

Chapter 14 / Capítulo 14

Health and Professional Practice in Argentina: Applied Research in Patient Care, Workforce Training, and Health Interventions (English Version)

ISBN: 978-9915-704-19-7

DOI: 10.62486/978-9915-704-19-7.ch14

Pages: 140-151

©2025 The authors. This is an open access article distributed under the terms of the Creative Commons Attribution (CC BY) 4.0 License.

Prenatal care in the genomic era / El Prenatal en la era genómica

Samila Sousa De Paula¹ ✉, José Vicente Postorivo Nauman¹ ✉

¹Facultad de Medicina y Ciencias de la salud, Universidad Abierta Interamericana.

ABSTRACT

Introduction: with the advancement of fetal medicine, the genomic era has assumed a central role by enabling the reading of the genetic code and the early detection of hereditary anomalies. Technologies such as whole genome sequencing (WGS) and next-generation sequencing (NGS) have facilitated the identification of well-defined genetic disorders as well as multifactorial genetic predispositions during preconception and conception stages. This has allowed carriers and medical teams to assess the risks of mutation transmission and to make informed decisions regarding reproductive health. Moreover, both invasive and non-invasive tests in prenatal care remain key tools for screening and diagnosing genetic alterations at various stages of gestation, promoting essential care for maternal and fetal health.

Method: a literature review was conducted with the aim of analyzing advances in prenatal care within the context of the genomic era. The search was carried out between November 2024 and January 2025 in the PubMed, Google Scholar, SciELO, and Virtual Health Library (VHL) databases, covering publications from the past fifteen years.

Results: the review showed that many couples present with latent hereditary conditions that can be identified during the preconception period, and that both advanced and non-advanced genomic technologies are crucial for detecting numerous anomalies. Additionally, 95 % of professionals consider genetic counseling to be a key component in making informed reproductive decisions.

Conclusions: this review confirmed that genomics has significantly advanced prenatal care; however, its application must be ethical, accessible, and consistently guided by specialized professionals who support families through genetic counseling, helping them to understand, decide, and face potential challenges with responsibility and support.

Keywords: Fetal Anomalies; Genetic Sequencing; Prenatal Diagnosis; Genomic Technologies; Genetic Counseling.

RESUMEN

Introducción: con el avance de la medicina fetal, la era genómica destacó un papel central al permitir la lectura del código genético y la detección temprana de anomalías hereditarias. Tecnologías como la secuenciación de todo el genoma (GS) y la secuenciación de próxima generación (NGS) han facilitado la identificación de trastornos genéticos bien definidos y las predisposiciones genéticas multifactoriales en las etapas preconcepcionales y concepcionales. Esto ha permitido a los portadores y al equipo médico evaluar los riesgos de transmisión de mutaciones y tomar decisiones informadas sobre la salud reproductiva. Además, las pruebas invasivas y no invasivas en el seguimiento prenatal siguen siendo herramientas clave para el rastreo y diagnóstico de alteraciones genéticas en diferentes etapas de la gestación, promoviendo cuidados esenciales para la salud materna y fetal.

Método: se realizó una revisión bibliográfica con el objetivo de analizar los avances del cuidado

prenatal en el contexto de la era genómica. La búsqueda se llevó a cabo entre noviembre de 2024 y junio de 2025 en las bases de datos PubMed, Google Académico, SciELO y Biblioteca Virtual en Salud (BVS), abarcando publicaciones de los últimos quince años.

Resultados: se evidenció que muchas de las parejas presentan condiciones hereditarias ocultas identificables en el período preconcepcional, y que las tecnologías genómicas avanzadas y no avanzadas son de suma importancia para el rastreo de las innumerables anomalías. Además, el 95 % de los profesionales destaca el asesoramiento genético como un componente clave para la toma de decisiones reproductivas informadas.

Conclusiones: esta revisión permitió verificar que la genómica ha progresado el cuidado prenatal de manera significativa, pero que su aplicación debe ser ética, accesible y acompañada siempre por profesionales especializados, que ayuden a las familias mediante el asesoramiento genético a comprender, decidir y enfrentar los posibles desafíos con responsabilidad y apoyo.

Palabras clave: Anomalías Fetales; Secuenciación Genética; Diagnóstico Prenatal; Tecnologías Genómicas; Asesoramiento Genético.

INTRODUCTION

Prenatal care is a set of systematic, periodic actions that precede birth to prevent, diagnose, and treat potential factors contributing to maternal and perinatal mortality.^(1,2,3,4,5)

In recent years, maternal-fetal medicine has made significant advances in research,^(6,7,8,9,10) knowledge, and technologies that improve medical monitoring of pregnancy and in the detection and diagnosis of genetic diseases, ranging from chromosomal and monogenic disorders to congenital anomalies during the preconception and prenatal periods.^(2,3,11,12,13,14,15)

Chromosomal genetic diseases are defined by alterations in the number or structure of chromosomes and can be:^(16,17,18)

- Numerical: e.g., trisomy 21 and monosomy X.
- Structural: e.g., Cri du Chat syndrome.

We can also define monogenic genetic diseases as those caused by mutations in a single specific gene. They can follow inheritance patterns:⁽¹⁸⁾

- Autosomal dominant: e.g., achondroplasia.
- Autosomal recessive: e.g., cystic fibrosis.
- X-linked: e.g., hemophilia.

We can categorize congenital anomalies into three different groups according to their cause:^(3,4,6)

- Polygenic or multifactorial causes: caused by the joint mutation of multiple genes combined with triggering environmental factors. E.g.: cleft palate.
- Genetic causes: caused by an alteration of a major gene or changes in the number or structure of a chromosome. E.g.: Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13).
- Environmental causes: caused by the influence of external factors (teratogens) that can interfere with the development of the embryo or fetus during pregnancy.

Preventing genetic disorders throughout a patient's life should be a global health priority.

The impact of these disorders during childhood and adulthood can be extremely limiting for some people. In addition, the constant search for answers to name the “condition” is arduous. This is because, to make a diagnosis, the medical team must correctly interpret the results and have extensive knowledge of the parents’ genetic history, advanced maternal age (AMA), history of congenital malformations (CM), whether either parent is a carrier of chromosomal abnormalities (CAC), and a history of recurrent miscarriages (RM), which is one of the most common complications of early pregnancy. It is estimated that about 60 % of spontaneous abortions in the first trimester are caused by sporadic chromosomal abnormalities, usually numerical, and that between 6 % and 12 % of stillborn babies also have chromosomal abnormalities.^(3,6,7,8,13)

Increasingly, couples are seeking preconception care. This is related to cultural and social issues specific to the younger generations; however, beyond that, there are also couples with genetic risk factors who seek preconception counseling to avoid the transmission of unwanted conditions.

With new whole-genome sequencing (WGS) and next-generation sequencing (NGS) technologies, it is possible to assess the risks of transmission for well-defined monogenic disorders, multifactorial genetic predispositions, and the couple’s reproductive health. In other words, it allows specific reproductive outcomes to be anticipated, providing the couple with the necessary knowledge about the dangers of having an affected child and offering alternatives, such as prenatal genetic testing or the use of donor gametes to prevent diseases in the child.^(4,7,8,9,10,13)

Genetic testing is also helpful for prenatal diagnosis, especially when ultrasound findings are detected during consultations, which “flag” the pregnancy as high risk. From there, the specialist can begin to perform a series of invasive and non-invasive tests at different stages of pregnancy to look for well-defined genetic disorders.

Ultrasound markers, including nuchal translucency, can be evaluated. In the first trimester, maternal blood samples are collected to detect the free beta-subunit of human chorionic gonadotropin and pregnancy-associated plasma protein. In the second trimester, alpha-fetoprotein and human beta-chorionic gonadotropin are evaluated to identify neural tube defects (non-invasive tests).^(5,6,7,11,12)

These tests can be successfully performed to evaluate abnormalities during pregnancy, providing essential guidance for managing the pregnancy. If the risk of congenital disabilities is high, invasive procedures are recommended, such as:

- Chorionic villus sampling, which can be performed in the first trimester of pregnancy by taking a biopsy of the placenta containing fetal cells.
- Amniocentesis which is performed by taking a sample of amniotic fluid during the second and third trimesters (recommended for pregnant women over 35 years of age).
- Percutaneous umbilical blood sampling, which involves extracting fetal blood through the umbilical cord; however, as this procedure carries a risk of miscarriage, it is rarely used in medical practice.^(5,7,12)

Another first-tier prenatal test for detecting aneuploidies, microdeletions, microduplications, and copy number variations is chromosomal microarray analysis (CMA). This diagnostic test allows for the evaluation and detection of variations in DNA copy number, i.e., the detection of polymorphisms (called duplications or deletions). This test is indicated after invasive tests have been performed.⁽¹⁵⁾

The implementation of these technologies in obstetric clinics not only helps doctors make accurate diagnoses but also allows them to advise families on how to proceed.

A good result can reassure the couple, but an unfavorable result does not necessarily mean the end of their dream of becoming parents or the mandatory termination of the pregnancy. On the contrary, it provides the family with the genetic knowledge necessary to understand the challenges they may face. For moments like this, there is “genetic counseling,” a process in which the specialist communicates with the couple to provide information, helping them understand the possible physical, social, and cultural limitations the child may or may not have. In other words, it is possible to live with some genetic diseases. Of course, the degree of restriction will depend on the extent of the disease and the management each family can undertake.^(6,14,16,17)

METHOD

A literature review was conducted to analyze advances in prenatal care in the genomic era. The search for articles began in November 2024 and was conducted across the PubMed, Google Scholar, SciELO, and Virtual Health Library (VHL) databases, covering publications from the last 15 years.

Descriptors in Spanish, English, and Portuguese were used, either alone or in combination, such as: “prenatal diagnosis,” “genomic medicine,” “congenital anomalies,” “genetic testing,” “preconception diagnosis,” and “genomic technologies.”

The inclusion criteria considered complete: free articles published in the three languages mentioned above that addressed: diagnosis of congenital anomalies during the prenatal period; genomic medicine; prenatal and preconception diagnosis; associated comorbidities; and other complementary diagnostic tests to genome sequencing.

Duplicate articles, publications without conclusive data for the research objective, and texts that did not specifically address prenatal care or genetic screening tests were excluded.

The selection of studies was based on the reading of titles, abstracts, and full texts, in accordance with the established criteria. The analysis of the collected material was conducted qualitatively, prioritizing clinical, diagnostic, and bioethical aspects related to the application of genomics in the prenatal period.

The research question

What is the contribution of the genomic era to the detection of congenital diseases during the prenatal period?

RESULTS

Through searches of PubMed Central, Google Scholar, SciELO, and other sources, we analyzed the 18 articles included in this review. We identified five main themes that reflect the transformations and the importance of prenatal care in the genomic era.

Preconception diagnosis results

Preconception genomic testing can be helpful for both individuals and couples. In this graph, the authors represent the estimated percentage of couples with hidden hereditary conditions detected by whole genome sequencing (WGS); the identification of treatable predispositions

in individuals with relevant family histories; the detection of carriers of recessive mutations through next-generation sequencing (NGS)-based testing; the risk of transmission of X-linked diseases (especially when the mother is a carrier); and the frequency of severe recessive genetic disorders in newborns.^(7,8,13)

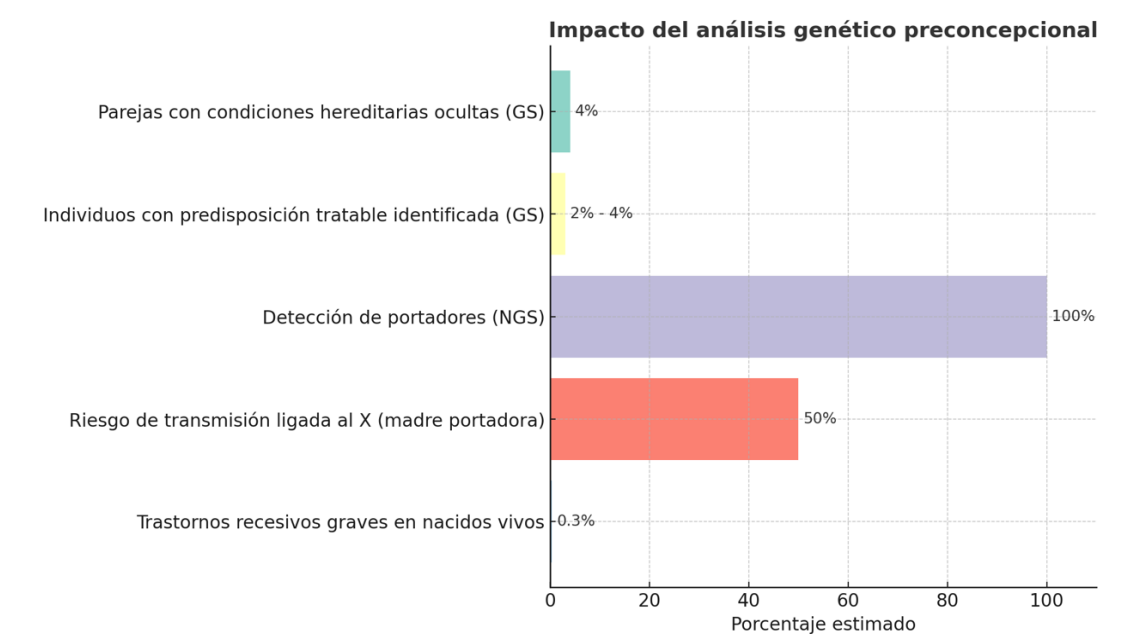


Figure 1. Applications of preconception genomic medicine

Prenatal diagnosis results

The data obtained in the articles analyzed show that accurate prenatal diagnosis requires the combined use of clinical and genetic tools, as well as a detailed assessment of the parents' family history, such as previous congenital malformations in previous pregnancies (CMP), one or both parents being carriers of chromosomal abnormalities (PAC), advanced maternal age (AMA), and a history of recurrent miscarriages (AH).

Table 1. Values found regarding genetic disorders and their relationship with pregnancy		
Category		Percentage (%)
1	Spontaneous abortions (clinically recognized)	15 - 25
2	Spontaneous abortions caused by chromosomal abnormalities	60
3	Numerical chromosomal abnormalities	86
4	Autosomal trisomies	27
5	Polyploidy	10
6	X chromosome monosomy	9
7	Structural rearrangements	2
8	Double or multiple trisomies	0,7
9	First trimester miscarriages with abnormalities	>50 %
10	Stillbirths with chromosomal abnormalities	6
11	Stillbirths with structural abnormalities (autopsy)	25

Below is a table summarizing the literature on genetic disorders and their relationship to pregnancy. In addition, we highlight the prevalence of each (table 1).^(3,4,7)

Invasive and non-invasive tests

Below, in table 2, we can see the data compiled in the literature that mentions the different prenatal diagnostic methods for congenital abnormalities, using a comparative table that highlights non-invasive and invasive tests and advanced prenatal genetic tests at different stages of pregnancy, as well as the function and application of each one.^(5,6,12)

Table 2. Prenatal Tests					
Type of Test	Name of Test	Use and Function	Week of Pregnancy	Application	
Non-invasive	Cell-free fetal DNA (CfDNA)	Detection of fetal aneuploidies (trisomies 21, 18, 13).	≥ 10 weeks	Early and accurate genetic screening.	
Non-invasive	Serum markers (1st trimester)	Assessment of the risk of aneuploidies and neural tube defects.	10-13 weeks	Combined screening with nuchal translucency.	
Non-invasive	Serum markers (2nd trimester)	Detection of neural tube defects and aneuploidies.	16-18 weeks	Second trimester biochemical screening.	
Non-invasive	Obstetric ultrasound	Fetal structural assessment and growth.	Routine throughout pregnancy	Detection of malformations and fetal monitoring.	
Non-invasive	Targeted ultrasound	Detection of subtle malformations with high resolution.	≥ 18 weeks	Detailed study of fetal organs.	
Non-invasive	Nuchal translucency	Measurement of nuchal fold for risk of aneuploidy.	11-13 weeks	Early screening combined with serum markers.	
Invasive	Amniocentesis	Chromosomal analysis, alpha-fetoprotein levels, acetylcholinesterase.	≥ 15 weeks	Diagnostic confirmation of abnormalities.	
Invasive	Chorionic villus sampling	Early fetal analysis (does not detect neural tube defects).	10-12 weeks	Early genetic diagnosis.	
Invasive	Percutaneous umbilical blood sampling	Rapid chromosomal evaluation or diagnosis of fetal anemia.	≥ 18 weeks	Genetic confirmation or intrauterine transfusion.	
Advanced genetics	Chromosomal microarray (CMA)	Detect submicroscopic CNVs.	After invasive testing	Used when there are fetal malformations or suspicion of a structural genetic abnormality.	
Advanced genetics	Genetic panels / NSG	Identify specific monogenic diseases.	After invasive testing	Indicated when there is a specific clinical suspicion, such as a specific syndrome detected by ultrasound.	
Advanced genetics	Whole exome sequencing (WES)	Comprehensive evaluation of complex genetic syndromes.	After invasive testing	Used when there is no clear diagnostic suspicion and the case requires a broader genetic search.	

Genetic counseling and reproductive discussions

In figure 2, We present the fundamental elements of the genetic counseling process, based on their importance as identified by the expert professionals in the analyzed articles. These elements include: informed autonomous decision-making (95 %), detailed assessment of familial genetic risk (90 %), correct interpretation of genetic results (85 %), guidance from the specialist regarding reproductive options (80 %), and emotional and psychological support, which is essential for coping with genetic results (70 %).⁽¹⁴⁾

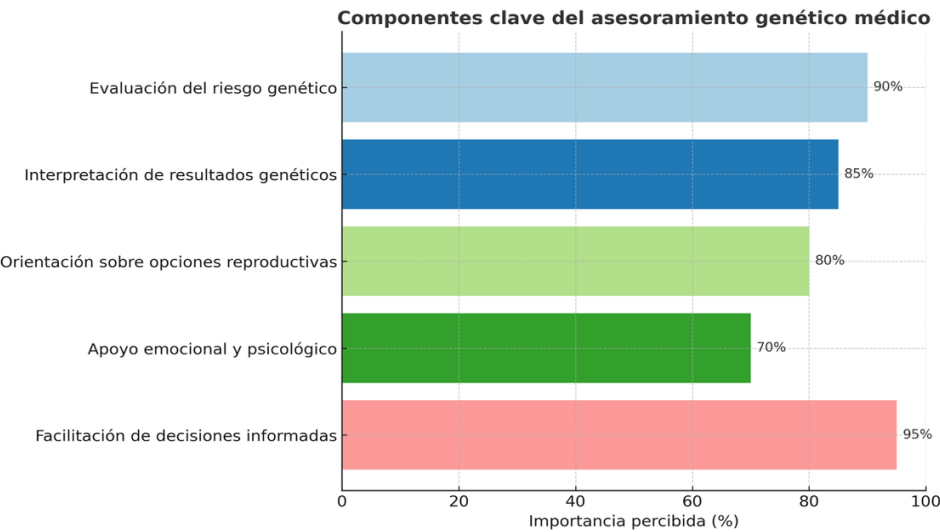


Figure 2. Components of genetic counseling

Ethical and social implications of genomic diagnosis

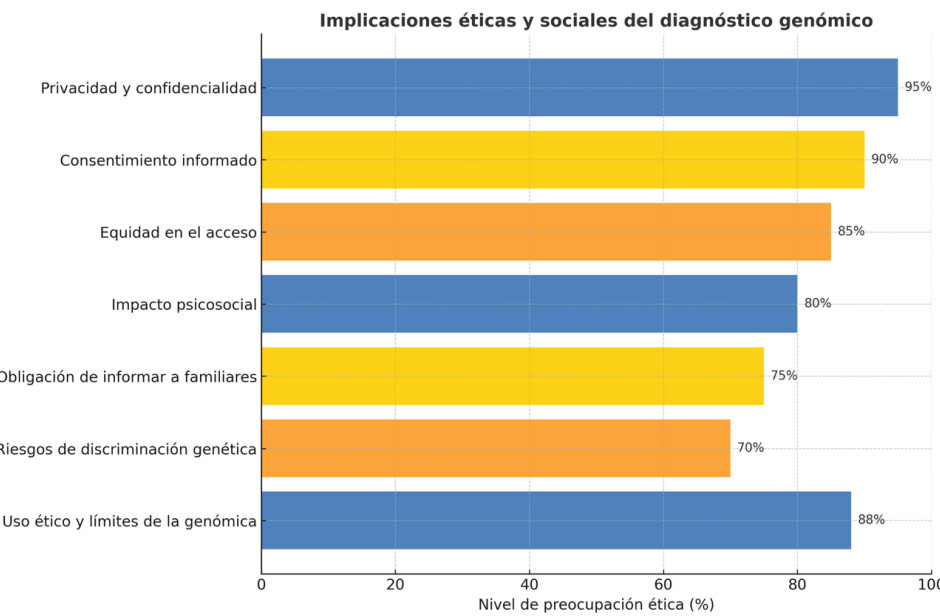


Figure 3. Ethical implications

Figure 3 shows the main ethical and social aspects associated with the implementation of genomic diagnosis, as well as the estimated level of concern they generate in clinical practice and in the specialized literature.^(16,17)

DISCUSSION

Based on the results, the field of genomics has significantly transformed prenatal care in recent years, broadening the diagnostic landscape and enabling interventions to be applied at the early stages of the reproductive process. Throughout this review, it became clear that the incorporation of genetic tools, both preconceptionally and during pregnancy, has significantly increased the ability to identify hereditary risks, congenital malformations, and monogenic or chromosomal disorders that can affect the newborn.

In the preconception setting, the data support the clinical utility of whole-genome sequencing (WGS) and next-generation sequencing (NGS), especially in couples with a family history of genetic diseases. It is estimated that some of these couples may carry previously undiagnosed hereditary conditions, making these tests a valuable tool for anticipating reproductive risks. In addition, identifying carriers of recessive mutations and evaluating X-linked variants can enable effective prevention of serious diseases that manifest early in childhood.^(7,8,13)

During the prenatal period, the combination of biochemical markers, ultrasound, and genetic testing has proven essential for early detection of fetal abnormalities. It has been observed that about 60 % of first-trimester miscarriages are caused by chromosomal abnormalities, mostly numerical, such as autosomal trisomies and X chromosome monosomy. These findings reinforce the need for systematic genetic evaluation when there is a history of recurrent miscarriages or congenital malformations, especially in women of advanced maternal age.

Among the available tests, the detection of cell-free fetal DNA (cfDNA) has gained ground due to its high sensitivity for detecting aneuploidies without putting the fetus at risk (5,6,12). In addition, we can use serum markers, such as alpha-fetoprotein or human chorionic gonadotropin, during screening. However, when suggestive ultrasound findings or altered markers are identified, invasive tests such as amniocentesis or chorionic villus sampling are still necessary to confirm the diagnosis.

The use of technologies such as chromosomal microarray (CMA) and whole exome sequencing (WES) has been key in complex cases, especially when there is no definite diagnostic suspicion. For this reason, they are indicated after invasive tests have been performed, allowing the detection of submicroscopic genetic variants and rare syndromes that would otherwise go unnoticed.

On the other hand, this technological evolution has highlighted the indispensable role of genetic counseling. The various articles analyzed agree that this process should not be limited to interpreting results, but should also provide emotional support, help families understand the clinical and social implications of the diagnosis, and facilitate informed and autonomous reproductive decisions. In fact, it has been reported that up to 95 % of specialists consider the main objective of counseling to be empowering couples to make ethically responsible and informed decisions.⁽¹⁴⁾

However, these diagnostic technologies are not without challenges. The ethical implications of genomic diagnosis were a cross-cutting theme in the studies reviewed. Data confidentiality,

the need for truly informed consent, the psychosocial impact of diagnosis, and equity in access to testing are issues that require rigorous regulation and an interdisciplinary approach. Of particular concern are the handling of unsolicited findings, the possibility of genetic discrimination, and the ongoing debates surrounding the use of genomics for embryo selection.^(16,17)

Against this backdrop, prenatal care is being redefined in countless ways and is becoming a process that encompasses not only clinical issues, diagnosis, and disease prevention, but also an opportunity to monitor the psychological aspects and well-being of each patient closely. It is also essential to highlight the social challenges that fetal medicine still faces regarding the cost-benefit of these advanced technologies and their limitations in public health. Therefore, the results of this review reaffirm the importance of consolidating public policies that guarantee universal access to genomic technologies for the entire population, as this is essential for the prevention and diagnosis of congenital anomalies during the prenatal period.

CONCLUSIONS

Prenatal care has changed significantly in recent years, and this review provides a deeper understanding of how genetic technologies are transforming the detection, interpretation, and management of congenital diseases.

Based on the analysis of the selected articles, it was evident that tools such as genome sequencing, next-generation sequencing, non-invasive and invasive testing, and advanced molecular studies offer new possibilities for both healthcare professionals and families who wish to plan a pregnancy with greater certainty or screen for abnormalities during pregnancy.

The results show that genetic diagnosis, both preconception and prenatal, can be an essential way to anticipate risks, guide decisions, and reduce uncertainty at very sensitive times. But it also became clear that access to these technologies is not yet equal for everyone, and that many ethical issues need careful consideration. Genetic information, when not well explained or accompanied, can cause distress or doubts, which is why the role of genetic counseling is so important.

In summary, this review confirmed that genomics has made significant progress in prenatal care. Still, its application must be ethical, accessible, and always accompanied by specialized professionals who provide genetic counseling to help families understand, make informed decisions, and face potential challenges with responsibility and support.

ACKNOWLEDGMENTS

First and foremost, I want to thank God for everything He has done in my life, for sustaining, strengthening, and guiding me to this important moment.

To my mother, with all my love and deep admiration: thank you for being my rock, for your unconditional love, and for always being by my side. This achievement is also yours.

To my grandparents, who raised me with tenderness, values, and commitment, and to my friends, the family I chose, who accompanied me with affection, sincere words, and constant support.

I also thank my professors for every lesson and for accompanying me on this journey. In particular, I thank Dr. José Postorivo, my thesis advisor, for his guidance, his patience, and for believing in this work from the beginning.

And to all the women in my life—my mother, my grandmothers, my sisters, my friends—thank you for being my inspiration, my support, and my role models. Thank you for shaping, with love and strength, the woman I am today.

From the bottom of my heart, thank you.

BIBLIOGRAPHIC REFERENCES

1. Gregg AR. Futuristic look at genetic and birth defect diagnoses and treatments. *Clin Obstet Gynecol*. 2017 Oct 5; 60(4):867-77. https://journals.lww.com/clinicalobgyn/abstract/2017/12000/futuristic_look_at_genetic_and_birth_defect.20.aspx
2. Levy B, Stosic M. Traditional prenatal diagnosis: past to present. *Methods Mol Biol*. 2019;1885:3-22. <https://pubmed.ncbi.nlm.nih.gov/30506187/>
3. Rappaport VJ. Prenatal diagnosis and genetic screening—integration into prenatal care. *Obstet Gynecol Clin North Am*. 2008 Sep;35(3):435-58.
4. Pietrzyk JJ. Multifactorial dependence of congenital malformations. *Folia Med Cracov*. 1993;34(1-4):97-103. <https://pubmed.ncbi.nlm.nih.gov/8175069/>
5. Dungan JS. Pruebas prenatales para detectar trastornos genéticos y defectos congénitos. *Manuales MSD versión para público general*; 2024. <https://www.msdmanuals.com/es/hogar/salud-femenina/detección-de-trastornos-genéticos-antes-y-durante-el-embarazo/pruebas-prenatales-para-detectar-trastornos-genéticos-y-defectos-congénitos>
6. Argentina.gob.ar. Anomalías congénitas y discapacidad: equipos de salud. Argentina.gob.ar; 2025. <https://argentina.gob.ar/sites/default/files/2020/05/anomalias-congenitas-discapacidad-equipos-salud.pdf>
7. Garcia-Herrero S, Simon B, Garcia-Planells J. The reproductive journey in the genomic era: from preconception to childhood. *Genes*. 2020 Dec 19;11(12):1521.
8. Human Reproduction Update. OUP accepted manuscript. *Hum Reprod Update*. 2020.
9. Rogers A, Lucas De Jong, Waters W, Rawlings LH, Simons K, Gao S, et al. Extending the new era of genomic testing into pregnancy management: a proposed model for Australian prenatal services. *Aust N Z J Obstet Gynaecol*. 2024 Apr 5.
10. Lercher JM, Asan Y, Alberola TM, Rodríguez-Iglesias B, Jiménez-Almazán J, et al. Comprehensive carrier genetic test using next-generation deoxyribonucleic acid sequencing in infertile couples wishing to conceive through assisted reproductive technology. *Fertil Steril*. 2015 Nov 1;104(5):1286-93.
11. Santesmases MJ. Una historia de la autonomía fetal: orígenes de la genética médica y del diagnóstico prenatal. *CSIC.es*. 2025. <https://digital.csic.es/handle/10261/274589>.
12. van der Meij KRM, Sistermans EA, Macville MVE, Stevens SJC, Bax CJ, Bekker MN, et al. TRIDENT-2: national implementation of genome-wide non-invasive prenatal testing as a first-tier screening test in the Netherlands. *Am J Hum Genet*. 2019 Dec;105(6):1091-101.

13. Burguete A, Bermúdez-Morales VH, Madrid-Marina V. Medicina genómica aplicada a la salud pública. *Salud Pública Méx.* 2009 Jan 1;51(Suppl 1):s379-85. https://www.scielo.org.mx/scielo.php?script=sci_arttext&pid=S0036-36342009000900003
14. Andrea I. El asesoramiento genético: evolución, actualidad y retos en la era genómica. *Rev Haban Cienc Méd.* 2021;20(5). http://scielo.sld.cu/scielo.php?pid=S1729-519X2021000500006&script=sci_arttext&tlng=pt
15. Zemet R, Van L. Impact of prenatal genomics on clinical genetics practice. *Best Pract Res Clin Obstet Gynaecol.* 2024 Sep 1;102545.
16. Macarov M, Meiner V, Chalk M, Hochner H, Shkedi-Rafid S. Clinicians' attitudes towards parental choice in the era of advanced genomic tests in pregnancy. *Prenat Diagn.* 2021 Jul 19;41(9):1066-73.
17. Pérez Tamayo R. Algunos aspectos de los problemas éticos generados por el Programa Internacional del Genoma Humano. *Perinatol Reprod Hum.* 2004;18(1):4-9. https://www.scielo.org.mx/scielo.php?script=sci_arttext&pid=S0187-53372004000100002
18. Guillén-Navarro E, Ballesta-Martínez MJ, López-González V. Genética y enfermedad. Concepto de genética médica. *Nefrología.* 2011 May 1;2(1):3-10. <https://www.revistanefrologia.com/es-genetica-enfermedad-concepto-genetica-medica-articulo-X2013757511002585>

FINANCING

None.

CONFLICT OF INTEREST

None.

AUTHORSHIP CONTRIBUTION

Conceptualization: Samila Sousa De Paula, José Vicente Postorivo Nauman.

Data curation: Samila Sousa De Paula, José Vicente Postorivo Nauman.

Formal analysis: Samila Sousa De Paula, José Vicente Postorivo Nauman.

Drafting - original draft: Samila Sousa De Paula, José Vicente Postorivo Nauman.

Writing - proofreading and editing: Samila Sousa De Paula, José Vicente Postorivo Nauman.