

Not every chronic cough is tuberculosis: Congenital diaphragmatic hernia mimicking active tb in an indigenous patient — a diagnostic dilemma in primary care

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SUMMARY

Tuberculosis (TB) continues to disproportionately affect the indigenous Orang Asli population in Malaysia, often presenting diagnostic challenges in primary care settings. We report a case of a 59-year-old Orang Asli man identified during targeted community TB screening following multiple TB-related deaths in Kampung Bumiputera Dalam, Rengit, Malaysia. Although asymptomatic at the initial screening, he demonstrated strong Mantoux positivity and abnormal chest radiography findings. Delayed follow-up revealed persistent cough, raising concerns about active pulmonary tuberculosis. However, repeated sputum microbiological investigations, including GeneXpert MTB/RIF, were negative, and the radiological findings remained stable. Radiological reassessment revealed an incidental congenital diaphragmatic hernia, providing an alternative explanation for the symptoms. Following a multidisciplinary discussion, a diagnosis of latent tuberculosis infection (LTBI) was made. The patient completed a three-month rifampicin-isoniazid regimen despite temporary treatment interruption. This case highlights the importance of correlating microbiological evidence with radiological findings in TB-endemic settings and maintaining a broad differential diagnosis to avoid unnecessary treatment. This underscores the central role of family medicine in balancing disease containment with accurate patient-centered care in vulnerable Indigenous populations.

INTRODUCTION

Tuberculosis remains a significant public health concern in Malaysia, especially among Orang Asli communities, where socioeconomic vulnerability, geographic isolation, and limited healthcare access challenge early diagnosis of the disease.¹ Community-based TB screening programs are vital, fostering a sense of shared responsibility and collective effort to improve health outcomes in high-risk populations.²

In TB-endemic settings, clinical decision-making should consider a broad differential diagnosis to avoid overreliance on pre-test probability and public health considerations, which can lead to unnecessary treatment and anxiety. Delivering comprehensive Tuberculosis Awareness among indigenous communities is important to facilitate early preventive and targeted treatment of the disease. However, community resistance and communication barriers limit

these opportunities.² Recognizing non-tuberculous conditions that mimic pulmonary TB helps clinicians feel assured of their thorough evaluation. As latent tuberculosis cases increase, the need to start preventive treatment earlier may reduce the risk of active tuberculosis progression by approximately 5 to 10 %.³

Congenital diaphragmatic hernia (CDH) is typically identified in infancy, and its presentation in adults is uncommon.⁴ When present, displacement of the abdominal contents into the thoracic cavity may contribute to gastrointestinal reflux and chronic cough, creating a clinical picture that overlaps with pulmonary disease. Recognition of such atypical presentations is particularly important in primary care settings, where initial diagnostic pathways are established.

We present a case of an Orang Asli patient with a persistent cough and chest radiographic abnormalities initially suggestive of pulmonary TB but ultimately diagnosed with congenital diaphragmatic hernia. This dilemma stirs primary care anxiety with the urgency to treat the active disease while simultaneously establishing the correct diagnosis. This case highlights the diagnostic challenges faced by family physicians in tuberculosis-endemic communities. This underscores the importance of maintaining a broad differential diagnosis when molecular testing does not support an active infection.

CASE PRESENTATION

A 59-year-old Orang Asli man from Kampung Bumiputera Dalam, Rengit, with no known medical illness, underwent targeted TB mass screening on May 27, 2025, following several TB-related deaths within the community. At screening, the patient was asymptomatic, without cough, fever, weight loss, or constitutional symptoms. The Mantoux test demonstrated a 20 mm induration. A chest radiograph performed on the same date showed blunting of the right costophrenic angle, which was suggestive of pleural effusion or pleural thickening.

The patient presented late for follow-up at Klinik Kesihatan Rengit on July 25, 2025, complaining of a persistent cough for approximately three weeks. Investigations revealed sputum AFB smear ×3 negative, sputum culture with no

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Fig. 1: Chest X-ray PA view taken on 11.8.2025.

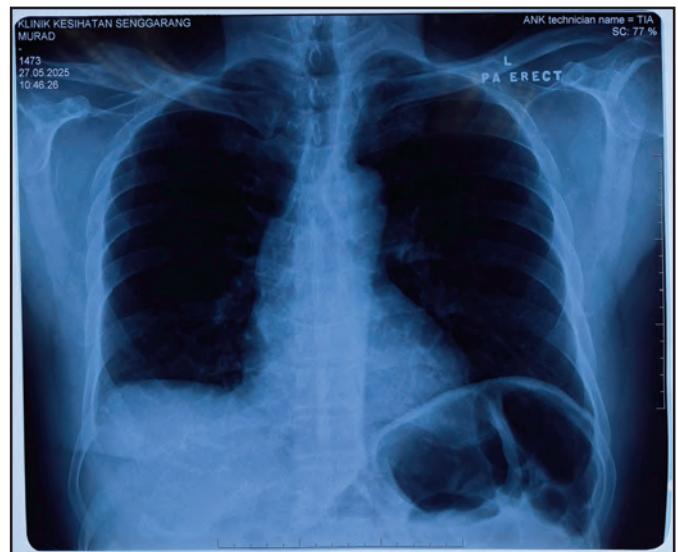


Fig. 2: Chest X-ray PA view taken on 27.5.2025.

growth, and an ESR of 5 mm/hr. The patient was subjected to a chest X-ray at a nearby health clinic with an available radiology unit, and serial chest radiographs (in May and August 2025) demonstrated persistent right costophrenic angle blunting with an incidental left congenital diaphragmatic hernia.

Given the ongoing symptoms and radiographic abnormalities, GeneXpert MTB/RIF testing was performed, and a multidisciplinary discussion with a respiratory physician was conducted. This collaborative approach aims to build confidence in diagnosis and ensure comprehensive patient care.

Considering the strong Mantoux reactivity, epidemiological exposure, stable radiographic findings, and absence of microbiological evidence of active disease, a diagnosis of latent tuberculosis infection was made. The patient commenced treatment with Akurit-2 tablets on August 15, 2025, for a planned three-month regimen. Treatment was interrupted due to default for approximately one month but was restarted on October 22, 2025, and completed on January 22, 2026.

The patient was concurrently managed at Klinik Kesihatan Rengit under the care of a Family Medicine Specialist for latent tuberculosis infection (LTBI). He completed the prescribed LTBI treatment and remained compliant with scheduled follow-up visits at 3, 6, 9 months, 1 year after treatment initiation.

The incidental finding of a left congenital diaphragmatic hernia on serial chest radiography was reviewed and discussed with both the radiologist and respiratory physician. Following counselling regarding the diagnosis, potential

complications, and available management options, the patient declined further investigations, specialist referral, or surgical intervention. He was asymptomatic and had lived comfortably with the condition for many years without any perceived impact on his daily activities or overall health. In addition, transportation difficulties posed a significant barrier, as the referral hospital was located approximately 45 km from his residence in Rengit.

Given the patient's informed decision, absence of symptoms, and stable clinical condition, no surgical referral was made at that time. The patient was advised on symptoms that would warrant immediate medical attention and continued primary care follow-up.

DISCUSSION

This case illustrates the diagnostic challenges encountered when assessing tuberculosis in high-risk Indigenous populations identified through community screening. Strong Mantoux reactivity and abnormal chest radiography in a TB-affected community naturally raise concerns regarding active disease; however, microbiological confirmation remains essential to avoid unnecessary treatment. Negative sputum AFB smears, negative GeneXpert MTB/RIF results, low inflammatory markers, and the absence of constitutional symptoms increase diagnostic confidence that active TB is unlikely, guiding clinicians toward appropriate management strategies.

The incidental finding of congenital diaphragmatic hernia provided a plausible alternative explanation for chronic cough, likely related to gastroesophageal reflux or gastric content irritation. Structural thoracic abnormalities, such as congenital diaphragmatic hernia, should be considered in

the differential diagnosis when respiratory symptoms persist despite negative microbiological tests, especially in TB-endemic regions where clinical suspicion is high. Recognizing these conditions can prevent misdiagnosis and unnecessary antituberculous therapy.⁵

Chronic cough is one of the most common presenting complaints encountered in primary care and frequently prompts evaluation for pulmonary tuberculosis (TB), particularly in endemic regions and among under-served populations.⁶ As the indigenous population carries a higher risk of tuberculosis infection, detailed screening with a holistic approach needs to be carried out simultaneously, while the team faced difficult challenges in poor awareness and community barriers.⁷ In Malaysia, primary care clinicians often face the challenge of balancing early TB detection with the risk of overdiagnosis, especially when symptoms and chest radiography findings are nonspecific. Diagnostic uncertainty may arise when radiological abnormalities, such as pleural thickening, coexist with persistent respiratory symptoms, and microbiological investigations remain negative. Pleural thickening is non-specific and may lead to overinterpretation of the disease, which can be caused by malignancy or benign processes.⁸

Differentiating latent tuberculosis infection (LTBI) from active pulmonary tuberculosis remains challenging in endemic settings, particularly within high-risk indigenous populations.⁹ In this case, strong Mantoux reactivity and abnormal chest radiography in the context of recent TB-related deaths heightened suspicion for active disease. However, repeated negative sputum AFB smears, negative GeneXpert MTB/RIF results, absence of constitutional symptoms, low inflammatory markers, and stable radiographic findings reduced the likelihood of active pulmonary tuberculosis diagnosis. Epidemiological risk alone should not supersede microbiological evidence, as diagnostic anchoring may lead to overtreatment and an increase in the incidence of multidrug-resistant tuberculosis.

Tuberculosis screening among Orang Asli communities requires pragmatic diagnostic approaches that consider stigma, mobility barriers, and high default rates. Although the Mantoux test is inexpensive, the completion of the 72-hour reading is often unreliable despite active outreach efforts. Fear, stigma, and avoidance behavior may limit engagement, even when healthcare teams visit the community directly. Single-encounter investigations, such as interferon-gamma release assays (IGRAs), may offer practical advantages despite their higher cost, particularly in populations with poor follow-up adherence.¹⁰

In this patient, sputum AFB smears were negative despite radiographic abnormalities suggestive of pleural pathology, creating uncertainty regarding the diagnosis of active tuberculosis. GeneXpert MTB/RIF was used to facilitate rapid molecular assessment. While mycobacterial culture remains the diagnostic gold standard, its prolonged turnaround time limits immediate clinical decision-making in community-based primary care settings. The combined use of IGRA and GeneXpert represents a context-adapted strategy that balances diagnostic accuracy with operational feasibility in remote indigenous settings.

The incidental congenital diaphragmatic hernia provided a plausible non-infective explanation for the persistent cough, which was likely related to gastric content irritation or reflux. Hence, a basic chest radiograph is mandatory in health clinics for screening chronic cough. The rare incidence of adult congenital diaphragmatic hernia, at 0.17%-0.20% in the population, may not be the first diagnosis in a patient presenting with a chronic cough.⁴ Structural thoracic abnormalities that contribute to gastric irritation may mimic pulmonary tuberculosis symptoms, such as persistent cough, which may cause a dilemma in diagnosing active tuberculosis in high-risk groups. Multidisciplinary consultation is essential for refining the diagnosis and avoiding unnecessary multidrug therapy.

Primary care clinicians working in rural settings frequently encounter diagnostic uncertainties when managing tuberculosis among high-risk Indigenous populations. Limited access to advanced imaging, delayed laboratory turnaround times, and challenges in patient follow-up require family medicine practitioners to balance public health priorities with individualized clinical judgement. In this case, the need to differentiate latent tuberculosis infection from active pulmonary disease was particularly important, as both undertreatment and overtreatment have significant consequences in communities experiencing ongoing transmission and TB-related mortality.

While tuberculosis remains a key consideration in symptomatic individuals from high-risk populations, clinicians must maintain a broad differential diagnosis for such cases. The incidental finding of congenital diaphragmatic hernia in this patient highlights the importance of evaluating the structural and non-infectious causes of respiratory symptoms. A holistic assessment reduces the risk of diagnostic bias and supports safer and more accurate clinical decision-making.

In conclusion, this case highlights the diagnostic complexity of evaluating chronic cough in tuberculosis-endemic primary care settings, where heightened clinical suspicion must be carefully balanced against the consideration of alternative diagnoses. Radiological abnormalities, such as pleural thickening, may be misleading when interpreted in isolation, particularly when molecular investigations do not support active infection. Recognition of uncommon structural conditions, including congenital diaphragmatic hernia, is essential to prevent unnecessary anti-tuberculosis treatment and ensure patient-centered care. Differentiating latent tuberculosis infection from active disease remains particularly challenging among high-risk Orang Asli populations, where epidemiological risk may inadvertently contribute to stigma or over-medicalization if not guided by objective clinical and microbiological evidence.

Continuity of care and structured follow-up imaging were critical in this case, allowing radiological stability to be recognized and supporting a diagnosis of latent rather than active tuberculosis. In resource-limited rural environments, family physicians play a pivotal role in coordinating culturally sensitive evidence-based care, integrating multidisciplinary input, and adapting diagnostic strategies to local realities. Strengthening clinical reasoning while

maintaining a broad differential diagnosis is essential for achieving accurate treatment decisions, sustaining patient trust, and supporting effective tuberculosis containment within vulnerable indigenous communities.

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