Innovative Research in Applied, Biological, and Chemical Sciences

ISSN: 3005-8449

INNOVATIVE RESEARCH JOURNALS

Research Article **IRABCS, vol. 3, issue 1, pp. 82-87, 2025**

Received: May 25, 2025
Revised: June 29, 2025
Accepted: June 29, 2025
https://doi.org/10.62497/irabcs.158

Early Diagnostic Utility of Genexpert for the Detection of *Mycobacterium tuberculosis*

Dr. Usman Aslam[1,2], Dr. Muhammad Shams ul Haq[2], Dr. Muhammad Yousuf[3], Dr. Aleena Batool[2], Dr. Usman Ali[1], Dr. Komal Zaman[4], and Dr. Syed Muhammad Asif Shah[5]

1. *Medical Officer, Department of General Surgery, SUM THQ Hospital Jand, District Attock, Pakistan*
2. *Medical Officer, Department of General Surgery, The Health Service Executive (HSE), Ireland*
3. *Medical Officer, Department of General Medicine, Allama Iqbal Teaching Hospital, D.G.Khan, Pakistan*
4. *WMO, Department of Emergency Medicine, Allied Hospital, Faisalabad. Pakistan*
5. *Medical Officer, Department of General Surgery, DHQ Hospital Bahawalnagar, Pakistan*
6. *Email any correspondence to: Dr. Usman Aslam (Usman.aslam.ghazian@gmail.com)*

How to cite: Aslam U, Haq MS ul, Yousuf M, Batool A, Ali U, Zaman K, et al. Early Diagnostic Utility of Genexpert for the Detection of *Mycobacterium tuberculosis*. IRABCS. 2025; 3(1): 82-87. DOI: https://doi.org/10.62497/irabcs.158 Available from: https://irjpl.org/irabcs/article/view/158

Abstract

Introduction: Tuberculosis (TB) remains a leading cause of morbidity and mortality worldwide, necessitating accurate and rapid diagnostic methods to improve early detection and treatment outcomes.

Objective: To evaluate the diagnostic performance of the GeneXpert MTB/RIF assay compared with sputum smear microscopy in suspected pulmonary TB patients

Methodology: This was a descriptive, cross-sectional study conducted over a period of six months (January to June 2024) at THQ Hospital Jand, District Attock. This cross-sectional study was conducted on 150 patients with suspected pulmonary TB. Demographic and clinical data were recorded, and all participants underwent sputum smear microscopy and GeneXpert MTB/RIF testing. Smear microscopy was used as the reference standard to evaluate diagnostic performance in terms of sensitivity, specificity, predictive values, likelihood ratios, and accuracy. SPSS version 20 was used to analyze the data. Relationships between continuous variables were ascertained using the Pearson correlation coefficient. P-values less than 0.05 were deemed statistically significant.

Results: The mean age of participants was 36.40 ± 17.12 years, with 74 (49.33%) males and 76 (50.67%) females. Most patients resided in rural areas (111; 74.0%) and were married (107; 71.33%), with farmers constituting the largest occupational group (94; 62.67%). Common symptoms included fatigue (140; 93.33%), chest pain (139; 92.67%), unintentional weight loss (137; 91.33%), and coughing up mucus or blood (117; 78.0%). GeneXpert detected TB in 123 (82.0%) patients compared to 85 (56.67%) by smear microscopy. Among smear-negative patients, GeneXpert identified 43 additional cases. The assay showed moderate diagnostic performance with strong agreement with smear microscopy.

Conclusion: GeneXpert MTB/RIF assay offers a rapid and more sensitive alternative to smear microscopy, particularly beneficial for detecting smear-negative TB cases, thereby improving case detection in high-burden settings.

Keywords: Tuberculosis, GeneXpert MTB/RIF, smear microscopy, diagnostic accuracy, rifampicin resistance

Introduction

Mycobacterium tuberculosis is still the cause of tuberculosis (TB), a chronic and dangerous public health hazard, with low-income and developing nations having the greatest rates of infection and death [1]. Formerly referred to as the "white plague," tuberculosis (TB) most often attacks the lungs (pulmonary TB), however it may also spread to other organs, resulting in extrapulmonary TB [2]. Night sweats, fever, persistent cough, and unexpected weight loss are the disease's hallmarks [3]. Since it is carried by airborne particles from infected people, early and precise diagnosis is crucial to stopping its spread [4]. In low- and middle-income countries (LMICs), where diagnostic and healthcare infrastructure is often insufficient, the worldwide burden of tuberculosis (TB) is still high [5].

The WHO reported 27,000 cases of multidrug-resistant TB (MDR-TB) in 2016, making Pakistan one of the high-burden TB nations in the world [6]. Pakistan is one of the nations with the highest TB

burdens in the world. There are significant drawbacks to traditional diagnostic procedures, including as polymerase chain reaction (PCR), culture methods, and sputum smear microscopy [7]. Despite being extensively used and reasonably priced, sputum smear microscopy has limited sensitivity and need a substantial bacterial load for detection [8]. Although culture-based techniques, which are regarded as the gold standard, are more sensitive, they take weeks to provide findings, which delays the start of therapy [9]. This delay promotes the ongoing spread of tuberculosis in the community in addition to negatively impacting the results for individual patients [10].

Molecular diagnostic techniques as the GeneXpert MTB/RIF test have transformed medication resistance screening and TB detection in order to overcome these obstacles [11]. Within two hours, M. tuberculosis and rifampicin resistance may be identified using the GeneXpert system, an automated, cartridge-based nucleic acid amplification test (NAAT) [12]. Its excellent sensitivity in smear-negative and HIV co-infected individuals makes it a useful tool in high-burden settings, and its quick turnaround time enables the early beginning of appropriate medication [13].

Numerous studies have shown how well GeneXpert works to improve TB diagnosis, particularly in high-burden settings [14]. For those with suspected MDR-TB and HIV-associated TB, the WHO recommends using GeneXpert as the first diagnostic test [15]. Although cost, infrastructure, and operational limitations continue to be obstacles, low-income nations like Pakistan have included it in their national TB control programs [16]. Despite these obstacles, GeneXpert may greatly improve early TB identification and management when used in specialized and outlying healthcare institutions.

Research Objective

This study aims to evaluate the early diagnostic utility of GeneXpert for detecting *Mycobacterium tuberculosis* in suspected pulmonary TB cases, focusing on its sensitivity, specificity, and potential role in strengthening TB control strategies in high-burden regions.

Materials and Methods

Study Design and Setting

This was a descriptive, cross-sectional study conducted over a period of six months (January to June 2024) at THQ Hospital Jand, District Attock. The study setting was the pulmonology ward, where patients presenting with respiratory symptoms suggestive of TB were routinely evaluated.

Inclusion and Exclusion Criteria

Inclusion criteria comprised adult male and female patients of all age groups who were clinically suspected of having pulmonary TB and had undergone both sputum smear microscopy and GeneXpert MTB/RIF assay. Exclusion criteria included patients already on anti-TB treatment, those with incomplete laboratory records, extrapulmonary TB cases, and individuals who did not provide consent.

Sample Size

Convenience sampling was used to recruit 150 individuals from the eligible inpatient and outpatient population who presented with suspected pulmonary tuberculosis throughout the research period. The single-center design and the goal of include all consecutive patients who met the inclusion criteria between January and June 2024 served as justifications for the convenience sampling strategy. The study's exploratory goals and attempt to replicate actual diagnostic procedures precluded a formal a priori power or sample size estimate. The final sample size, however, is in line with previous observational studies conducted in the real world that assess GeneXpert's diagnostic efficacy in TB detection [17]. The discussion thread has recognized this restriction.

Data Collection

Data collection involved obtaining sputum specimens from eligible patients following standard biosafety protocols. Laboratory results from both sputum smear microscopy and GeneXpert MTB/RIF assay were recorded. All diagnostic data were entered into a pre-designed proforma to ensure systematic and uniform documentation. Patient demographic information, clinical presentation, and test results were collected. Confidentiality of patient data was maintained throughout the process.

Statistical Analysis

SPSS version 20 was used to analyze the data. The mean ± standard deviation was computed for continuous variables. Relationships between continuous variables were ascertained using the Pearson correlation coefficient. P-values less than 0.05 were regarded as statistically significant. To assess GeneXpert's diagnostic performance, the following metrics were computed: sensitivity, specificity, positive likelihood ratio, negative likelihood ratio, illness prevalence, positive predictive value, negative predictive value, and accuracy.

Ethical Approval

Ethical approval for the study was obtained from the Institutional Review Board (IRB) of THQ Hospital Jand, District Attock. Written informed consent was taken from all participants or their guardians in the case of minors, ensuring adherence to ethical guidelines for human subject research.

Results

The 150 study participants had a mean age of 36.40 ± 17.12 years, including 74 males (49.33%) and 76 females (50.67%), shown in table 1. Most were from rural areas (n=111; 74.0%), while 39 (26.0%) lived in urban settings. A majority were married (n=107; 71.33%), and 43 (28.67%) were single. Regarding occupation, 94 (62.67%) were farmers, 46 (30.67%) were unemployed, and 10 (6.67%) were employed in other professions.

Table 1: Demographic, clinical characteristics, and age distribution of study participants.

Variable	Category	Frequency	Percent
Gender	Male	74	49.33
	Female	76	50.67
Residence	Rural	111	74.00
	Urban	39	26.00
Marital Status	Married	107	71.33
	Single	43	28.67
Occupation	Unemployed	46	30.67
	Employed	10	6.67
	Farmer	94	62.67

Figure 1 shows the distribution of clinical signs and symptoms among the 150 study participants. Coughing up mucus or blood was reported by 117 patients (78.0%), chest pain or pain with breathing/coughing by 139 patients (92.67%), unintentional weight loss by 137 patients (91.33%), and fatigue by 140 patients (93.33%).

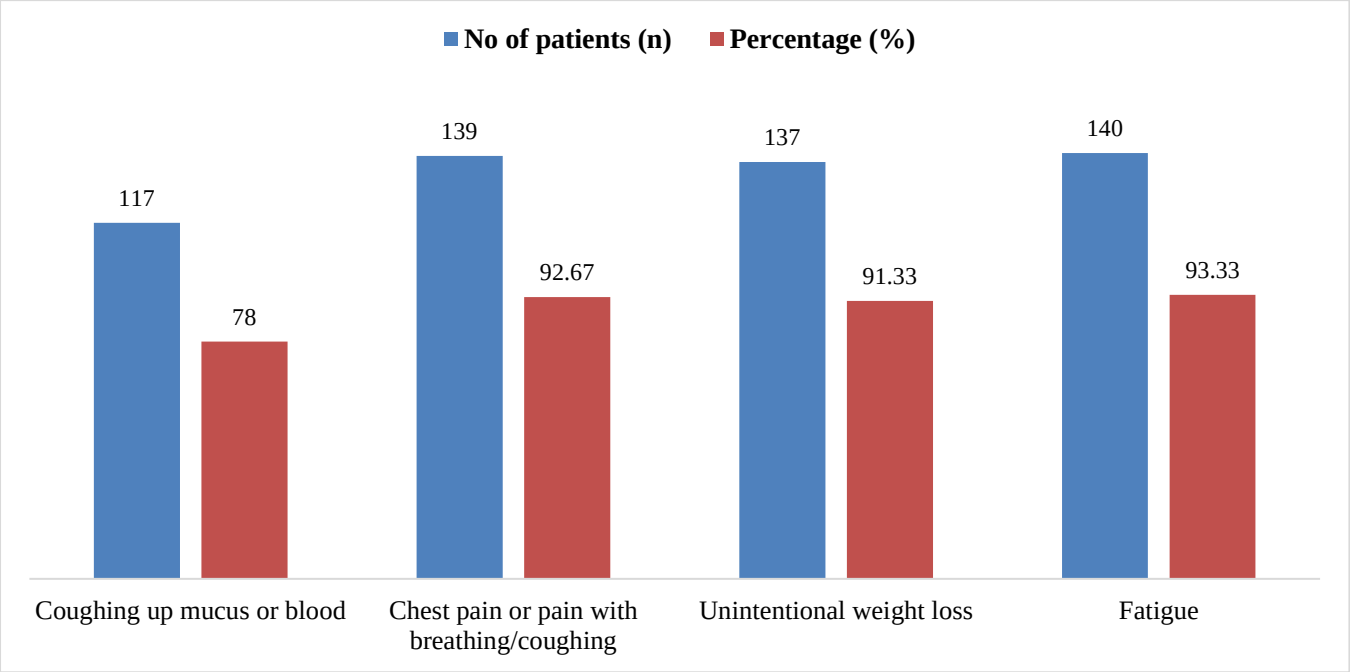

Figure 1: Distribution of clinical signs and symptoms among study participants (n = 150)

Figure 2 presents the cross-tabulation of smear microscopy and GeneXpert results among the 150 study participants. Of the 85 smear-positive cases, 80 were also GeneXpert positive, while 5 were GeneXpert negative. Among the 65 smear-negative cases, 43 were GeneXpert positive and 22 were GeneXpert negative. Overall, GeneXpert detected 123 positive cases and 27 negative cases.

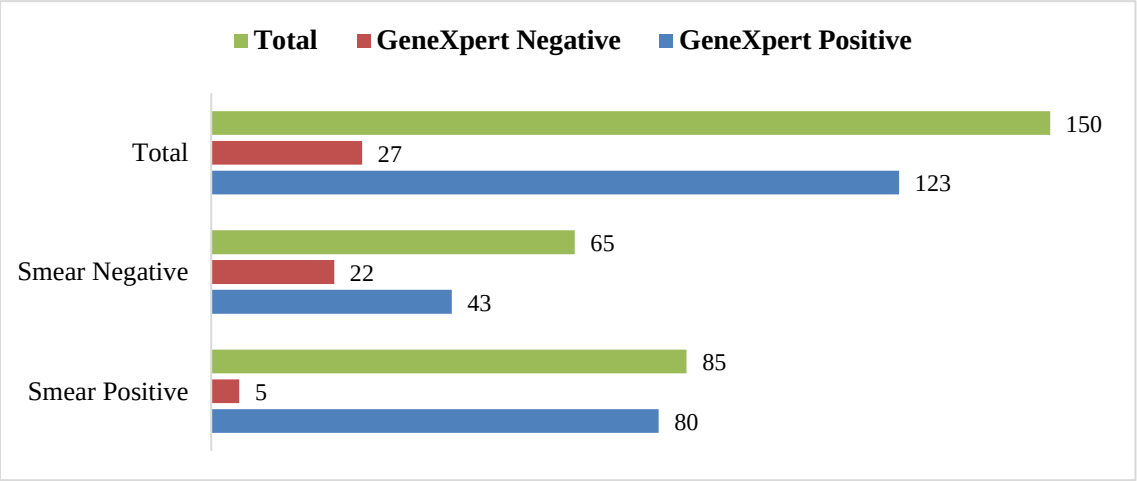

Figure 2: Cross-tabulation of smear microscopy vs GeneXpert results

The GeneXpert MTB/RIF assay's diagnostic performance is compiled in Table 2. The test showed a 70.31% (95% CI: 49.82–86.25) specificity and a 68.29% (95% CI: 59.29–76.39) sensitivity. 2.30 (95% CI: 1.27–4.17) was the positive likelihood ratio, while 0.45 (95% CI: 0.32–0.64) was the negative likelihood ratio. The study population had an 82.0% disease prevalence (95% CI: 74.90–87.79). 91.30% (95% CI: 85.29–95.0) was the positive predictive value, while 32.76% (95% CI: 25.43–41.04) was the negative predictive value. With a Pearson correlation value of 0.46 ($p < 0.001$) and an overall accuracy of 68.67% (95% CI: 60.50–75.98), GeneXpert and smear microscopy data showed a moderately favorable association.

Table 2: Diagnostic performance of the GeneXpert MTB/RIF assay compared to sputum smear microscopy.

Variables	Value	95% CI
Sensitivity	68.29%	59.29 – 76.39
Specificity	70.31%	49.82 – 86.25
Positive likelihood ratio	2.30	1.27 – 4.17
Negative likelihood ratio	0.45	0.32 – 0.64
Disease prevalence	82.0%	74.90 – 87.79
Positive predictive value	91.30%	85.29 – 95.0
Negative predictive value	32.76%	25.43 – 41.04
Accuracy	68.67%	60.50 – 75.98
Pearson correlation coefficient	0.46	$p < 0.001$

Discussion

This research assessed the GeneXpert MTB/RIF assay's early diagnostic value for detecting Mycobacterium tuberculosis in probable pulmonary tuberculosis patients by comparing it to sputum smear microscopy. Comparing GeneXpert to smear microscopy, we found that it had an overall accuracy of 68.67%, with a sensitivity of 68.29% and a specificity of 70.31%. These findings are in line with earlier research that shown that GeneXpert had better detection rates in smear-negative individuals with a high sensitivity (88.6%) and specificity (93.6%) when compared to culture [18]. For example, according to a meta-analysis, GeneXpert showed a pooled sensitivity of roughly 67% and specificity of almost 98% for smear-negative pulmonary TB. Variability was noted across various patient populations and regional settings, indicating similar performance in various contexts [19].

Of the 65 smear-negative patients in our group, 43 (66.15%) tested positive for GeneXpert, demonstrating its usefulness in finding extra instances that traditional microscopy would otherwise overlook. This is consistent with GeneXpert's WHO-endorsed function of enhancing TB case identification, particularly in smear-negative individuals [18]. This kind of improvement is especially crucial in high-burden, resource-constrained environments where transmission management may be greatly impacted by prompt diagnosis.

Our study's excellent positive predictive value (91.30%) indicates how reliable GeneXpert is in confirming tuberculosis when the test result is positive. Similar results have been reported in earlier research, which further emphasizes GeneXpert's strong positive predictive value and supports its use as a trustworthy diagnostic tool for TB confirmation in a range of clinical contexts and specimen types [20]. A negative GeneXpert test does not, however, consistently rule out tuberculosis in this cohort because of our very low negative predictive value of 32.76%. Previous investigations have also noted this restriction, especially in areas with high disease incidence where low bacillary load or technical issues may result in false negatives [21].

Additionally, our demographic information reveals epidemiological patterns that are pertinent to TB prevention. The majority of patients were farmers (62.67%) and rural dwellers (74.0%), which is consistent with research that suggests a higher frequency of tuberculosis in rural areas because of the lack of access to healthcare and increased exposure to risk factors including malnutrition and inadequate ventilation [22]. Weight loss (91.33%), chest discomfort (92.67%), and exhaustion (93.33%) dominated the clinical presentation, which was in line with the symptom profile reported in other TB research [23].

Our findings generally corroborate earlier findings that GeneXpert is a useful early diagnostic tool in high-burden settings, especially for smear-negative individuals; nonetheless, its poor NPV and moderate sensitivity suggest the necessity for supplementary diagnostic techniques to prevent missed diagnoses.

Strengths and Limitations

This study's primary strength is its practical assessment of the GeneXpert MTB/RIF test in a resource-constrained, high-burden environment, which offers important proof of its diagnostic efficacy in comparison to sputum smear microscopy. The results' generalizability to comparable groups is reinforced by the inclusion of both smear-positive and smear-negative probable pulmonary tuberculosis patients, as well as thorough demographic and clinical characteristics. Furthermore, a thorough evaluation of test usefulness is provided by the study's direct comparison of sensitivity, specificity, predictive values, and likelihood ratios. The single-center design and the limited sample size (n=150) are drawbacks, nevertheless, and may restrict generalizability. The

lack of a culture-based gold standard makes it impossible to completely prove actual diagnostic accuracy, and convenience sampling may add selection bias. Additionally, operational viability and cost-effectiveness—two crucial factors for widespread deployment in rural areas—were not evaluated.

Conclusion

This research shows that the GeneXpert MTB/RIF assay has distinct benefits over sputum smear microscopy, especially when it comes to identifying instances that smear testing misses. It is a useful tool for prompt TB diagnosis and treatment start because to its quick turnaround time and capacity to concurrently detect rifampicin resistance. Implementing GeneXpert into current TB control programs, particularly for smear-negative and high-risk patients, can greatly improve case detection and help reduce disease transmission in high-burden settings, even though it cannot completely replace other diagnostic techniques due to certain limitations.

Conflict of interest

The authors state no conflict of interest.

Author Contributions

All authors contributed equally to this work. They were involved in the conception and design, data acquisition, analysis, and interpretation. All authors participated in drafting and critically reviewing the manuscript for important intellectual content. Each author has reviewed the final version to be published and agrees to be accountable for all aspects of the work.

References

1. Silva S, Arinaminpathy N, Atun R, Goosby E, Reid M. Economic impact of tuberculosis mortality in 120 countries and the cost of not achieving the Sustainable Development Goals tuberculosis targets: a full-income analysis. The Lancet Global Health. 2021 Oct 1;9(10):e1372-9. https://doi.org/10.1016/S2214-109X(21)00299-0
2. Pezzella AT. History of pulmonary tuberculosis. Thoracic surgery clinics. 2019 Feb 1;29(1):1-7. https://doi.org/10.1016/j.thorsurg.2018.09.002
3. Verma A, Singh V, Singh Chouhan AP, Dutt PK. TUBERCULOSIS: SYMPTOMS, DIAGNOSIS AND THE FIGHT AGAINST DRUG RESISTANCE. Biochemical & Cellular Archives. 2024 Oct 2. https://doi.org/10.51470/bca.2024.24.1-S.3337
4. Swaminathan N, Perloff SR, Zuckerman JM. Prevention of Mycobacterium tuberculosis transmission in health care settings. Infectious Disease Clinics. 2021 Dec 1;35(4):1013-25. https://doi.org/10.1016/j.idc.2021.07.003
5. Harries AD, Kumar AM, Satyanarayana S, Takarinda KC, Timire C, Dlodlo RA. Treatment for latent tuberculosis infection in low-and middle-income countries: progress and challenges with implementation and scale-up. Expert review of respiratory medicine. 2020 Feb 1;14(2):195-208. https://doi.org/10.1080/17476348.2020.1694907
6. Arif S, Mehboob Q, Arif W. Prevalence and Analysis of Drug Resistance Pattern of MDR-TB in Retreatment Cases at Allied Hospital, Faisalabad, Pakistan. Annals of Punjab Medical College. 2020 Dec 31;14(4):331-5. DOI: https://doi.org/10.29054/apmc/2020.994
7. Garg SK, Tiwari RP, Tiwari D, Singh R, Malhotra D, Ramnani VK, Prasad GB, Chandra R, Fraziano M, Colizzi V, Bisen PS. Diagnosis of tuberculosis: available technologies, limitations, and possibilities. Journal of clinical laboratory analysis. 2003;17(5):155-63. https://doi.org/10.1002/jcla.10086
8. Che-Engku-Chik CE, Yusof NA, Abdullah J, Othman SS, Said MH, Wasoh H. Detection of tuberculosis (TB) using gold standard method, direct sputum smears microscopy, PCR, qPCR and electrochemical DNA sensor: A mini review. Journal of Biochemistry, Microbiology and Biotechnology. 2016 Dec 30;4(2):16-21. https://doi.org/10.54987/jobimb.v4i2.305
9. Chopra KK, Singh S. Newer diagnostic tests for tuberculosis, their utility, and their limitations. Current Medicine Research and Practice. 2020 Jan 1;10(1):8-11. https://doi.org/10.1016/j.cmrp.2020.01.004
10. Shah HD, Nazli Khatib M, Syed ZQ, Gaidhane AM, Yasobant S, Narkhede K, Bhavsar P, Patel J, Sinha A, Puwar T, Saha S. Gaps and interventions across the diagnostic care cascade of TB patients at the level of patient, community and health system: a qualitative review of the literature. Tropical Medicine and Infectious Disease. 2022 Jul 15;7(7):136. https://doi.org/10.3390/tropicalmed7070136
11. Yadav S, Rawal G, Jeyaraman M, Jeyaraman N. Advancements in tuberculosis Diagnostics: a comprehensive review of the critical role and future prospects of Xpert MTB/RIF Ultra technology. Cureus. 2024 Mar 31;16(3). https://doi.org/10.7759/cureus.57311
12. Sahana KS, Prabhu AS, Saldanha PR. Usage of cartridge based nucleic acid amplification test (CB-NAAT/GeneXpert) test as diagnostic modality for pediatric tuberculosis; case series from Mangalore, South India. Journal of Clinical Tuberculosis and Other Mycobacterial Diseases. 2018 May 1;11:7-9. https://doi.org/10.1016/j.jctube.2017.12.002
13. Bartolomeu-Gonçalves G, Souza JM, Fernandes BT, Spoladori LFA, Correia GF, Castro IM, Borges PHG, Silva-Rodrigues G, Tavares ER, Yamauchi LM, Pelisson M, Perugini MRE, Yamada-Ogatta SF. Tuberculosis Diagnosis: Current, Ongoing, and Future Approaches. Diseases. 2024 Sep 3;12(9):202. doi: https://doi.org/10.3390/diseases12090202
14. Williams V, Calnan M, Edem B, Onwuchekwa C, Okoro C, Candari C, Cruz R, Otwombe K. GeneXpert rollout

in three high-burden tuberculosis countries in Africa: A review of pulmonary tuberculosis diagnosis and outcomes from 2001 to 2019. African Journal of Laboratory Medicine. 2022 Aug 30;11(1):1811. https://hdl.handle.net/10520/ejc-ajlab_v11_i1_a1811

15. Sasikumar C, Utpat K, Desai U, Joshi J. Role of GeneXpert in the diagnosis of mycobacterium tuberculosis. Advances in Respiratory Medicine. 2020;88(3):183-8. https://doi.org/10.5603/ARM.2020.0102
16. van den Boom M, Bennani K, Sismanidis C, Gunneberg C, Khawaja L, Safdar MA, Muhwa C, Heldal E, Cirillo DM, Khan AW, Fatima R. 2022 TB programme review in Pakistan: strengthening governance, with better patient diagnosis and treatment. IJTLD open. 2024 Mar 1;1(3):136-43. DOI: https://doi.org/10.5588/ijtldopen.23.0587
17. Patel S, Dadheech M, Maurya AK, Singh J, Purwar S, Rai N, Sarawagi R, Joshi A, Khadanga S, Patel Jr S, Rai NK. Assessment of the diagnostic utility of GeneXpert Mycobacterium tuberculosis/Rifampicin (MTB/RIF) assay in the suspected cases of tuberculous meningitis. Cureus. 2023 Apr 18;15(4). doi: 10.7759/cureus.37761.
18. Arora D, Dhanashree B. Utility of smear microscopy and GeneXpert for the detection of Mycobacterium tuberculosis in clinical samples. Germs. 2020 Jun 2;10(2):81. https://doi.org/10.18683/germs.2020.1188
19. Walusimbi S, Bwanga F, De Costa A, Haile M, Joloba M, Hoffner S. Meta-analysis to compare the accuracy of GeneXpert, MODS and the WHO 2007 algorithm for diagnosis of smear-negative pulmonary tuberculosis. BMC infectious diseases. 2013 Oct 30;13(1):507. https://doi.org/10.1186/1471-2334-13-507
20. Li H, Li Z, Cui X, Liu X, Zhao J, Zhang Y, Wang C, Li B, Fan Y, Han J, Xia Y, Xiong Z, Zou X, Zhu Y, Li M, Lu B, Cao B. Mycobacterium tuberculosis Detection in Diverse Clinical Specimens by GeneXpert MTB/RIF: A Large-Scale Retrospective Study. Infect Drug Resist. 2025 May 8;18:2401-2413. https://doi.org/10.2147/IDR.S514220
21. Jain J, Jadhao P, Banait S, Salunkhe P. Diagnostic accuracy of GeneXpert MTB/RIF assay for detection of tubercular pleural effusion. PLoS One. 2021 Jun 14;16(6):e0251618. https://doi.org/10.1371/journal.pone.0251618
22. Li A, Yuan SY, Li QG, Li JX, Yin XY, Liu NN. Prevalence and risk factors of malnutrition in patients with pulmonary tuberculosis: a systematic review and meta-analysis. Frontiers in Medicine. 2023 Aug 10;10:1173619. https://doi.org/10.3389/fmed.2023.1173619
23. Luies L, Du Preez I. The echo of pulmonary tuberculosis: mechanisms of clinical symptoms and other disease-induced systemic complications. Clinical microbiology reviews. 2020 Sep 16;33(4):10-128. https://doi.org/10.1128/cmr.00036-20