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Diagnostic Utility of GeneXpert in Pediatric Tuberculosis.

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Abstract

Background: Introduction: Tuberculosis (TB) in children remains a major diagnostic challenge, particularly in high-burden countries such as India, due to its paucibacillary nature and non-specific clinical presentation. Conventional diagnostic methods like smear microscopy have limited sensitivity, necessitating the use of rapid molecular diagnostics such as the GeneXpert MTB/RIF assay. The present study aimed to evaluate the diagnostic utility of GeneXpert in detecting pediatric tuberculosis and rifampicin resistance. ***Materials and Methods:*** This prospective observational study was conducted in the Department of Pulmonary Medicine at Mamata Academy of Medical Sciences, Hyderabad, from January to December 2025. A total of 50 pediatric patients (0–18 years) with suspected tuberculosis were included. Clinical samples including sputum, gastric aspirate, bronchoalveolar lavage, pleural fluid, and lymph node aspirates were analyzed using smear microscopy and GeneXpert. Diagnostic performance of GeneXpert was evaluated using smear microscopy as the reference standard. Data were expressed as N (%), and statistical analysis was performed using SPSS version 26. ***Results:*** Among 50 patients, GeneXpert detected Mycobacterium tuberculosis in 23 (46.0%) cases. It identified an additional 9 (18.0%) cases that were smear-negative. Sensitivity and specificity of GeneXpert were 87.5% and 92.6%, respectively, with a diagnostic accuracy of 90.0%. Rifampicin resistance was detected in 4 (17.4%) cases. The most common symptoms were fever (82.0%) and cough >2 weeks (76.0%). ***Conclusion:*** GeneXpert is a rapid, sensitive, and reliable diagnostic tool for pediatric tuberculosis, with the added advantage of early detection of rifampicin resistance, thereby aiding prompt and appropriate management.

Keywords: Pediatric tuberculosis; GeneXpert; Rifampicin resistance; Smear microscopy; Molecular diagnosis.

INTRODUCTION

Tuberculosis (TB) remains a major global public health concern and is one of the leading causes of morbidity and mortality among children, particularly in high-burden countries such as India [1]. Pediatric tuberculosis contributes significantly to the overall TB burden but is often underdiagnosed and underreported due to its paucibacillary nature and non-specific clinical presentation [2]. Children frequently present with symptoms such as prolonged fever, chronic cough, weight loss, and failure to thrive, which overlap with other common childhood illnesses, thereby complicating early diagnosis and timely initiation of treatment [3,4].

Conventional diagnostic methods, including smear microscopy and culture, have notable limitations in pediatric populations [5,6]. Smear microscopy has low sensitivity, especially in children who are often unable to produce adequate sputum samples, while culture, though considered the gold standard, is time-consuming and requires well-equipped laboratory infrastructure [7]. These challenges necessitate the use of rapid, sensitive, and specific diagnostic tools that can facilitate early detection of Mycobacterium tuberculosis and guide appropriate management [8].

The introduction of nucleic acid amplification tests, particularly the GeneXpert MTB/RIF assay, has revolutionized TB diagnostics [9]. This cartridge-based system enables rapid detection of Mycobacterium tuberculosis complex DNA and simultaneous identification of rifampicin resistance within a short turnaround time [10]. The assay has been endorsed by the World Health Organization for use in pediatric tuberculosis due to its improved sensitivity compared to smear microscopy and its ability to detect drug resistance early, which is critical for initiating appropriate therapy and improving outcomes [11].

Despite these advantages, the diagnostic performance of GeneXpert in pediatric populations varies across different clinical settings and specimen types. Therefore, the present study aimed to evaluate the diagnostic utility of GeneXpert in detecting tuberculosis and rifampicin resistance among pediatric patients in a tertiary care hospital setting.

MATERIALS AND METHODS

This hospital-based prospective observational study was conducted in the Department of Pulmonary Medicine at Mamata Academy of Medical Sciences, Bachupally, Hyderabad, over a period of one year from January 2025 to December 2025. The study aimed to evaluate the diagnostic utility of GeneXpert in pediatric tuberculosis. Institutional Ethics Committee approval was obtained prior to the commencement of the study, and informed consent was secured from parents or legal guardians of all participants.

A total of 50 pediatric patients (aged 0–18 years) with clinical suspicion of tuberculosis were included in the study. Suspected cases were identified based on the presence of persistent fever, chronic cough (>2 weeks), weight loss or failure to thrive, history of contact with an active tuberculosis case, or suggestive radiological findings. Children already on anti-tubercular therapy or those with incomplete clinical data were excluded from the study.

Relevant demographic details, clinical history, nutritional status, BCG vaccination status, and presenting symptoms were recorded using a structured proforma. Appropriate clinical samples were collected based on the site of disease, including sputum, gastric aspirate, bronchoalveolar lavage, pleural fluid, and lymph node aspirates, following standard aseptic precautions.

All samples were subjected to smear microscopy for acid-fast bacilli (AFB) using Ziehl–Neelsen staining. Simultaneously, samples were processed using the GeneXpert MTB/RIF assay, a cartridge-based nucleic acid amplification test (CBNAAT), which detects Mycobacterium tuberculosis complex DNA and rifampicin resistance by targeting the rpoB gene. The test was performed as per manufacturer's instructions, and results were reported as MTB detected or not detected, along with rifampicin sensitivity status.

Data were entered and analyzed using Statistical Package for Social Sciences (SPSS) version 26. Categorical variables were expressed as N (%). Diagnostic performance of GeneXpert was assessed in terms of sensitivity, specificity, positive predictive value, negative predictive value, and accuracy, taking smear microscopy as the reference standard. A p-value <0.05 was considered statistically significant.

RESULTS

The study included 50 pediatric patients with suspected tuberculosis. The majority of participants were aged >10 years (20 [40.0%]), followed by 5–10 years (18 [36.0%]) and <5 years (12 [24.0%]). There was a slight male predominance (28 [56.0%]) compared to females (22 [44.0%]). Regarding nutritional status, 19 (38.0%) children were normal, while 17 (34.0%) and 14 (28.0%) had moderate and severe malnutrition, respectively. A history of contact with tuberculosis was present in 21 (42.0%) cases. BCG vaccination scar was observed in 37 (74.0%) children. The most common presenting symptoms were fever (41 [82.0%]) and cough lasting more than two weeks (38 [76.0%]), followed by weight loss (29 [58.0%]) and lymphadenopathy (18 [36.0%]) (Table 1).

Table 1. Demographic and Clinical Characteristics of Study Population (n = 50)

Variable	Category	N (%)
Age Group (years)	<5 years	12 (24.0%)
	5–10 years	18 (36.0%)
	>10 years	20 (40.0%)
Gender	Male	28 (56.0%)
	Female	22 (44.0%)
Nutritional Status	Normal	19 (38.0%)
	Moderate malnutrition	17 (34.0%)
	Severe malnutrition	14 (28.0%)
History of Contact with TB	Present	21 (42.0%)
	Absent	29 (58.0%)
BCG Vaccination	Scar Present	37 (74.0%)
	Scar Absent	13 (26.0%)
Presenting Symptoms	Fever	41 (82.0%)
	Cough >2 weeks	38 (76.0%)
	Weight loss	29 (58.0%)
	Lymphadenopathy	18 (36.0%)

Various clinical samples were obtained for GeneXpert testing, with gastric aspirate being the most common (16 [32.0%]), followed by sputum (14 [28.0%]). Bronchoalveolar lavage samples accounted for 8 (16.0%), while pleural fluid and lymph node aspirates contributed equally with 6 (12.0%) samples each. This distribution reflects the diversity of specimen types used in pediatric tuberculosis diagnosis, particularly in cases where sputum collection is challenging (Table 2).

Table 2. Sample Type Distribution (n = 50)

Sample Type	N (%)
Sputum	14 (28.0%)
Gastric Aspirate	16 (32.0%)
Bronchoalveolar Lavage (BAL)	8 (16.0%)
Pleural Fluid	6 (12.0%)
Lymph Node Aspirate	6 (12.0%)

GeneXpert detected Mycobacterium tuberculosis in 23 (46.0%) cases, while 27 (54.0%) samples were reported as MTB not detected. This indicates a substantial diagnostic yield of GeneXpert in the study population, supporting its role as a rapid molecular diagnostic tool in pediatric tuberculosis (Table 3).

Table 3. Diagnostic Yield of GeneXpert (n = 50)

GeneXpert Result	N (%)
MTB Detected	23 (46.0%)
MTB Not Detected	27 (54.0%)

Among the 23 GeneXpert-positive cases, rifampicin sensitivity was observed in 19 (82.6%) patients, whereas 4 (17.4%) were found to have rifampicin resistance. This highlights the additional advantage of GeneXpert in simultaneously detecting drug resistance, which is critical for early initiation of appropriate therapy (Table 4).

Table 4. Rifampicin Resistance among GeneXpert Positive Cases (n = 23)

Result	N (%)
Rifampicin Sensitive	19 (82.6%)
Rifampicin Resistant	4 (17.4%)

Comparison between GeneXpert and smear microscopy showed that 14 (28.0%) cases were positive by both methods, while 2 (4.0%) cases were smear-positive but GeneXpert-negative. Notably, 9 (18.0%) cases were smear-negative but detected by GeneXpert, emphasizing its superior sensitivity. A total of 25 (50.0%) cases were negative by both methods (Table 5).

Table 5. Comparison of GeneXpert with Smear Microscopy (n = 50)

Smear Microscopy	GeneXpert Positive	GeneXpert Negative	Total
Positive	14 (28.0%)	2 (4.0%)	16 (32.0%)
Negative	9 (18.0%)	25 (50.0%)	34 (68.0%)
Total	23 (46.0%)	27 (54.0%)	50 (100%)

Using smear microscopy as the reference standard, GeneXpert demonstrated a sensitivity of 87.5% and specificity of 92.6%. The positive predictive value and negative predictive value were 87.5% and 92.6%, respectively, with an overall diagnostic accuracy of 90.0%. These findings indicate that GeneXpert is a reliable diagnostic modality with high accuracy in detecting pediatric tuberculosis (Table 6).

Table 6. Diagnostic Performance of GeneXpert (Using Smear as Reference)

Parameter	Value (%)
Sensitivity	87.5%
Specificity	92.6%
Positive Predictive Value (PPV)	87.5%
Negative Predictive Value (NPV)	92.6%
Accuracy	90.0%

DISCUSSION

The present study demonstrated that GeneXpert detected Mycobacterium tuberculosis in 46% of pediatric cases, highlighting its substantial diagnostic yield in a clinically suspected population. Similar findings have been reported in previous studies, where GeneXpert positivity rates ranged between 30–50% depending on specimen type and disease severity. For instance, Nishal et al. reported a positivity of 30.76% in extrapulmonary samples, while other pediatric studies have shown improved detection rates when molecular methods are incorporated alongside conventional techniques [12,13]. These variations can be attributed to differences in sample types, bacillary load, and study settings. The relatively higher yield in the present study may be due to inclusion of multiple specimen types such as gastric aspirate and bronchoalveolar lavage, which are known to improve diagnostic outcomes in children.

In the present study, GeneXpert demonstrated high sensitivity (87.5%) and specificity (92.6%) when compared with smear microscopy, reinforcing its superiority over conventional methods. These findings are consistent with earlier literature, where GeneXpert has shown superior diagnostic accuracy compared to smear microscopy, which is known to have poor sensitivity in pediatric populations. A meta-analysis reported pooled sensitivity and specificity of approximately 87% and 97%, respectively [14], while another study documented sensitivity and specificity as high as 92.7% and 98.9% when compared with culture [15]. The improved performance of GeneXpert can be attributed to its ability to detect even low bacillary loads, which is particularly relevant in children where disease is often paucibacillary.

An important observation in the present study was the detection of additional 18% cases that were smear-negative but GeneXpert-positive, emphasizing its enhanced sensitivity in diagnosing cases that would otherwise be missed. This finding is in agreement with previous studies, which have consistently demonstrated that GeneXpert significantly increases case detection rates beyond smear microscopy. Studies have shown that smear microscopy has markedly lower sensitivity compared to GeneXpert, with some reporting smear sensitivity as low as 39% [15]. Additionally, molecular assays like GeneXpert provide rapid results within hours, compared to culture methods that may take several weeks, thereby facilitating early diagnosis and timely initiation of treatment.

The ability of GeneXpert to simultaneously detect rifampicin resistance is a significant advantage observed in this study, where 17.4% of cases showed resistance. Comparable findings have been reported in previous studies, though with varying prevalence rates depending on geographic and epidemiological factors. For example, a large-scale study reported rifampicin resistance in approximately 7.11% of TB-positive cases [16], while other pediatric studies have also emphasized the importance of early detection of drug resistance for appropriate management. Furthermore, GeneXpert has been widely recognized as a rapid and reliable tool for detecting both tuberculosis and drug resistance, contributing to improved patient outcomes and better control of disease transmission [17].

CONCLUSION

The GeneXpert MTB/RIF assay demonstrated high diagnostic utility in pediatric tuberculosis, with superior sensitivity and specificity compared to smear microscopy and the added advantage of rapid detection of rifampicin resistance. The assay significantly improved case detection, particularly among smear-negative children, facilitating early diagnosis and timely initiation of appropriate therapy. Its ability to provide accurate and rapid results makes it a valuable tool in the routine diagnostic workup of suspected pediatric tuberculosis in tertiary care settings, thereby contributing to improved clinical outcomes and better disease control.

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