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Pregnancy in pulmonary alveolar microlithiasis

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Clinical image description

A 32-year-old nepalese woman presented with dyspnea and dry cough. She had a non-confirmed diagnosis of pulmonary tuberculosis (TB) and completed 6-months of antibacilar treatment 10 years ago. She had no occupational exposure, smoking habits or familiar history of respiratory diseases. Auscultation revealed bilateral decreased and rude respiratory sounds. Chest radiograph showed a bilateral dense micronodular pattern. Blood tests were unremarkable. Because of tuberculosis suspicion, she started antibacilar treatment. Bronchoscopy ruled out infection and the pulmonary alveolar microlithiasis (PAM) diagnosis was considered due to characteristic radiologic pattern, stopping the antibacilar treatment. Genetic test (SLC34A2 mutation) confirmed the diagnosis. No mutation was detected in one brother. The patient remained stable, without respiratory failure.

Genetic counselling was provided, consanguinity was excluded and the husband was tested and no pathogenic genetic variants were found. Being PAM an autosomal recessive disease there is no risk of further inheritance in this couple. Despite knowing the risks and the uncertainty of the safety of labour, the patient became pregnant and decided to go through with the pregnancy. No complications were detected during the gestation and an eutocic delivery was possible without maternal or neonatal complications.

This is a rare genetic disease [1] that can be underestimated, especially in endemic areas of tuberculosis, provoking calcium microliths intra-alveolar accumulation and without directed therapy. Presently, the only effective therapy is lung transplantation. Classical CT scan pattern can confirm the diagnosis, avoiding invasive procedures. Genetic counselling must be offered and there is a chance of a pregnancy without complications in these patients.

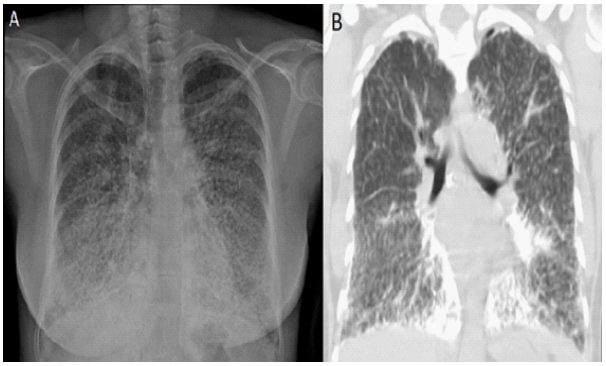

Figure 1: Chest X-ray with bilateral dense micronodular pattern (A); Chest CT-scan with high density (B).

Declarations

Contributorship: CF and DM choose the images from the exams, written and revised the manuscript.

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Informed consent/Patient Permission: obtained and attached.

References

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