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Between Molecular Precision and Healthcare Equity: A Multidisciplinary Analysis of Genetic Counseling

Between Molecular Precision and Care Equity: A Multidisciplinary Analysis of Genetics Counseling

Between molecular precision and equity in care: a multidisciplinary analysis of genetic advice

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ABSTRACT: Genetic counseling has become an essential tool in contemporary clinical practice, especially in the context of preventing hereditary diseases. This article aims to analyze the contribution of genetic counseling to the prevention of hereditary diseases, with an emphasis on Autism Spectrum Disorder, hereditary neoplasms, and sickle cell anemia. This is a narrative literature review, with an active search in the SciELO, PubMed, and Google Scholar databases, using descriptors such as "genetic counseling," "hereditary diseases," and "prevention." The results indicate that genetic counseling contributes significantly to the early identification of hereditary risks, the estimation of recurrence of genetic conditions, informed family planning, and the reduction of uncertainties associated with diagnosis. However, structural barriers related to the scarcity of specialized services in the Brazilian Unified Health System (SUS), the lack of professional training, and regional and socioeconomic disparities in access are evident. It is concluded that genetic counseling represents a multidimensional care technology, whose effectiveness depends on the integration of molecular precision, psychosocial support, and equitable public policies. Strengthening genetic counseling in Brazil requires expanding the service network, professional training, and the inclusion of genetics in health science curricula.

Keywords: Genetic counseling. Hereditary diseases. Prevention. Clinical genetics. SUS.

ABSTRACT: Genetic counseling has become an essential tool in contemporary clinical practice, especially for preventing hereditary diseases. This article aims to analyze the contribution of genetic counseling to the prevention of hereditary diseases, with emphasis on Autism Spectrum Disorder, hereditary neoplasms, and sickle cell anemia. This is a narrative bibliographic review with active searches in the SciELO, PubMed, and Google Scholar databases, using descriptors such as "genetic counseling", "hereditary diseases", and "prevention". The results indicate that genetic counseling significantly contributes to the early identification of hereditary risks, the estimation of the recurrence risk of genetic conditions, informed family planning, and the reduction of diagnostic uncertainties. However, structural barriers are evident, including the scarcity of specialized services in the Brazilian public health system (SUS), a lack of professional training, and regional and socioeconomic disparities in access. It is concluded that genetic counseling is a multidimensional healthcare technology whose effectiveness depends on the integration of molecular precision, psychosocial support,

and equitable public policies. Strengthening genetic counseling in Brazil requires expanding the service network, enhancing professional training, and integrating genetics into health curricula.

Keywords: Genetic counseling. Hereditary diseases. Prevention. Clinical genetics. SUS.

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1. INTRODUCTION

Advances in genetics and molecular biology have promoted significant changes in clinical practice, especially with regard to the prevention, diagnosis and treatment of Hereditary diseases. In this context, genetic counseling stands out as a This is an essential strategy, as it enables the identification of genetic risks and assists individuals and Families in informed decision-making. According to the National Center for Biotechnology According to NCBI (2018), genetic counseling consists of a process of communication focused on understanding the risks related to the occurrence or recurrence of genetic conditions within the family.

The incorporation of genetic testing into clinical practice has increased the demand for professionals. qualified to interpret laboratory results and advise patients about the implications. Medical and psychosocial aspects of hereditary diseases. In this context, genetic counseling It has become integrated into healthcare as an interdisciplinary practice, keeping pace with advancements. of molecular diagnostic technologies and personalized medicine (SILVA et al., 2022).

In addition to contributing to diagnosis, genetic counseling plays a role. fundamental in the prevention of hereditary diseases. Early identification of individuals with Risk allows for the adoption of preventive measures and continuous monitoring, favoring Early interventions and better clinical outcomes (OLIVEIRA et al., 2023).

In the context of public health, especially in women's and family health care, the Genetic counseling plays a prominent role. According to the Good Practices Portal of Fiocruz, this approach contributes to informed reproductive decision-making and conscious, based on scientific evidence (FIOCRUZ, 2023).

Furthermore, genetic counseling has significant impacts on emotional aspects and psychological factors of patients and their families. According to Santos et al. (2022), a Proper genetic counseling helps reduce anxiety and promotes understanding of Genetic risk, contributing to safer and more informed health decisions.

Given this context, the relevance of genetic counseling as a tool becomes evident. fundamental to preventive and personalized medicine. In this sense, the following is presented. Research problem: how does genetic counseling contribute to prevention

What is the role of hereditary diseases in clinical practice?

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2. THEORETICAL FRAMEWORK

Genetic counseling (GC) is a specialized communication process that aims to...
To provide information, support, and guidance to individuals and families who are at risk of or have presented symptoms.
to develop hereditary genetic conditions. This practice integrates knowledge of
medical genetics, psychology, and bioethics, allowing patients to understand the
The nature of the genetic condition, its inheritance mechanisms, the risks of recurrence, and the...
possibilities for prevention, diagnosis, and treatment. According to Sheldon Reed, who is responsible for
Introducing the term "genetic counseling" in the 1940s, the central objective of this
The approach is to help individuals make informed decisions that are compatible with their needs.
Personal and family values. In this context, the AG (General Assembly) is not limited to the transmission of information.
It doesn't use techniques, but seeks to promote patient autonomy in frequently encountered situations.
marked by uncertainties and significant emotional impacts (Martins; Menezes, 2022).
In addition to its informative function, genetic counseling plays a role
fundamental to humanizing healthcare, especially in light of advances in
molecular genetics and precision medicine. The process involves collecting and analyzing the history
family history, the creation of pedigrees, the assessment of genetic risks, and the interpretation of
Laboratory tests, enabling an individualized approach for each case.
However, its relevance transcends biomedical aspects, as it encompasses ethical issues.
Psychosocial and reproductive factors associated with genetic diagnosis. According to Alonso et al.
(2021), genetic counseling contributes to the reduction of anxiety and stigma.
associated with hereditary conditions, strengthening the family support network and promoting
conscious decision-making. In this way, the AG is currently recognized as a
important assistive technology, capable of integrating scientific knowledge, support
Emotional well-being and the promotion of patients' rights constitute one of the pillars of genetics.
contemporary clinical practice. (Alonso et al., 2021; Martins; Menezes, 2022)
Understanding the genetic basis of hereditary diseases is fundamental to the practice of
Genetic counseling, guiding the identification of inheritance patterns and the estimation of...
Recurrence risks. The main Mendelian inheritance patterns include autosomal
Dominant, autosomal recessive, X-linked, and mitochondrial. In autosomal inheritance.
In recessive syndrome, both copies of the gene must be altered for the disease to manifest, being
The presence of asymptomatic carriers in the population is common (SILVA et al., 2020; COSTA et
al., 2024).

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Hereditary risk assessment is a central step in genetic counseling and
It is based on the analysis of family history, the construction of pedigrees, and identification.
of risk factors. Martins and Menezes (2022) highlight that the perception of "risk" is
influenced by clinical, cultural, and social aspects, it is essential that the professional
Consider these dimensions when communicating the probabilities of occurrence or recurrence of
genetic conditions. In the context of sickle cell anemia, the most prevalent genetic disease in
In Brazil, couples where both partners are carriers (Hb AS) have a 25% probability of having a child.
A child with the disease (Hb SS) during each pregnancy (COSTA et al., 2024).

In addition to monogenic diseases, genetic counseling also addresses other conditions.
complex and multifactorial. Santos et al. (2025) demonstrate that most cases of
Autism Spectrum Disorder has a genetic origin, with high heritability and risk of...
Higher recurrence among siblings and monozygotic twins. In hereditary neoplasms, the
Identifying germline mutations in genes such as BRCA1 and BRCA2 allows us to estimate
high risks of developing neoplasms and adopting preventive measures (SOUZA et al.,
2024).

Advances in molecular diagnostic techniques have significantly expanded the
Possibilities for investigating the etiology of hereditary diseases. The conventional karyotype.
It identifies large-scale chromosomal alterations, but has limited resolution. PCR
It detects point mutations, such as that of sickle cell anemia, while CGH-array allows
to identify microdeletions and microduplications not visible in the karyotype, which is useful in
diagnosis of ASD and congenital malformations (BRAZ; AMBROSIO-ALBUQUERQUE,
2022; FONTES; SOUZA, 2022).

Next-generation sequencing (NGS) has revolutionized genetic diagnosis by
allow parallel analysis of multiple genes or the entire exome. Silva and Santos (2025)
They emphasize that NGS expands the ability to identify pathogenic variants, including in
conditions of previously unknown molecular etiology. In the context of malformations
In cases of lethal conditions, such as bilateral renal agenesis, NGS allows for the identification of genetic causes.
previously undetectable, although it also brings new ethical and emotional dilemmas for the
families.

Despite technological advancements, genetic testing continues to serve as a tool.
These are complementary and do not replace specialized clinical evaluation. Fontes and Souza (2022)
They emphasize that the diagnosis of ASD remains fundamentally clinical, since the
The genetic complexity of the disorder prevents the identification of a single responsible alteration.

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In all cases. In the field of oncogenetics, the sequencing of susceptibility genes
Cancer screening allows for risk assessment and informs decisions regarding screening and prophylactic surgeries. (SOUZA et al., 2024).

2.1 Clinical Applications of Genetic Counseling

2.1.1 TEA

Autism Spectrum Disorder has a high heritability rate, with a risk of recurrence.
between siblings ranging from 7% to 20% and concordance rates in monozygotic twins that
can exceed 80%. Santos et al. (2025) demonstrate that genetic counseling
It allows for more accurate estimates of reproductive risk, contributing to family decisions.
better informed. An increased risk is also observed in families with other cases of
ASD or ADHD, suggesting genetic overlap between these conditions (ROCHA et al., 2024).

Advances in molecular diagnostic techniques have broadened the scope of etiological investigation of TEA. Fontes and Souza (2022) highlight that methods such as CGH-array, the research of the syndrome Fragile X syndrome and the sequencing of the MECP2 and PTEN genes constitute tools.
Relevant complementary aspects. However, the authors emphasize that the diagnosis of ASD
It remains fundamentally clinical, given the genetic heterogeneity of the disorder.
It prevents the identification of a single alteration responsible for all cases.

In addition to risk assessment, genetic counseling plays a role in ASD.
important in humanizing care. Alonso et al. (2021) argue that AG helps in
Reducing the social stigma associated with the disorder and strengthening the family support network. Thus, The findings reinforce the need to integrate GA into multidisciplinary approaches.
to care for individuals with ASD, promoting more personalized assistance and humanized.

2.1.2 Hereditary Neoplasms

Genetic counseling in the context of hereditary neoplasms has become established as
central tool of personalized medicine and cancer prevention. Souza et al. (2024)
They demonstrate that the identification of germline mutations in genes of susceptibility to
Cancer allows for the estimation of elevated risks of developing neoplasms throughout life.

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Key genes associated with hereditary syndromes include BRCA1 and BRCA2 (cancer of breast and ovarian cancer), MLH1 and MSH2 (hereditary colorectal cancer), among others. The oncological genetic counseling process involves fundamental steps: data collection and analysis of family history, identification of criteria for genetic testing, sessions Pre- and post-testing, interpretation of results, and management recommendations. Individuals Carriers of pathogenic mutations may have up to an 80% increased risk of developing cancer. Breast cancer in BRCA1/2 carriers and 80-100% for colorectal cancer in Lynch syndrome treated (SOUZA et al., 2024).

Despite technological advances, cultural and infrastructural barriers persist. Souza et al. (2024) warn that telemedicine, while promising for expanding access in regions Remote work requires infrastructure and training that are not always available. Strengthening the Oncogenetics depends not only on technological expansion, but also on capacity building. professional and public policy frameworks that guarantee equitable access.

2.1.3 Genetic Diseases and Congenital Malformations

Sickle cell anemia is the most prevalent genetic disease in Brazil, with autosomal inheritance. recessive. Couples in which both partners are carriers (Hb AS) have a 25% risk of having a child with the recessive gene. son with the disease (Hb SS) in each pregnancy. Costa et al. (2024) and Silva et al. (2020) reinforce this. that the disease suffers from chronic neglect in counseling, despite its high prevalence, highlighting the importance of AG for informed family planning.

A common consensus among the studies is the scarcity of structured services. Genetic counseling in the SUS. Silva et al. (2020) identified that, in Pará, there is a lack of specialized centers force patients to rely on centralized care in the capital. with little continuity of counseling. Silva's research (2020) in Itabaiana-SE reveals that lack of information is the main obstacle to accessing AG, even for families. with children who have been diagnosed.

In the field of congenital malformations, Silva and Santos (2025) present the use of Next-generation sequencing (NGS) in the diagnosis of life-threatening conditions. NGS allows identifying previously undetectable genetic causes, enabling more accurate estimates of Recurrence. However, early detection imposes complex pregnancy decisions on families. which require psychosocial support, which is often lacking in the Brazilian public system.

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2.1.4 Psychosocial and Bioethical Aspects

Genetic counseling transcends the technical dimension of molecular diagnosis.
involving profound psychosocial and bioethical implications. Unlike others
Medical procedures, the AG deals with information about risks and uncertainties that affect not
not only the individual, but also their entire family. Alonso et al. (2021) argue that AG
plays an important role in humanizing care, helping to reduce the
social stigma and in strengthening the family support network.
One of the fundamental principles of genetic counseling is non-directivity, according to...
Which professional should provide clear information so that the patient can make decisions?
autonomous. Respect for autonomy implies guaranteeing the right to information and, equally, the
"Right not to know" — the possibility of refusing to know the results of tests.
genetic factors that may cause psychological distress (MARTINS; MENEZES, 2022; SILVA,
2020). The privacy and confidentiality of genetic information also deserve attention.
Special attention is needed, especially in light of technological advancements.
In the context of social inequalities, bioethical questions related to justice emerge.
distributive in access to genetic services. Silva (2020) and Silva et al. (2020) demonstrate
that vulnerable populations have significantly less access to genetic testing and
qualified advice, perpetuating health inequities. The unequal distribution of
The lack of genetic services in the national territory constitutes a situation of injustice that needs to be addressed.
This is addressed through public policies that guarantee equitable access.

2.1.5 Challenges and Perspectives of Genetic Counseling in the Brazilian Public Health System (SUS).

The implementation of genetic counseling within the Brazilian public health system (SUS) faces structural challenges.
There are educational and support programs that limit the population's access to this technology. Although the
Although the National Policy for Comprehensive Care in Clinical Genetics was established in 2009, its
Implementation in practice is still incipient and heterogeneous across different regions of the country.
(SILVA, 2020; SOUZA et al., 2024). The reference centers are concentrated in the regions
The South and Southeast regions, while the North, Northeast, and Midwest regions show a significant deficit of
professionals and services.
The lack of information and awareness about genetic counseling among
healthcare professionals constitute an additional barrier. Silva (2020) demonstrated that the lack of

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Information is the main obstacle to accessing AG, even for families with children.
diagnosed. Primary care professionals are often unaware of the criteria for
Identifying genetic risk perpetuates underdiagnosis and a lack of guidance.
adequate. Furthermore, the number of professionals trained in medical genetics is insufficient.
to meet the demand of the population.

Regarding perspectives for strengthening the AG within the SUS (Brazilian Public Health System), the following stand out: expansion of the network of
Clinical genetics services with decentralization; implementation of programs of
Telemedicine for remote regions; training of primary care professionals; and inclusion.
of genetics content in undergraduate curricula (COSTA et al., 2024; SILVA et al., 2020;
SOUZA et al., 2024). Genetic counseling is not only a tool of
preventive medicine, but also an instrument of social justice and a guarantee of rights.
reproductive.

3. MATERIALS AND METHODS

This is a literature review study, conducted through an electronic search in the database.
from data from the CAPES Periodicals Portal, with the aim of identifying publications on
Genetic counseling. The search strategy was conducted using relevant descriptors.
The topic was then selected, and eligibility criteria were subsequently applied to select the studies.
The inclusion criteria adopted were scientific articles published in the last five years.
years, available for free in full text (open access) and that addressed aspects
related to genetic counseling in clinical practice, research, or care in
health. Duplicate works, studies unavailable in full, and abstracts were excluded.
scientific events, letters to the editor, editorials and publications not directly related to the objective
of the review.

After applying the selection criteria, 13 articles were included for analysis. The studies
The selected data were organized into a data extraction table developed by the authors.
considering the following variables: article title, year of publication, study objective and
main conclusions. The data analysis was performed descriptively, with the aim of
to identify trends, research approaches, and contributions from recent literature on
genetic counseling.

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4. RESULTS AND DISCUSSION

The application of the eligibility criteria resulted in the selection of 13 scientific articles. Published in the last five years and made available in open access. The studies analyzed They presented different approaches related to genetic counseling, including their application in clinical practice, supporting patient decision-making, identifying hereditary risks, the use of genetic testing, and the ethical challenges involved in the process of advice.

The systematization of the extracted information made it possible to observe that genetic counseling It is widely recognized as an essential tool for promoting understanding of hereditary diseases, assisting in the interpretation of genetic tests, and supporting decision-making. reported by patients and their families. In general, the conclusions of Studies have converged on the importance of multidisciplinary work and clear communication. between professionals and users and the expansion of access to counseling services. genetic, highlighting the positive impact of these services on prevention and early diagnosis. and in the management of conditions of genetic origin. The main data relating to the title, year of The publication, objective, and conclusion of each study were organized in a table **(Table** 1) for to facilitate the comparison and synthesis of the evidence found.

Table 1 – Articles selected from the database: CAPES Journals

Title	Authors	Objective:	Conclusion
Pregnancy at an advanced age and genetic counseling: a study on high-risk conceptions.	Martins and Menezes, 2022	This article presents the concepts of risk surrounding what biomedicine considers advanced maternal age, in order to define what is classified as a high-risk pregnancy.	The research found that the classification of an ideal maternal age for pregnancy is relative and subject to change, depending on the socio-historical context of each society.
Autism spectrum disorder: uncertain origin and impasses in the humanization process.	Alonso *et* al., 2021 Clarify	concepts and particularities, in order to promote a better understanding of the spectrum as a tool for humanization.	There is an urgent need to promote understanding of ASD in light of the exclusion and dehumanization suffered by patients with this conditi Genetic counseling, which aims to reduce social stigma and increase family support, is just as essential as early diagnosis, in order to...

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Title	Authors	Objective	Conclusion
			to reduce cognitive damage.
Autism spectrum disorder: The importance of genetic counseling.	Rocha *et al.,* 2024.	The general objective of this article is to analyze, through a literature review, the characteristics of ASD, in order to provide appropriate family genetic counseling.	After a thorough review of the literature, it is clear that ASD is a syndrome that is difficult to elucidate. There is no single specific biomarker, but rather numerous genetic factors and biological aspects that, together, They can trigger the disease.
Genetic counseling in oncology nursing	Silva and Lopes, 2023	To describe the processes of genetic counseling in oncology, to explain factors about hereditary cancer, and to explain the role of nursing in a multidisciplinary genetic counseling team.	Therefore, it is clear that, among the studies analyzed, there is an informative study on the barriers in genetic counseling processes, the qualification of the professionals involved, the socioeconomic barriers, and the limited access of families to information.
Sickle cell anemia, early diagnosis and genetic counseling in sickle cell disease: a literature review.	Costa *et al.,* 2024	This literature review reports on the consequences of AF in patients' lives and the importance of early diagnosis and genetic counseling.	Therefore, this review presents the clinical picture of AF and reinforces the importance of early diagnosis and genetic counseling, since it is a genetic and hereditary disease.
Sickle Cell Disease, Ancestry and Counseling Genetics: gender relations and reproductive rights in State of Pará, Amazon	Silva, Oliveira and Filgueiras, 2020	At the Pará Regional Blood Center, we investigated 60 people with sickle cell anemia using a semi-structured questionnaire to understand clinical manifestations, socio-racial relationships, gender, income, reproductive rights, genetic counseling, and identity.	Genetic counseling is in its early stages in Pará, and there is no specific sector for it at the Blood Center.
The impact of oncogenetics and the	Souza et al., 2024	This article provides a detailed review of	In conclusion, it can be said that expanding these services is...

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Title:	Authors	Objective:	Conclusion
Relevance of genetic counseling in public health		To highlight the relevance of genetic counseling in the context of public health, emphasizing its importance in expanding the accessibility to genetic diagnostic services, especially for vulnerable populations	This is essential not only for the early detection of genetic mutations, but also for ensuring that preventive care is equitable and accessible, especially in regions where health services are limited.
Genetic counseling: access for families of individuals with congenital diseases.	Silva, 2020	To identify the availability of access to Genetic Counseling (GC) services for families of individuals with congenital diseases in Itabaiana-SE.	The results point to the need for greater investment in publicizing the AG services already provided and in expanding these services.
Autism spectrum disorder (ASD): From genetic classification to molecular diagnosis	Fontes and Souza, 2022.	Aims to address the genetic classification of ASD and review the diagnostic advances of the disorder based on techniques of... molecular biology.	The genetic classification of ASD highlights the importance of considering genetic factors to broaden knowledge about the disorder, since most current diagnostic guidelines are predominantly based on clinical aspects. Furthermore, molecular biology techniques contribute to the etiological identification of cases, making genetic counseling and clinical diagnosis more efficient.
Prenatal diagnosis of bilateral renal agenesis: technological advances and impacts on genetic counseling.	Silva and Santos, 2025	This study aimed to explore the use of advanced technologies in the diagnosis of ARB and its implications. for the genetic counseling.	In conclusion, early diagnosis of ARB, combined with genetic counseling, promotes safer pregnancy decisions.
Genetic counseling and autism: new perspectives	Santos et al., 2025.	Analyzing the relevance of genetic counseling in the context.	Genetic counseling is crucial for clarifying the disorder, being

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Title	Authors	The goal	Conclusion
for diagnosis and intervention		of autism, focusing on genetic factors and the risk of recurrence of disorder.	It is important in both the public and private sectors to inform families about the risks of recurrence and the genetic factors involved, given the high impact of the disorder on the population. general.
Oncogenetic counseling as a care technology in oncology nursing: an integrative review.	Almeida *et al,* 2021	To identify, based on scientific production in the field of nursing, the role of nurses in oncogenic counseling, highlighting their practices and challenges.	Nurses are ideally positioned within the healthcare system; having a knowledge base about the professional's role in the field prepares the nursing workforce to provide skillful care with a focus on oncogenic counseling.
Genetic testing as an auxiliary tool for the diagnosis of Disorder of Autism Spectrum	Braz and Albuquerque, 2022	This article presents a survey of the main tests available to aid in the diagnosis of children suspected of having Autism Spectrum Disorder (ASD), including advantages, disadvantages, and their influence on therapeutic management and genetic counseling, based on... in a literature review.	Autism Spectrum Disorder (ASD) is a multifactorial and heterogeneous condition with increasing prevalence, making it difficult to identify its etiology in each patient. However, the development of knowledge about the genetic and molecular aspects of autism, coupled with the improvement of biotechnological tools for diagnosis, such as PCR, karyotyping, CGH arrays, and sequencing, has contributed to the screening of the condition. These tests, along with the patient's clinical history, help in elucidating the diagnosis.

The analysis of the selected studies reveals a growing consensus regarding the relevance of

Genetic counseling in the management of Autism Spectrum Disorder (ASD), especially

Given the advances in understanding the genetic mechanisms involved in its etiology.

Although ASD is recognized as a multifactorial and heterogeneous condition, the results

The findings demonstrate that genetic factors play a substantial role in their

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development, justifying the incorporation of genetic counseling as a component
essential assistance to affected individuals and their families.

In this context, SANTOS et al. (2025) present robust evidence of the high
heritability of ASD, highlighting that the risk of recurrence is significantly influenced.
based on the degree of kinship, being higher between siblings and monozygotic twins. The authors
They emphasize that genetic counseling allows for more accurate risk estimates.
reproductive, contributing to more informed family decisions. This perspective is
corroborated by ROCHA et al. (2024), who identify a strong genetic contribution to the
occurrence of the disorder and emphasize that knowledge of hereditary factors makes it possible to avoid
not only to guide future pregnancies, but also to broaden family understanding about
condition. Thus, both studies converge in recognizing that counseling
Genetics transcends the mere transmission of technical information, configuring itself as
A strategic tool for family planning and for reducing uncertainty.
frequently associated with the diagnosis.

Meanwhile, advances in molecular diagnostic techniques have expanded
significantly increases the possibilities for etiological investigation of ASD. (Sources and Souza)
(2022) highlight that methods such as chromosome microarray analysis, research on
Fragile X syndrome and the sequencing of the MECP2 and PTEN genes constitute tools.
Complementary materials relevant to clinical practice. According to the authors, the incorporation of these
The methodologies favor the identification of genetic alterations associated with the disorder.
making genetic counseling more assertive and targeted. Similar results are
described by BRAZ AND AMBROSIO-ALBUQUERQUE (2022), who emphasize the usefulness of
Tests such as PCR, karyotyping, CGH-array, and genetic sequencing are used in the diagnostic process.
highlighting their contribution to defining therapeutic conduct and providing family guidance.

However, despite the recognized contribution of molecular tools, studies
The analyses also point to important limitations. FONTES AND SOUZA (2022) emphasize that
The diagnosis of ASD remains fundamentally clinical, given the complexity
The genetic makeup of the disorder prevents the identification of a single alteration responsible for all of the symptoms.
cases. This observation is particularly relevant when considering the heterogeneity
The genetics of ASD hinders the universal application of diagnostic protocols based on...
exclusively in molecular tests. Thus, although technological advances have
While expanding the capacity for genetic research does not replace clinical evaluation, these advancements are not a substitute for clinical evaluation.
specialized, but they act as complementary tools capable of increasing precision.

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diagnosis and the quality of guidance provided to families.

Another relevant aspect identified in the studies refers to the psychosocial impact of genetic counseling. ALONSO et al. (2021) present a differentiated approach to to emphasize that the benefits of this process are not limited to the identification of factors. hereditary factors are not included in the calculation of recurrence risk. The authors argue that the Genetic counseling plays an important role in humanizing care. helping to reduce the social stigma associated with ASD and strengthening the support network. familiar. This perspective broadens the traditional understanding of genetic counseling, often focused on biomedical aspects, highlighting its contribution to Coping with the emotional, social, and psychological repercussions of the diagnosis.

A comparison between the studies reveals that, although different approaches have been used... adopted, there is complementarity between the results. While SANTOS et al. (2025) and ROCHA et al. (2024) focus their analyses on heritability and recurrence risks. familiar, FONTES and SOUZA (2022) and BRAZ and AMBROSIO-ALBUQUERQUE (2022) They direct their discussions toward the diagnostic advances provided by molecular genetics. In turn, ALONSO et al. (2021) broaden the debate by incorporating aspects related to Humanization of care and psychosocial support. This diversity of approaches This demonstrates that genetic counseling has a multidimensional character, encompassing everything from From etiological investigation to family support and reproductive decision-making.

Given this scenario, it is observed that genetic counseling is assuming an increasingly important role. increasingly relevant in assisting people with ASD. In addition to contributing to identification Regarding possible genetic causes and for estimating the risk of recurrence, this practice favors Empowering families, understanding the disorder, and accessing strategies. more appropriate diagnostic and therapeutic approaches. Thus, the findings of the analyzed studies reinforce the need to expand access to clinical genetics services and to integrate the Genetic counseling and multidisciplinary approaches to the care of individuals with ASD, promoting more personalized, humanized and based care. scientific evidence.

4.1 Genetic Counseling – Neoplasms

The analysis of the studies by SOUZA et al. (2024) and ALMEIDA et al. (2021) reveals a theoretical and practical convergence regarding the central role of genetic counseling (GC) in

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Personalized medicine and in the prevention of neoplasms. Both studies support that AG
It is an indispensable tool for the early diagnosis and management of hereditary risks.
corroborating the idea that the identification of genetic mutations is a watershed moment for
patient survival and quality of life. However, when comparing the perspectives
As presented, distinct focuses become apparent which, when integrated, offer a broader perspective.
A holistic view of the challenges to consolidating this practice.

While SOUZA et al. (2024) direct their analysis towards systemic impact and health
collective, emphasizing the relevance of public policies and the need to reduce the
regional disparities, ALMEIDA et al. (2021) focus on the sphere of professional practice,
specifically in the role of oncology nursing. The contrast between these approaches is
Clearly, the first group of authors defends telemedicine as a vector of equity, capable
to overcome geographical and economic barriers for vulnerable populations (Souza et al., 2024).
On the other hand, ALMEIDA et al. (2021) argue that, regardless of the technology, the
The success of the AG depends on the professional's positioning on the front line of care, in
that the empathetic relationship and the nurse's technical skills are crucial for adherence.
from the patient to the counseling process.

There is a remarkable critical synthesis when observing the issue of continuing education. SOUZA et al.
(2024) demonstrate that medical awareness is a predictive factor for the increase in
referrals, which underscores the fragility of the integration between oncology and genetics in
current clinical practice. This finding is complemented by the results of ALMEIDA et al.
(2021), which, through an integrative review, point to a worrying shortage of
qualified knowledge among the nursing class, despite the existence of resolutions
(such as COFEN Resolution No. 468/2014) which legitimize this practice as advanced. Therefore,
The counterpoint here is not technical competence, but a lack of specific training.
while SOUZA et al. (2024) point out flaws in the standardization of family history collection,
ALMEIDA et al. (2021) highlight the urgent need for institutional protocols and
robust academic training so that nursing can fully exercise its functions.
skills.

Furthermore, the comparative analysis shows that, despite technological advances, the barriers remain.
Cultural and infrastructural challenges persist. Souza et al. (2024) warn that telemedicine, although
While promising, it requires infrastructure and training that are often not available in
low-income regions. In parallel, ALMEIDA et al. (2021) suggest that the path to
Strengthening the practice lies in incorporating AG as an assistive technology, which

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It requires that the nurse stop being merely an executor and start acting as a risk manager and educator, which would reduce costs and increase the effectiveness of the health system. In short, the transition to an effective oncogenetic care model requires an approach. multidimensional. It is concluded that the strengthening of oncogenetics does not depend exclusively from technological expansion via telemedicine (SOUZA et al., 2024), nor solely from the individual training of nursing professionals (ALMEIDA et al., 2021), but from the intersection of Both: the implementation of public policies that guarantee access, combined with training. continuous and multidisciplinary, ensuring that genetic counseling ceases to be a A fragmented practice is being transformed into a standard and equitable component of cancer care.

4.2 Genetic Counseling – Public Health

The discussion on the importance of genetic counseling (GC) in contemporary medicine. reveals a complex scenario, in which technological advances and the expansion of knowledge Genetic challenges clash with structural barriers to access and socioeconomic disparities. joint analysis of the studies by MARTINS E MENEZES (2022), COSTA et al. (2024), SILVA et al. (2020), SILVA (2020) and SILVA E SANTOS (2025) allows a critical synthesis on the Perception and implementation of AG in Brazil.

The perception of "risk" is the fundamental starting point in different specialties. MARTINS AND MENEZES (2022) argue that, while in traditional obstetrics the age Advanced maternal age is often labeled as an independent risk factor in genetics. This classification is seen as relative and susceptible to socio-historical variations. Counterpoint is essential because it demystifies the centrality of age, shifting the focus to... analysis of Mendelian inheritance and individual genetic predisposition factors.

Similarly, when we observe monogenic diseases such as sickle cell anemia (SCA), Genetic counseling is taking on the dimensions of a clinical and social urgency. COSTA et al. (2024) Silva et al. (2020) reinforce that AF is the most prevalent genetic syndrome in the world, but suffers from chronic neglect in genetic counseling. The study by SILVA et al. (2020) shows an additional layer of complexity: the intersection of gender, race, and class. Women Those with AF not only face more severe symptoms, but also live with low incomes. significantly smaller, which shows that counseling cannot be dissociated from biocultural context and the reality of structural racism in the country.

A cross-sectional consensus between the studies by SILVA (2020) and SILVA et al. (2020) is the scarcity

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structured care services in the Unified Health System (SUS). The research by SILVA (2020)
In Itabaiana-SE, it is revealed that the lack of information is the main obstacle, including for families.
with children diagnosed. While groups with greater purchasing power are able to circumvent
These limitations, imposed through private services, leave the most vulnerable populations as they are.
Without support, this perpetuates a lack of conscious reproductive planning and suffering.
biopsychosocial. This gap is exacerbated by regional disparity, in which states in the Region
Northern regions, such as Pará, have a marked shortage of specialized centers, depending on...
hematological services are centralized in capital cities and lack continuity in other areas.
advice.

The negligence described by COSTA et al. (2024) in counseling patients with trait
The lack of information about sickle cell disease and individuals with the disease results in chronic misinformation, hindering...
Informed reproductive decision-making and appropriate clinical adaptation of patients.
In contrast to the precarious access to basic services, the study by SILVA and SANTOS
(2025) presents the technological frontier: the use of next-generation sequencing (NGS) in
diagnosis of lethal malformations, such as bilateral renal agenesis. If, on the one hand, NGS and
While artificial intelligence increases diagnostic accuracy, it also brings new ethical dilemmas and...
Emotional factors. Early detection imposes complex and often difficult pregnancy decisions on families.
Sometimes traumatic, requiring a support structure that, as we saw earlier,
It rarely exists in the Brazilian public system.

The comparison between these productions demonstrates the "importance of counseling."
"Genetic" is a concept that, in theory, is accepted, but in practice, is highly stratified.
While biomedicine advances in the molecular identification of serious diseases, the system of
The health system fails to provide the basic support that transforms this technical knowledge into...
Autonomy and reproductive rights. The transition to a comprehensive care model,
defended by COSTA et al. (2024) and SILVA and SANTOS (2025), it must therefore go beyond
Technology requires public policies that promote equity and ensure that information...
Genetics should not be a class privilege, but a right of citizenship for all families.
regardless of region or socioeconomic status.

FINAL CONSIDERATIONS

This literature review allowed us to analyze the contribution of genetic counseling.
for the prevention of hereditary diseases in clinical practice. The findings demonstrate that the

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Genetic counseling (GC) operates on multiple dimensions: in the early identification of risks.
genetic factors, in estimating the recurrence of conditions such as ASD and sickle cell anemia, in
Informed family planning and psychosocial support for patients and their families.
However, significant challenges persist in the Brazilian context. The scarcity of services
specialized in the SUS (Brazilian Public Health System), the regional concentration of genetics centers in the South and
In the Southeast region, the lack of professional training and socioeconomic inequalities limit the
Equitable access to genetic counseling. These barriers perpetuate misinformation and...
lack of adequate guidance for vulnerable populations. It is concluded that the
Genetic counseling is an essential tool in preventive and personalized medicine.
but its effectiveness depends on the integration between diagnostic accuracy, psychosocial support and the
Public policies that guarantee universal and equitable access. Strengthening the AG in Brazil.
This requires expanding the service network, providing professional training, and including genetics in...
curricula in the health field.

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