungs were firm and non-compliant. The right lung weighed 1,280 grams and the left lung 1,200 grams. The pleural surface was smooth and shiny. The cut surface was gritty, firm, and consolidated. There was honeycombing of the parenchyma. Microscopic examination of decalcified lung showed lamellated calcium microliths filling most of the alveoli, with no fibrosis or inflammation. All other organs were within normal parameters. Toxicology was performed and was negative.

Discussion: This case highlights the importance of histology in correlating non-specific gross findings with microscopic findings. This is an unusual case because the decedent did not know she had PAM, and her death was earlier than most cases reported in the literature. The lack of fibrosis and cor pulmonale findings could indicate a rapid progression, even though it is a disease known for its slow and progressive course. Establishing the diagnosis after death has significance because it allows siblings to be tested and monitored for the disease.

Reference(s):

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2. Siddiqui, N. and Fuhrman, C. (2011). Best Cases from the AFIP: Pulmonary Alveolar Microlithiasis. *RadioGraphics*, 31(2), pp.585-590.
3. Saito, A. and McCormack, F. (2016). Pulmonary Alveolar Microlithiasis. *Clinical Chest Medicine*, 37(3), pp.441-448.

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