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Case Report

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A unique association of diaphragmatic hernia and pulmonary tuberculosis in an infant

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Abbreviations: CXR: Chest X Ray; CT: Computed Tomography; CDH: Congenital Diaphragmatic Hernia; TB: Tuberculosis.

Case report

We report a healthy-appearing 7-month-old girl of non-related ancestry, uneventful pre - and postnatal development and past medical history, who was investigated for lung Tuberculosis (TB) in her mother. Tuberculin skin test was highly positive (20 mm induration, normal range 0-5 mm in unvaccinated children). Chest X-ray (CXR) revealed a surprising image suggestive of the presence of abdominal contents in the thoracic cavity (Figure 1); the unobscured lung parenchyma showed no signs of TB. Computed Tomography (CT) imaging detected branching linear opacities and patchy consolidation in the right upper lobe, intrathoracic lymphadenopathy and confirmed retrosternal dislocation of colon (Figure 2 A,B,C). Thus, the diagnosis of concurrent lung TB and Congenital Diaphragmatic Hernia (CDH) was established. The 4-drug regimen of antituberculotics was initiated, i.e. isoniazid, rifampicin, pyrazinamide and ethambutol. The hernia repair was deferred to prevent abdominal dissemination of *Mycobacterium tuberculosis*.

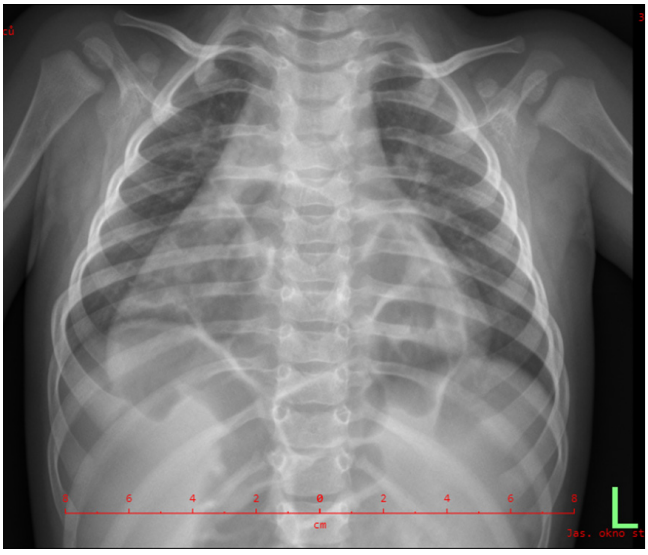

Figure 1: Chest X ray of the 7-month-old girl with diaphragmatic hernia and tuberculosis depicting the dilated colon in the thoracic cavity which precludes the visibility of lung parenchyma and the heart shadow.

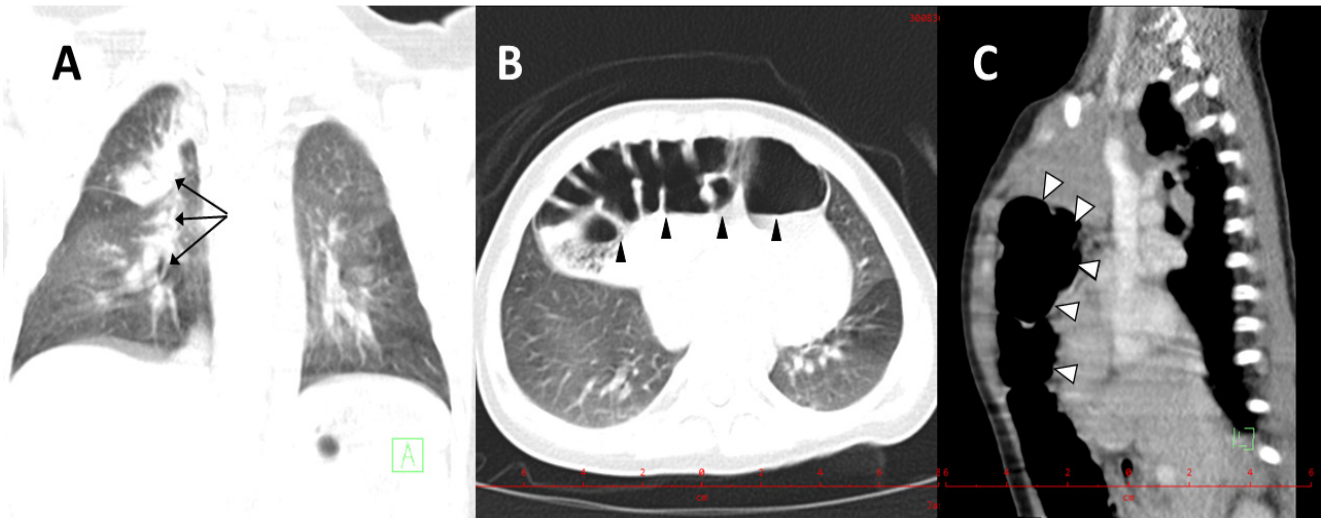

Figure 2: Chest CT of the 7-month-old girl with diaphragmatic hernia and tuberculosis.
(A) Anteroposterior CT (2D reconstruction). Patchy consolidation, branching linear opacities, and inthrathoracal lymphadenopathy in the upper right lobe are indicated by arrows; **(B)** Axial CT of the chest. Trasversal colon in the anterior thoracic cavity (headarrows); **(C)** Sagittal chest CT (2D reconstruction) depicting the retrosternal presence of the colon in the thoracic cavity (headarrows).

CDH represents the most common intrathoracic fetal anomaly (1:3,000 live births; male to female ratio 2:1). The abdominal contents protrude into the thoracic cavity through a developmental defect in the diaphragm. Ranging from a small aperture in the posterior muscle rim to a complete absence of diaphragm, it may interfere with normal lung development. Postero-lateral hernias, also known as Bochdalek hernias, are the most common (70-75%), followed by anterior defects or Morgagni hernias and central hernias [1]. Clinical presentation is variable; most typically, tachydyspnea, tachycardia, cyanosis and caved-in appearance of the abdomen are apparent shortly after birth. CDH is often diagnosed via antenatal ultrasound. In some cases, it remains oligosymptomatic till adulthood. The incidence of TB in the Czech Republic is very low (4,3/100 000) [2]. The population at highest risk are the youngest children living in TB households [3].

The occurrence of lung TB in a patient with CDH has not been reported so far and we presume the association to be coincidental. Of note, several cases of late-presenting CDH, treated erroneously as TB due to a misinterpreted CXR, have been referred [4]. Our case demonstrates the necessity to actively investigate TB household contacts, even if asymptomatic. It also emphasizes the utility of CT imaging in case of negative or inconclusive chest X ray in selective screening for tuberculosis in infants at high risk of TB acquisition.

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