Pulmonary alveolar microlithiasis: often misdiagnosed in children

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Accepted 7 July 2019

DESCRIPTION

An 8-year-old girl presented with the complaints of gradually progressive dry cough for 2 months, which was aggravated at night. There was no history of fever, fast breathing or breathing difficulty, or noisy breathing. Her parents visited a local practitioner where she was investigated and, based on only chest radiograph findings, diagnosis of miliary tuberculosis (TB) was made and, she started on category-I anti-tubercular therapy (ATT). Despite a course of ATT, she continued to have symptoms, and so she was referred to our institute for further evaluation. Her clinical details and investigation were reviewed. She was born to nonconsanguineous parents, second in birth order, delivered at term by normal vaginal delivery. Her neonatal period was uneventful. She was partly immunised, and developmental was appropriate for age. On examination, her vitals were within normal limit; there was no pallor, cyanosis, oedema or lymphadenopathy. Her weight and height for age were between 0 and −1 SD. On chest examination, air entry was equal on both sides, and there was no added sound. Other systemic examination findings were also normal. Her chest X-ray showed dense reticulonodular opacities in bilateral lungs field, looking like 'sandstorm' appearance (figure 1A). The differential diagnoses that were considered based on the clinico-radiological investigations were miliary TB, immunodeficiency disorder, acyanotic congenital heart disease, fungal infection (histoplasmosis) and pulmonary haemorrhage. There was no history of contact with a TB case and no weight loss in the last 3 months. Moreover, in TB workup, Mantoux test was negative, the GenXpert test did not detect Mycobacterial tuberculosis in induced sputum and bronchoalveolar lavage (BAL). Flexible bronchoscopy did not reveal any structural abnormalities, and there was no PAS-positive material or hemosiderin laden macrophage detected in BAL. Fungal culture in BAL was negative. Detailed immunodeficiency workup was done which was unremarkable (HIV: nonreactive, IgG: 563 mg/dL, IgM: 211 mg/dL, IgA: 154 mg/dL, IgE: 186 mg/dL). High-resolution CT (HRCT) scan of the chest was done, which revealed high attenuation linear areas (density equivalent to calcium) in bilateral lungs predominantly in posterior location suggestive of pulmonary alveolar microlithiasis (PAM) (figure 1B,C). Based on the clinical profile and HRCT chest findings, a diagnosis of PAM was considered.

PAM is a rare, autosomal recessive disease characterised by lamellar deposition of calcium phosphate or 'microliths' within the alveoli. PAM is uncommon in childhood, including neonate and infancy.[1 2] It is caused by the mutation in the SLC34A2 gene which code for a phosphate transporter in type II alveolar cell responsible for the removal of degraded phosphate, which results in microliths accumulation in the alveoli.[2] Children usually remain asymptomatic at the time of diagnosis; however, they can present with non-productive cough, exertional dyspnoea, pneumothorax, digital clubbing and so on. PAM is usually diagnosed by characteristic chest radiograph with 'sandstorm' appearance, which is considered pathognomonic of PAM. HRCT shows diffuse micronodular calcified densities with thickening of the septa.[3] Other diagnostic modalities are BAL, nuclear scan, lung biopsy and detection of SLC34A2 mutation, which can help to establish the diagnosis.[2 4] Military TB, sarcoidosis, histoplasmosis,

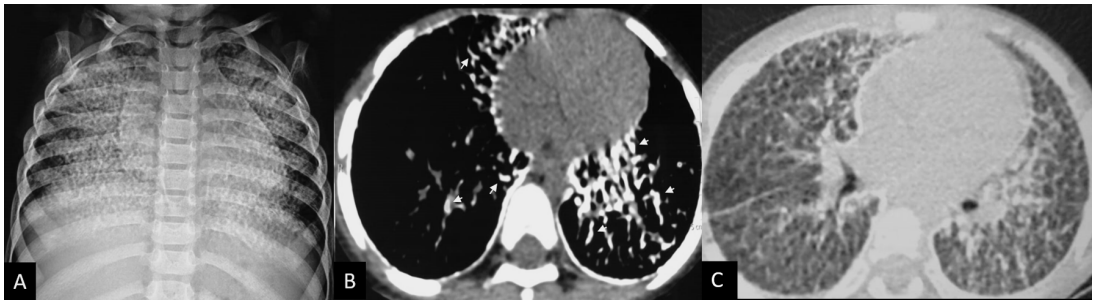

Figure 1 Chest radiograph showing dense reticulonodular opacities in bilateral lungs (A). HRCT soft tissue (B) and lung (C) window confirming high-density linear areas of calcification (arrows) in bilateral lungs predominantly in posterior location suggestive of alveolar microlithiasis. HRCT, high-resolution CT.

To cite: Kumar P, Binit S, Goyal JP. *BMJ Case Rep* 2019;**12**:e230776. doi:10.1136/bcr-2019-230776

Learning points

- Bilateral diffuse opacity in lung fields has several differentials; the common ones are military tuberculosis, sarcoidosis, histoplasmosis, idiopathic pulmonary haemorrhage and pulmonary alveolar microlithiasis (PAM).
- Empirical ATT should be avoided in the absence of microbiological or other supportive evidence for TB.
- The diagnosis of PAM can be established with typical radiological appearance.

and idiopathic pulmonary haemorrhage are the close differential for PAM. Although, various therapies have been described, namely, whole lung lavage, glucocorticoid and bisphosphonate, currently, there is no curative treatment for PAM. Lung transplantation promises some hope but the real benefit is still under investigation.[2 5]

Contributors PK and JPG were involved in conceptualisation and drafting of manuscript. SB was involved in manuscript design. All the authors reviewed the manuscript.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Parental/guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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