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Applications of molecular biology in the prenatal diagnosis of hereditary diseases.

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ABSTRACT

Introduction: Prenatal diagnosis plays a fundamental role in the early identification of genetic alterations and hereditary diseases during pregnancy. This study aimed to analyze the applications of molecular biology techniques in prenatal diagnosis, highlighting their benefits, limitations, and the role of the biomedical professional in this context. Methodology: This is a qualitative research study developed through a literature review. The search was conducted in the Google Scholar, SciELO, LILACS, and PubMed databases, using the descriptors "Prenatal Diagnosis", "Molecular Biology", and "Mutation", combined with the Boolean operators AND and OR.

After applying the inclusion and exclusion criteria, 10 articles published between 2020 and 2025 were selected. Results: The studies demonstrated that techniques such as PCR, FISH, NGS, exome sequencing, and non-invasive prenatal testing have high sensitivity in detecting fetal genetic alterations. NIPT stood out for reducing invasive procedures and increasing maternal-fetal safety. Exome and genome sequencing allowed the identification of mutations associated with hereditary diseases and chromosomal abnormalities. Recent technologies have also demonstrated greater diagnostic accuracy in complex genetic alterations. The importance of the biomedical professional in the execution, validation, and interpretation of molecular tests was also observed.
Discussion: Advances in molecular biology have increased diagnostic accuracy and strengthened precision medicine. However, challenges related to the cost of technologies, laboratory infrastructure, and the interpretation of genetic variants still persist. Final considerations: It is concluded that molecular biology techniques constitute essential tools for improving the prenatal diagnosis of hereditary diseases.

Descriptors: Prenatal diagnosis; Molecular biology; Hereditary diseases; Next-generation sequencing; Non-invasive prenatal testing.

1 INTRODUCTION

Prenatal diagnosis is an important tool in modern medicine.
allowing for the early detection of genetic and chromosomal alterations, as well as
fetal malformations, even during pregnancy. This approach makes it possible not only to
adequate monitoring of maternal and fetal health, but also genetic counseling and...
safer reproductive decision-making (SANSEVERINO; KESSLER; BURIN, 2020;
SILVA et al., 2022).

Traditionally, prenatal care involves a set of clinical and laboratory actions.
including imaging tests, such as morphological ultrasound, and maternal biochemical tests,
used for screening for fetal anomalies, especially trisomies. Despite their
Despite their relevance, these methods have limitations regarding diagnostic accuracy, which has driven...
the development of more sensitive and specific techniques (YANG; TAN, 2020; PETERSEN et al.,

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2021).

In this context, molecular biology has revolutionized prenatal diagnosis by introducing methods capable of directly analyzing fetal genetic material. Techniques such as Polymerase Chain Reaction (PCR), Fluorescence In Situ Hybridization (FISH) and the Next-Generation Sequencing (NGS) has significantly expanded detection capabilities. of genetic alterations, allowing the identification of everything from aneuploidies to associated point mutations hereditary diseases (BERISHA, 2020).

One of the biggest recent advances is the development of non-invasive prenatal testing. (NIPT), based on the analysis of circulating cell-free fetal DNA (cffDNA) in maternal plasma. This method It exhibits high sensitivity and specificity for screening for chromosomal abnormalities. reducing the need for invasive procedures such as amniocentesis and chorionic villus sampling. chorionic villus sampling, associated with gestational risks (MORTAZAVIPOUR, 2022; MOFFARREJ; BIANCHI, 2023).

In addition to aneuploidies, molecular biology techniques have enabled the identification early onset of several hereditary diseases, including hemoglobinopathies, monogenic diseases and Rare conditions. In Brazil, the importance of diagnosing diseases such as anemia stands out. Sickle cell anemia and thalassemias, especially in populations with a higher genetic prevalence, reinforces the importance of targeted screening in carrier couples (MINISTRY OF HEALTH, 2022; BENDER; HOBBS; WARE, 2021).

The incorporation of technologies such as NGS and chromosome microarrays also It broadened the diagnostic spectrum, allowing the identification of microdeletions, microduplications, and Genetic variants with greater precision. These approaches increase diagnostic yield. especially in cases of fetuses with structural malformations, contributing to a better understanding the genotype-phenotype correlation (BORRELL, 2023).

Despite technological advancements, the use of these tools requires integration between healthcare professionals, adequate laboratory infrastructure, and careful interpretation of Results. Ethical issues, such as the handling of incidental findings and variations in meaning. Uncertainty also makes them increasingly relevant in this scenario (LI; WANG; ZHANG, 2023).

In this context, the role of the biomedical professional in the implementation, execution, and... validation of molecular tests. This professional is involved from the selection of methodologies and the... Quality control, bioinformatic analysis, and data interpretation, following guidelines. established to ensure the reliability of laboratory results (ZHONG, 2021; BUSTIN, 2023).

Furthermore, the consolidation of genomic medicine has contributed to the standardization of

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diagnostic protocols and for expanding access to technologies, including in contexts with Limited resources. Integration between health services, specialized laboratories, and policies. Public access is essential to ensure more accurate, accessible and equitable diagnoses (LEISTNER, 2022; BRAZIL, 2022).

Therefore, the present work aims to analyze the applications of the techniques of Molecular biology in the prenatal diagnosis of hereditary diseases, highlighting its importance. for the advancement of early diagnosis. Specifically, it seeks to investigate the main methods. used in this context, evaluate their benefits and limitations in clinical diagnosis and analyze the The biomedical professional's contribution to the application, interpretation, and validation of these tests, highlighting their... role in improving the detection of hereditary diseases.

2 METHODOLOGY

This study is a qualitative research project, conducted through a literature review. focusing on prenatal diagnosis and its importance for the early identification of diseases hereditary. The bibliographic survey was conducted in the Google Scholar database, SciELO, LILACS, and PubMed databases, without language restrictions, covering publications from the last five years. years (2020 to 2025). The descriptors "Prenatal Diagnosis", "Molecular Biology" and were used. "mutation", obtained from the Health Sciences Descriptors (DeCS), combined with the Boolean AND and OR connectors are used to refine the search.

The inclusion criteria considered articles published between 2020 and 2025, whether review articles or Experimental articles that had a direct relationship to the proposed theme were excluded. paid, those with incomplete content, or those that strayed from the topic. Although the time frame of Although the research was conducted between 2020 and 2025, some earlier references were included, considered essential for the theoretical foundation and historical contextualization of the topic.

3 RESULTS

Based on a structured search conducted in the databases, 832 articles were identified. By Through reading titles and abstracts and applying inclusion and exclusion criteria, 10 were selected. articles (Figure 1) for the analysis corpus

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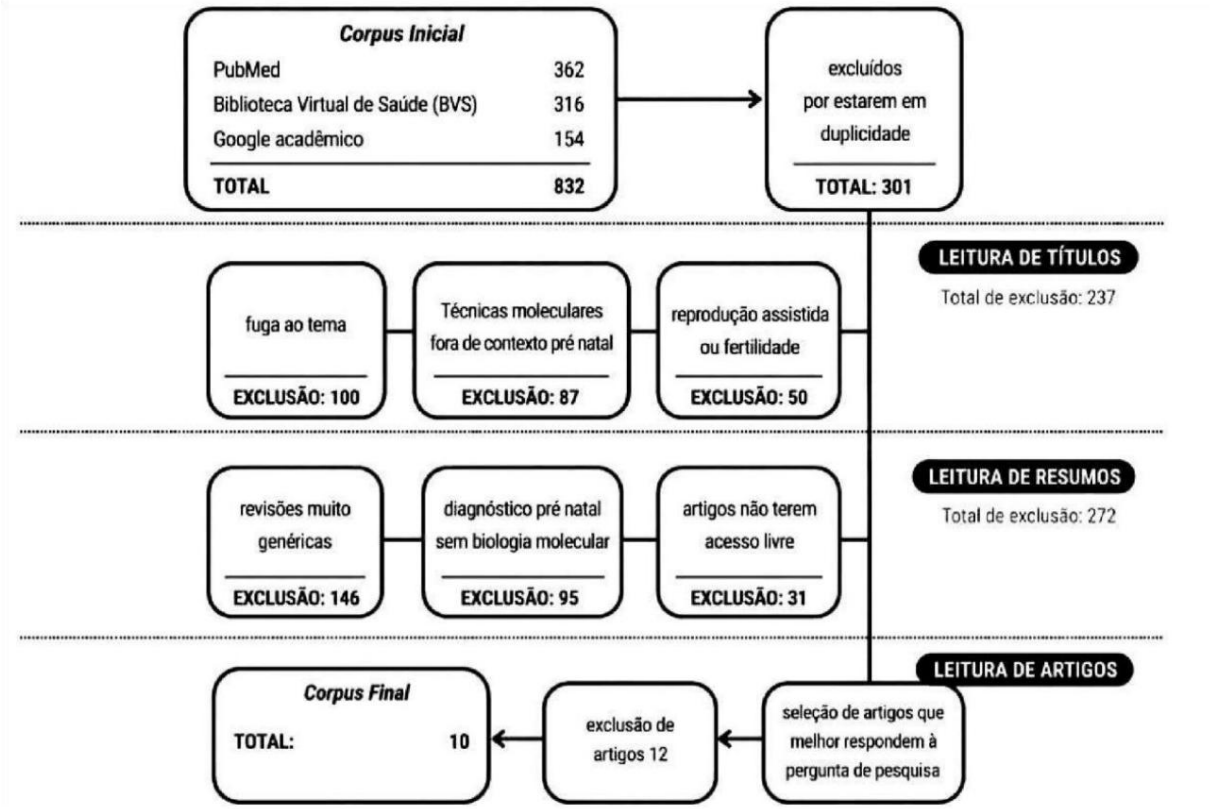

Figure 1. Flowchart of article selection.

Among the 10 selected articles, the main molecular biology techniques were analyzed.
used in the prenatal diagnosis of genetic diseases, such as NGS, exome, genome, PCR and analysis.
of cell-free fetal DNA. The performance of these techniques, their limitations, and their [unclear/unclear] were also evaluated.
Clinical application in pregnant women. Details of the evaluated articles are available (information).
(regarding the title, authors, year of publication, objectives and results) in Table 1

Table 1. Characteristics of the selected scientific articles, according to title, authors, and year of publication.
publication, the objective and the results.

Title	Authors:	Target Year	Results
Usefulness of long-read sequencing in delineating a rare and extensive deletion of the beta-globin gene not detected by sequencing of Sanger syndrome in prenatal diagnosis in a family with hereditary persistence of fetal hemoglobin.	Lee *et al.*	2025 To investigate whether third-generation long read sequencing (LRS) is effective for prenatal diagnosis of hereditary persistence of fetal hemoglobin (PHHF) — a rare form of beta thalassemia that may not be detected by traditional methods such as Sanger sequencing.	LRS improves prenatal diagnosis of rare conditions, such as PHHF, surpassing the... limitations of Sanger sequencing. Chromosomal microarrays (CGH) also help identify deletions associated with beta-thalassemia. Only LRS accurately defines the size and breakpoints of the deletions, with

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				Confirmation possible by PCR.
Unbalanced inherited reciprocal translocation with tetrasomy 18p11.32p11.21 and trisomy 9q34.3 in a fetus revealed by cell-free fetal DNA (cffDNA) testing: cytogenetic and cytogenomic characterization in prenatal diagnosis.	ARDISIA *et al.*	2024	Investigate and characterize chromosomal alterations in a fetus of a pregnant woman carrying a balanced reciprocal translocation, after a high-risk result in NIPT for aneuploidy of chromosome 18, using cytogenetics, FISH, and microarray.	The results highlight the high sensitivity of NIPT with massive parallel sequencing and the usefulness of cytogenetics, cytogenomics, and molecular biology techniques, working synergistically, to characterize and confirm the findings. positive NIPT results.
Prenatal diagnosis and molecular genetic analysis of recurrent maternal trisomy 18 in two consecutive pregnancies.	CHEN *et al.*	2023	Present the prenatal diagnosis and molecular genetic analysis of recurrent maternal trisomy 18 in two consecutive pregnancies.	Prenatal diagnosis and molecular genetic analysis confirmed recurrent maternal trisomy 18 in two consecutive pregnancies. NIPT proved useful for the rapid prenatal diagnosis of recurrent trisomy 18 in these circumstances.
Non-invasive prenatal testing: a revolutionary journey in prenatal testing.	ABEDALTHAGAF I *et al.*	2023	Present the evolution of non-invasive prenatal testing (NIPT), highlighting its technological basis and clinical applications in the detection of fetal genetic abnormalities, especially aneuploidies.	NIPT revolutionized prenatal screening because it is non-invasive and offers high accuracy compared to... other methods. Despite the advances, it is necessary to carefully evaluate the benefits, risks, and evidence before expanding their use, especially in Rare conditions. Expansion should be ethical, evidence-based, and with appropriate counseling.
Prenatal exome sequencing analysis in fetuses with various ultrasound findings.	BORRELL *et al.*	2023	Evaluate the use of Exome sequencing to detect copy number variations and genetic mutations associated with developmental disorders in fetuses with mild ultrasound findings, but without a prior indication for this test.	Fifty-nine pregnant women who underwent invasive examinations due to fetal ultrasound abnormalities were evaluated. They were identified different types of anomalies and 6 cases of common aneuploidies were excluded from the analysis.
Advances in molecular medicine: unraveling the complexity of diseases and pioneering precision medicine.	BUSTIN; JELLINGER	2023	Application of molecular techniques to detect and analyze genetic and molecular biomarkers in patient samples, defining the era of personalized medicine.	As AI technology evolves, its integration into molecular medicine will drive significant improvements, shaping the future of healthcare toward more precise and personalized care.

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Next-generation sequencing technology: current trends and advancements.	SATAM *et al.*	2023	Comprehensive overview of NGS technology, highlighting its transformative impact in diverse areas, including clinical genomics, cancer research, infectious diseases, surveillance, and microbiome analysis.	The future of NGS is highly promising, with the potential to drive scientific discoveries and generate significant advances in health, agriculture, the environment, and various other areas.
Emerging technologies for prenatal diagnosis: the application of whole genome sequencing and... RNA	LIU; VOSSAERT	2022	To provide an overview of recent advances and demonstrate how they can overcome the limitations of current clinical methods in the prenatal diagnosis of genetic diseases.	The experience of postnatal genetic diagnosis tends to guide and shape analytical expectations regarding its application in the prenatal context.
Application of next-generation sequencing in laboratory medicine.	ZHONG *et al.*	2021	Presenting current sequencing technologies (short and long reading) and highlight the clinical application of NGS in hereditary diseases, oncology, and infectious diseases.	NGS technologies and bioinformatics tools will continue to evolve and become the primary means of diagnosis and the standard of care for genomic analyses, meeting the growing demands of precision medicine.
Non-invasive prenatal testing: current perspectives and future challenges	CARBONE *et al.*	2021	Reduce both the incidence and prevalence of hereditary diseases, which have a strong impact on the psychological and economic aspects of people's lives and represent a burden for national health systems.	Due to technical issues, NIPT remains a screening test and not a diagnostic test. However, the increasing accuracy of the methodology is promising, including for the widespread use of this test in screening for monogenic diseases.

4. DISCUSSION

Prenatal diagnosis plays a key role in the early identification of abnormalities.
genetic and hereditary diseases, contributing to the proper monitoring of pregnancy and to
Safer clinical decision-making. In recent years, advances in molecular biology...
They have significantly expanded diagnostic capabilities during pregnancy, allowing for greater...
precision in detecting fetal anomalies through increasingly sensitive molecular techniques
and specific (ABEDALTHAGAFI et al., 2023; LIU; VOSSAERT, 2022).

The results of this review show that molecular biology techniques have played a significant role.
a central role in advancing the prenatal diagnosis of hereditary diseases, especially with the
incorporation of technologies such as next-generation sequencing (NGS), non-prenatal testing

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invasive penicillin testing (NIPT) and exome sequencing. These findings are consistent with the literature, which highlights a significant transition from traditional methods to approaches more sensitive and specific molecules, expanding the ability to detect changes. fetal genetics (ZHONG et al., 2021; SATAM et al., 2023).

The high sensitivity of NIPT observed in the analyzed studies reinforces its relevance as prenatal screening tool, especially for aneuploidies such as trisomy 18. These The results corroborate the findings of Abedalthagafi et al. (2023), who point to NIPT as a Revolutionary technology due to its precision and non-invasive nature. However, as Also highlighted by Carbone et al. (2021), the test should still be interpreted as a method. screening, requiring confirmation through invasive and complementary techniques, which This highlights the importance of integrating different diagnostic methodologies.

Furthermore, the use of exome and genome sequencing has demonstrated great potential for to identify mutations associated with fetal developmental disorders, especially in cases with Inconclusive ultrasound findings. This result is consistent with Borrell et al. (2023). which demonstrate the increase in diagnostic yield with the use of these technologies, which allows The detection of genetic variants that would not be identified by conventional methods. Thus, a significant advance in the correlation between genotype and phenotype is observed, which contributes for more accurate diagnoses.

Another relevant point identified was the superiority of newer technologies, such as... Long read sequencing (LRS) is used in the detection of complex genetic alterations, such as large gene deletions. As demonstrated by Lee et al. (2025), this approach overcomes the limitations of traditional sequencing, such as the Sanger method, allowing for greater precision in identification of breakpoints and structural variations. This advance reinforces the trend of Continuous evolution of molecular techniques in the context of prenatal diagnosis.

In this context, the role of the biomedical scientist in the application of molecular biology techniques stands out. Prenatal diagnosis. The use of advanced methodologies, such as NGS, NIPT, and sequencing. Exome sequencing requires the involvement of qualified professionals to perform the tests and ensure proper control. quality control and analysis of the generated data. Thus, the biomedical professional acts directly in ensuring the... reliability of laboratory results, especially given the complexity of the analyses. genetics and the need for interpretation based on scientific evidence (ZHONG et al., 2021; BUSTIN; JELLINGER, 2023).

Despite the progress, the studies also highlight important limitations. Among them, the following stand out: if the high cost of technologies, the need for specialized infrastructure and the complexity in interpretation of variants of uncertain meaning. These findings are consistent with those of Liu and

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Vossaert (2022), who emphasize that, although emerging technologies expand the potential

However, while there are still technical and analytical challenges to overcome for its comprehensive diagnosis, there are still challenges to overcome.

clinical implementation.

In addition to technical limitations, ethical issues are also becoming increasingly relevant. especially with regard to the handling of unexpected genetic discoveries in the context of prenatal diagnosis. As discussed by Abedalthagafi et al. (2023), the expansion of the use of NIPT should be conducted cautiously, based on scientific evidence and accompanied by... Proper genetic counseling is essential to ensure that the benefits outweigh the potential risks. psychological and social impacts on patients.

Regarding practical implications, the results of this research indicate that... The incorporation of molecular biology techniques in prenatal diagnosis contributes significantly for precision medicine, allowing for earlier interventions and targeted. In addition, these technologies assist in clinical decision-making and in Genetic counseling, promoting better reproductive planning and risk reduction. (BUSTIN; JELLINGER, 2023).

Thus, it becomes evident that the biomedical professional plays a strategic role in the consolidation of Genomic medicine and the advancement of prenatal diagnosis of hereditary diseases. Its role integrated into multidisciplinary teams, it contributes to the safe and effective implementation of these Technologies in clinical practice. Furthermore, the continuous training of these professionals becomes... Essential for keeping up with the evolution of molecular techniques and ensuring the quality of services. laboratory tests (SATAM et al., 2023; ZHONG et al., 2021).

From a theoretical perspective, the advancement of these methodologies reinforces the consolidation of genomics as a fundamental pillar of modern medicine. The trend is that NGS and other technologies will continue. evolving and becoming standard in genetic diagnosis, further expanding access to these tools in different clinical contexts (ZHONG et al., 2021).

Finally, the findings of this review reinforce that molecular biology techniques are... significantly transforming prenatal diagnosis, increasing the accuracy in detection of hereditary diseases and contributing to the consolidation of precision medicine. However, for For these advances to be effectively incorporated into clinical practice, further progress is still needed. of challenges related to the standardization of methods, the interpretation of genetic variants, and the expanding access to technologies. In this context, future studies should focus on improvement of bioinformatics tools and in the clinical validation of these methodologies, especially in the diagnosis of monogenic diseases and rare conditions, in order to make the prenatal diagnosis is becoming increasingly safe, accessible and effective (SATAM et al., 2023; LIU;

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VOSSAERT, 2022).

FINAL CONSIDERATIONS

This study allowed us to analyze the applications of molecular biology in prenatal diagnosis of hereditary diseases, highlighting the relevance of these techniques for the early detection of Genetic and chromosomal changes during pregnancy. Based on the literature review conducted, It was observed that methods such as PCR, FISH, NGS, exome sequencing, and prenatal testing did not... Invasive penicillin testing (NIPT) represents significant advances in diagnostic medicine, providing greater Sensitivity, specificity, and safety in fetal assessment.

The results analyzed demonstrated that molecular technologies have been contributing to the Early identification of hereditary diseases, including aneuploidies, hemoglobinopathies, and other diseases. monogenic, allowing for better pregnancy monitoring and genetic counseling. more assertive. Furthermore, it was found that NIPT revolutionized prenatal screening by to reduce the need for invasive procedures, decrease maternal and fetal risks, and expand the diagnostic accuracy.

Despite technological advancements, limitations related to the high cost of [technology] still exist. methodologies, the need for specialized laboratory infrastructure, and the interpretation of Genetic variants of uncertain significance have also been identified. Related ethical challenges have also been identified. to the use of these technologies, especially regarding the management of incidental findings and the expansion of Access to molecular testing.

In this context, the importance of the biomedical professional in the execution, validation, and Interpretation of molecular tests, ensuring quality, reliability and safety of results. results. The biomedical scientist plays a fundamental role in the development of genomic medicine and in Integration of new technologies into clinical diagnosis.

Finally, it is concluded that molecular biology plays an essential role in improving prenatal diagnosis, contributing to more precise, personalized, and preventative medicine. It is suggested that future research explore expanding the clinical applicability of sequencing. genetic, as well as strategies that promote the democratization of access to technologies molecular technologies in the healthcare system.

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