GENETIC LITERACY IN ADOLESCENTS WITH GENETIC DISORDERS: A BIOPSYCHOSOCIAL AND CLINICAL APPROACH

ALFABETIZAÇÃO GENÉTICA EM ADOLESCENTES COM TRANSTORNOS GENÉTICOS: UMA ABORDAGEM BIOPSICOSSOCIAL E CLÍNICA

ALFABETIZACIÓN GENÉTICA EN ADOLESCENTES CON TRASTORNOS GENÉTICOS: UN ENFOQUE BIOPSICOSOCIAL Y CLÍNICO

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ABSTRACT

Genetic literacy constitutes an essential component in contemporary medicine, particularly in the management of adolescents with genetic disorders. This population group faces complex challenges encompassing biomedical, psychological, and social dimensions. The aim of this chapter is to analyze the role of genetic literacy in adolescents with genetic diseases, its relationship with psychological adaptation, and its clinical implications. The components of genetic literacy, associated factors, empirical evidence, and the role of genetic counseling are reviewed. Evidence suggests that, although levels of genetic literacy are often low, adolescents may present adequate levels of psychological adaptation, indicating the influence of psychosocial factors. An integrative biopsychosocial model is proposed to optimize clinical and educational interventions.

Keywords: Genetic Literacy. Adolescents. Genetic Diseases. Psychological Adaptation. Genetic Counseling.

RESUMO

A alfabetização genética constitui um componente essencial na medicina contemporânea, particularmente no manejo de adolescentes com transtornos genéticos. Esse grupo

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populacional enfrenta desafios complexos que abrangem dimensões biomédicas, psicológicas e sociais. O objetivo deste capítulo é analisar o papel da alfabetização genética em adolescentes com doenças genéticas, sua relação com a adaptação psicológica e suas implicações clínicas. São revisados os componentes da alfabetização genética, fatores associados, evidências empíricas e o papel do aconselhamento genético. As evidências sugerem que, embora os níveis de alfabetização genética sejam frequentemente baixos, os adolescentes podem apresentar níveis adequados de adaptação psicológica, o que indica a influência de fatores psicossociais. Propõe-se um modelo integrador biopsicossocial que permita otimizar intervenções clínicas e educacionais.

Palavras-chave: Alfabetização Genética. Adolescentes. Doenças Genéticas. Adaptação Psicológica. Aconselhamento Genético.

RESUMEN

La alfabetización genética constituye un componente esencial en la medicina contemporánea, particularmente en el manejo de adolescentes con trastornos genéticos. Este grupo poblacional enfrenta desafíos complejos que abarcan dimensiones biomédicas, psicológicas y sociales. El objetivo de este capítulo es analizar el papel de la alfabetización genética en adolescentes con enfermedades genéticas, su relación con la adaptación psicológica y sus implicaciones clínicas. Se revisan los componentes de la alfabetización genética, factores asociados, evidencia empírica y el rol del asesoramiento genético. La evidencia sugiere que, aunque los niveles de alfabetización genética suelen ser bajos, los adolescentes pueden presentar adecuados niveles de adaptación psicológica, lo que indica la influencia de factores psicosociales. Se propone un modelo integrador biopsicosocial que permita optimizar intervenciones clínicas y educativas.

Palabras clave: Alfabetización Genética. Adolescentes. Enfermedades Genéticas. Adaptación Psicológica. Asesoramiento Genético.

1 INTRODUCTION

The advancement of medical genetics has profoundly transformed the understanding, diagnosis, and management of multiple diseases. However, this progress has generated a growing need for patients to understand complex genetic concepts in order to actively participate in decision-making about their health (Abrams et al., 2015; Hurle et al., 2013).

In this context, genetic literacy emerges as a fundamental tool, especially in vulnerable populations such as adolescents with genetic disorders. Adolescence represents a critical stage of development characterized by the construction of identity, the search for autonomy, and the consolidation of cognitive and social skills (Alderson, 2007; McGill et al., 2019).. The coexistence of these characteristics with a chronic genetic condition introduces additional challenges that impact both psychological well-being and adherence to treatment (Wakefield et al., 2016).

Various studies have indicated that adolescents with genetic diseases constitute a risk group for emotional alterations, including anxiety, depression and difficulties in social integration. However, a remarkable resilience has also been observed in many of these patients (Forrest et al., 2010).

This chapter aims to comprehensively analyze genetic literacy in adolescents with genetic disorders, exploring its relationship with psychological adaptation and its relevance in clinical practice.

2 CONCEPTS OF GENETIC LITERACY

Genetic literacy is defined as an individual's ability to obtain, understand, and use genetic information to make informed decisions about their health (Hurle et al., 2013; Lea et al., 2011).

This concept is structured in four main dimensions (Abrams et al., 2015) :(FIG. 1)

2.1 FAMILIARITY

Degree of exposure and comfort with concepts such as genes, chromosomes, mutations, and inheritance.

2.2 COMPREHENSION

Ability to interpret genetic information and establish relationships between concepts.

2.3 GENETIC ARITHMETIC

Ability to understand probabilities, risks, and inheritance patterns (Kaphingst et al., 2012).

Genetic literacy is a subcomponent of health literacy, but it is more complex due to the abstract and probabilistic nature of genetic information (Nutbeam, 2000).

3 ADOLESCENCE AND GENETIC DISORDERS

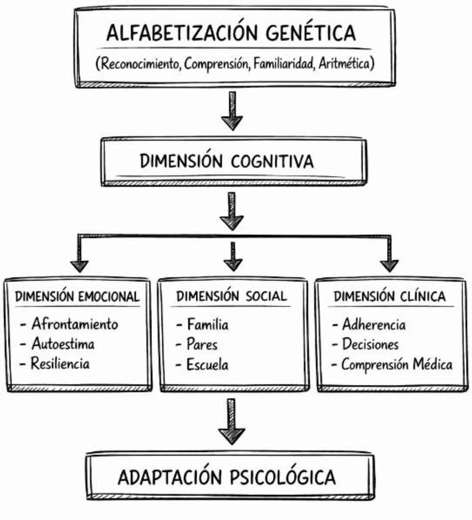
Adolescence is a transitional stage characterized by biological, psychological, and social changes. In this period, individuals develop their identity, autonomy, and decision-making skills (Alderson, 2007).

When a teen has a genetic disorder, multiple stressors are added:

- Chronicity of the disease
- Frequent medical checkups
- Functional limitations
- Social stigmatization
- Uncertainty about the forecast (Biesecker & Peters, 2001)

Figure 1

Conceptual model. Biopsychosocial model of genetic literacy in adolescents



Modelo integrador que ilustra la interacción entre alfabetización genética y factores psicosociales en la adaptación de adolescentes con trastornos genéticos.

Ability to identify basic genetic terms used in clinical practice.

3.1 PSYCHOLOGICAL IMPACT

Teens with genetic diseases may experience:

- Anxiety and depression
- Low self-esteem
- Alterations in body image
- Difficulties in social relationships
- Fear related to future reproduction

These manifestations have been widely documented in the literature, highlighting the emotional impact of genetic knowledge and the associated uncertainty (Wakefield et al., 2016; McGill et al., 2019).

3.2 SOCIAL DIMENSION

Social impact includes:

- Peer integration issues
- Long-term family dependency
- Limitations on school and recreational activities

The family and social environment plays a determining role in the adolescent's adaptation, acting as a protective or risk factor depending on the quality of the support received (Forrest et al., 2010; Bronfenbrenner, 1989).

Despite these challenges, many adolescents maintain similar life aspirations to their peers, highlighting the importance of interventions that promote their autonomy. (FIG.2)

4 LEVELS OF GENETIC LITERACY IN ADOLESCENTS

The available evidence indicates that adolescents with genetic disorders generally have low levels of genetic literacy (Haga et al., 2013; Kaphingst et al., 2012).

Among the most relevant findings:

- Low recognition of specialized terms
- Partial understanding of complex concepts
- Difficulties in interpreting probabilities
- Better performance on everyday concepts such as "heredity"

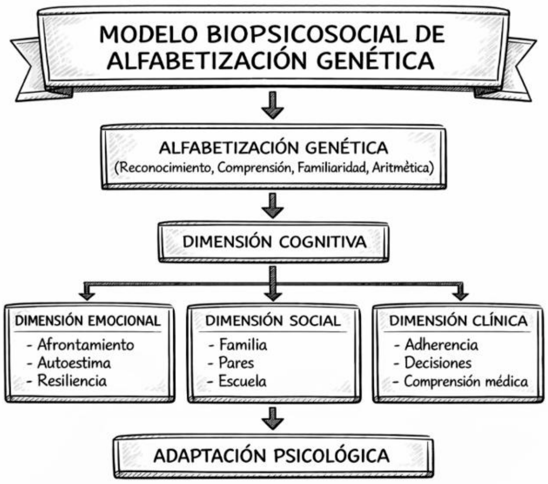
These results suggest that genetic knowledge in adolescents is:

- Fragmented
- Superficial

- Dependent on educational level (Lea et al., 2011)

Figure 2

The figure represents a biopsychosocial model of genetic literacy, where genetic knowledge acts as the starting point of the process. This is channeled through the cognitive dimension, which in turn influences three key areas: emotional (coping, self-esteem, resilience), social (family, peers, school) and clinical (adherence, decision-making, medical understanding). The interaction of these dimensions converges in psychological adaptation, showing that genetic understanding is not only informative, but also decisive in the integral adjustment of the patient



5 PSYCHOLOGICAL ADAPTATION

Psychological adaptation is defined as the degree to which an individual manages to adjust emotionally and cognitively to a chronic condition (Wakefield et al., 2016).

This construct includes:

- Effective coping
- Self-esteem
- Social integration
- Existential well-being

5.1 KEY FINDINGS

Studies have shown that:

- Adolescents may have adequate levels of psychological adjustment
- There is a significant capacity for resilience
- Social support plays a determining role (Forrest et al., 2010; Wakefield et al., 2016)

5.2 RELATIONSHIP TO GENETIC LITERACY

A particularly relevant finding is that:

There is not always a direct relationship between genetic literacy and psychological adaptation (McGill et al., 2019).

This suggests that:

- Knowledge is not the only determinant of well-being
- Emotional and social factors have a significant weight (Bronfenbrenner, 1989)

6 FACTORS ASSOCIATED WITH GENETIC LITERACY

Genetic literacy is influenced by multiple variables (Abrams et al., 2015).

6.1 EDUCATIONAL LEVEL

There is a direct relationship between schooling and genetic understanding (Lea et al., 2011).

6.2 FAMILY ENVIRONMENT

The educational level of the parents significantly influences the adolescent's understanding (Forrest et al., 2010).

6.3 ACCESS TO INFORMATION

The quality of doctor-patient communication is key (Kaphingst et al., 2016).

6.4 COMPLEXITY OF DIAGNOSIS

More complex diseases generate greater difficulty in understanding (Haga et al., 2013).

7 ROLE OF GENETIC COUNSELING

Genetic counseling is a fundamental process in the management of patients with genetic diseases (Resta et al., 2006).

7.1 DEFINITION

It is an educational and psychosocial process that:

- Explain the nature of the disease
- Facilitates decision-making
- Promotes psychological adaptation (Biesecker, 2001)

7.2 MAIN FUNCTIONS

- Translating complex information into understandable language
- Assess patient understanding
- Reduce anxiety and uncertainty
- Support reproductive decisions (Resta et al., 2006)

7.3 IN ADOLESCENTS

In this population, counseling should:

- Adapt to the cognitive level
- Include emotional support
- Promoting progressive autonomy (Alderson, 2007)

8 CLINICAL IMPLICATIONS

Genetic literacy has important applications in medical practice (Lea et al., 2011):

8.1 CLINICAL COMMUNICATION

- Use of clear language
- Understanding Check
- Visual and didactic support (Kaphingst et al., 2016)

8.2 THERAPEUTIC ADHERENCE

- Greater understanding → better adhesion
- Reduction of errors in disease management (Haga et al., 2013).

8.3 PUBLIC HEALTH

- Incorporation of genetics in school education
- Health literacy programs (Nutbeam, 2000)

9 BIOPSYCHOSOCIAL INTEGRATIVE MODEL

A conceptual model based on three dimensions is proposed (Bronfenbrenner, 1989):

9.1 COGNITIVE DIMENSION

- Genetic Literacy

9.2 EMOTIONAL DIMENSION

- Psychological adaptation
- Coping Strategies

9.3 Social dimension

- Family Support
- Peer network
- Educational context

Integrative hypothesis

Psychological adjustment in adolescents with genetic disorders depends more on psychosocial factors than on the level of genetic literacy alone (Wakefield et al., 2016).

10 LIMITATIONS IN CURRENT RESEARCH

- Scarcity of studies in the adolescent population (McGill et al., 2019)
- Predominance of cross-sectional designs
- Lack of specific tools for adolescents
- Limited evidence in Latin American contexts (Penchaszadeh, 2000)

11 FUTURE LINES OF RESEARCH

- Longitudinal studies
- Development of educational interventions
- Assessing the Impact of Genetic Counseling
- Integration of digital tools (O'Connor et al., 2016)

12 CONCLUSIONS

Genetic literacy is an essential component in the comprehensive management of adolescents with genetic disorders (Hurle et al., 2013). Although levels of knowledge are usually limited, adolescents may present an adequate psychological adaptation, which shows the importance of psychosocial factors (Wakefield et al., 2016).

The approach to this population must be comprehensive, incorporating educational strategies, emotional support and adequate genetic counseling. The implementation of biopsychosocial models will optimize clinical results and improve the quality of life of these patients.

13 GENETIC LITERACY IN ADOLESCENT GYNECOLOGY

Adolescent gynecology represents a particularly relevant clinical setting for genetic literacy, given that many decisions at this stage are directly related to reproductive health (Committee on Adolescent Health Care, 2017).

- Pubertal development
- Ovarian function
- Future fertility
- Risk of genetic transmission
- Sexual and reproductive health

In adolescents with genetic disorders, these dimensions become more complex, since genetic information directly influences intimate, identity, and reproductive decisions.

13.1 GENETIC DISORDERS OF GYNECOLOGICAL RELEVANCE

The most relevant conditions include:

- Disorders of Sexual Differentiation (DSD)
- Primary ovarian insufficiency (e.g. Turner syndrome)
- Sex chromosomal abnormalities (e.g., Klinefelter syndrome in mosaic variants)
- Hereditary diseases with reproductive impact (cystic fibrosis, dystrophies, etc.)
- Carriers of X-linked diseases

These conditions require genetic understanding to address fertility and hereditary risk (McCullough & Chervenak, 2008). As well as:

- Reproductive Options
- Medical interventions (e.g., fertility preservation)

14 GENETIC LITERACY AND REPRODUCTIVE HEALTH

One of the most critical aspects is the adolescent's ability to understand reproductive implications (Chen & Goodson, 2007).

14.1 EARLY REPRODUCTIVE DECISION-MAKING

Although reproduction may seem distant, in adolescents with genetic diseases, questions arise such as:

- Can I have children?
- Will my disease be transmitted?
- What options are there to prevent transmission?

Here, genetic literacy has a direct impact on:

- Understanding genetic risk
- Perception of control
- Anticipatory anxiety (Wakefield et al., 2016)

14.2 REPRODUCTIVE TECHNOLOGIES AND GENETIC UNDERSTANDING

Genetic knowledge is key to understanding options such as:

- Preimplantation genetic diagnosis
- Prenatal diagnosis
- Reproductive counseling (Lyerly et al., 2009)

Without adequate literacy, these options can:

- Being misunderstood
- Generating false expectations
- Increase anxiety

15 ETHICAL DIMENSION IN ADOLESCENTS

Genetic literacy in gynecology is deeply linked to ethical dilemmas (Ross et al., 2013).

15.1 AUTONOMY VS. GUARDIANSHIP

In adolescents there is a tension between:

- Right to information
- Decision-making capacity
- Role of parents/guardians

The level of genetic literacy conditions the real ability to exercise autonomy (Alderson, 2007).

15.2 RIGHT NOT TO KNOW

Some teens may prefer:

- Avoid knowing genetic risks
- Postponing decisions

This poses challenges in:

- Late-onset diseases (Biesecker, 2001).
- Serious Hereditary Conditions

16 CLINICAL COMMUNICATION IN GENETIC GYNECOLOGY

Communication in this context must be highly specialized in adolescents (Kaphingst et al., 2016).

16.1 COMMON BARRIERS

- Excessive technical language
- Information overload
- Failure to adapt to cognitive development (Lea et al., 2011)
- Biomedical approach without an emotional component

16.2 ADVANCED STRATEGIES

- Use of analogies (e.g., "genetic instruction manual")
- Visual Models (Simplified Pedigree)
- Teach-back technique
- Tiered communication (Kaphingst et al., 2016)

17 DIGITAL GENETIC LITERACY

Adolescents actively seek information in digital environments (Stellefson et al., 2011).

17.1 RISKS

- Disinformation
- Misinterpretation
- Internet-induced anxiety (O'Connor et al., 2016)

17.2 OPPORTUNITIES

- Educational apps
- Interactive Platforms
- Genetic inheritance simulators (Stellefson et al., 2013)

This opens a key line for innovative interventions.

18 LATIN AMERICAN PERSPECTIVE

This is a key point to increase editorial impact, since genetic literacy in Latin America presents structural and sociocultural particularities that influence its development and implementation (Penchaszadeh, 2000).

18.1 STRUCTURAL GAPS

In Latin America:

- Low access to genetic counseling
- Limited training in clinical genetics
- Inequality in health education

These limitations have been widely documented and reflect an unequal distribution of genetic resources and services in the region (Castillo-Fernandez et al., 2019; Paz-y-Miño et al., 2015).

18.2 SOCIOCULTURAL CONTEXT

Cultural factors influence:

- Perception of genetic diseases
- Reproductive decisions
- Stigma

Cultural beliefs can condition the interpretation of genetic disease, generating barriers in the acceptance of the diagnosis and in the search for care (Biesecker & Peters, 2001; Penhaszadeh, 2001).

18.3 RELEVANCE TO ECUADOR

In contexts such as Ecuador:

- Access to genetics is limited
- Health education is heterogeneous
- The family role is highly decisive

These characteristics reflect the structural and sociocultural inequalities that directly impact genetic literacy (Paz-y-Miño et al., 2015).

19 EXTENDED BIOPSYCHOSOCIAL MODEL WITH A GYNECOLOGICAL APPROACH

An expansion of the integrative model based on an ecological approach is proposed (Bronfenbrenner, 1989).

An expansion of your model is proposed:

19.1 COGNITIVE DIMENSION

- Genetic Literacy
- Reproductive understanding (Hurle et al., 2013)

19.2 EMOTIONAL DIMENSION

- Reproductive anxiety
- Body and sexual identity (Wakefield et al., 2016)

19.3 SOCIAL DIMENSION

- Family Support
- Cultural norms
- Access to health (Forrest et al., 2010)

19.4 CLINICAL DIMENSION

- Medical Interventions
- Gynecological follow-up
- Genetic counseling (Resta et al., 2006)

Extended hypothesis

Reproductive decision-making in adolescents with genetic disorders depends on the interaction between genetic literacy, emotional maturity, and sociocultural context. (McGill et al., 2019).

20 PRACTICAL IMPLICATIONS IN GYNECOLOGY

20.1 CLINICAL CONSULTATION

- Integrating genetics into gynecological history
- Assess patient understanding
- Include anticipatory counseling (Committee on Adolescent Health Care, 2017)

20.2 PATIENT EDUCATION

- Material adapted by age
- Using Visual Tools
- Progressive education (Kaphingst et al., 2016)

20.3 HEALTH POLICIES

- Incorporating Genetics into Adolescent Health Programs
- Strengthen genetic counseling
- Continuing Medical Education (World Health Organization, 2002)

21 CLINICAL INTERVENTION PROPOSAL

A structured model based on educational and psychosocial interventions is proposed (Lea et al., 2011).

4-phase model:

1. **Evaluation**
   - Level of genetic literacy
   - Emotional state (Wakefield et al., 2016)
2. **Personalized education**
   - Adapted language
   - Visual material (Kaphingst et al., 2016)
3. **Psychosocial support**
   - Clinical Psychology
   - Family work (Forrest et al., 2010)
4. **Longitudinal Tracking**
   - Periodic re-evaluation
   - Information adjustment (O'Connor et al., 2016)

22 EXTENDED CONCLUSION

Genetic literacy in adolescents with genetic disorders, particularly in the gynecological setting, is a fundamental axis in modern medicine (Hurle et al., 2013). Its impact transcends biomedical knowledge, influencing identity, mental health, and reproductive decisions (Wakefield et al., 2016).

The clinical approach must evolve towards integrative models that consider not only the transmission of information, but also the emotional, social and cultural context of the adolescent (Bronfenbrenner, 1989).

Genetic Literacy in Latin American Adolescents

1. Introduction

Genetic literacy in Latin American adolescents represents an emerging challenge in public health and clinical medicine. Despite the rapid advancement of medical genetics, its integration into clinical practice and health education remains limited in Latin America, generating a gap between scientific knowledge and its application in the general population (Abrams et al., 2015; Hurle et al., 2013).

Adolescents are a particularly vulnerable group, given that they are in a stage of cognitive and psychosocial transition, which influences their ability to understand complex genetic information and participate in decision-making related to their health (McGill et al., 2019).

2. Structural context in Latin America

2.1 Access to Genetics Services

In Latin America, access to clinical genetics services is limited and heterogeneous. The distribution of geneticists is uneven and services are often concentrated in highly complex urban centers (Penchaszadeh, 2000; Castillo-Fernández et al., 2019).

In countries such as Ecuador, this situation is aggravated by:

- Limitations in the health system
- Lack of integration of genetic counseling in primary care
- Economic and geographical barriers

This conditions late or non-existent access to adequate genetic information (Paz-y-Miño et al., 2015)

2.2 Genetics and health education

Formal education in genetics in Latin America has important limitations. Educational programs tend to focus on theoretical concepts, with little clinical application and little integration into health literacy (Dougherty et al., 2011).

This translates into:

- Superficial understanding of genetic concepts
- Difficulties in interpreting hereditary risks
- Limitations in informed decision-making (Lea et al., 2011).

3. Sociocultural factors

3.1 Family influence

In Latin American contexts, the family plays a central role in health decision-making. Adolescents rely heavily on their parents for the interpretation and management of genetic information (Forrest et al., 2010).

This can generate:

- Information Filtering
- Limitation of adolescent autonomy
- Biased interpretations of medical information

3.2 Beliefs and stigmatization

Several studies have shown that cultural beliefs influence the perception of genetic diseases, associating them in some cases with destiny, punishment or social stigma (Biesecker & Peters, 2001).

These beliefs can lead to:

- Denial of diagnosis
- Delay in seeking care
- Social isolation

4. Genetic literacy and the digital environment

Latin American adolescents have high access to the internet, but do not necessarily possess critical digital literacy skills (Stellefson et al., 2011).

This involves risks such as:

- Disinformation
- Misinterpretation of genetic information
- Self-Diagnosis Related Anxiety

However, the digital environment also represents an opportunity to implement innovative educational interventions (O'Connor et al., 2016).

5. Evidence in Latin American adolescents

The evidence in the region is limited, but it consists of noting:

- Low levels of genetic literacy
- Fragmented understanding of concepts
- Dependence on health professionals for decision-making (Haga et al., 2013; Kaphingst et al., 2012)

Despite this, it has been observed that:

- Many adolescents have adequate psychological adaptation
- Family support is a key protective factor (Wakefield et al., 2016).

6. Implications in adolescent gynecology

Genetic literacy acquires special relevance in adolescent gynecology, where it directly influences reproductive health and future decision-making. (FIG. 3)

6.1 Reproductive health

Understanding genetic concepts is essential to:

- Assess risk of hereditary transmission
- Planning for motherhood
- Understand conditions that affect fertility (Committee on Adolescent Health Care, 2017).

For example, adolescents with Turner syndrome require specific genetic education to understand their reproductive prognosis.

6.2 Teenage pregnancy

In adolescent pregnancy contexts, genetic literacy is often limited, restricting access to:

- Prenatal diagnosis
- Genetic counseling
- Informed decisions (Chen & Goodson, 2007).

6.3 Role of the gynecologist

In Latin America, the gynecologist frequently assumes genetic counseling functions due to the shortage of specialists, which highlights the need for training in genetic communication (Resta et al., 2006).

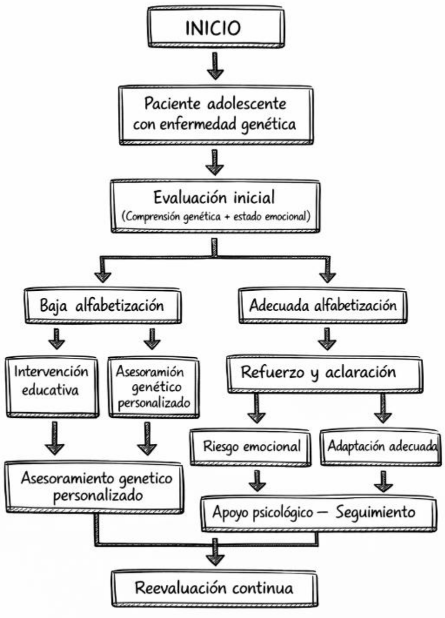
7. Model adapted to Latin America

An integrative model is proposed that includes:

- Individual level: genetic literacy and cognitive development
- Family level: parental education and communication dynamics
- Health level: access to services and quality of care
- Sociocultural level: beliefs, stigma and social norms (Bronfenbrenner, 1989).

Figure 3

Clinical flowchart. Approach to genetic literacy in adolescents



Flujograma de intervención clinica para mejorar la alfabetización genética y la adaptación psicológica en adolelescentes.

8. Intervention strategies

8.1 Clinical level

- Use of clear language
- Understanding Check

- Progressive education (Kaphingst et al., 2016)

8.2 Educational level

- Integrating Genetics into School Curricula
- Health literacy programs (Nutbeam, 2000)

8.3 Digital level

- Validated educational platforms
- Technology-based interventions (Stellefson et al., 2013)

8.4 Level of public policies

- Strengthening Genetics Services
- Inclusion of genetic counseling in primary care (World Health Organization, 2002)

9. Conclusions

Genetic literacy in Latin American adolescents is limited and influenced by structural, educational, and sociocultural factors. However, it represents a key opportunity to improve the quality of health care, particularly in the gynecological setting (Hurle et al., 2013).

The development of strategies adapted to the regional context will strengthen the autonomy of the adolescent, optimize decision-making and reduce health inequities.

Ethical Issues in Adolescent Genetic Literacy

1. Introduction

Genetic literacy in adolescents raises important ethical implications related to autonomy, confidentiality, informed decision-making, and the psychological impact of genetic knowledge (McBride et al., 2010; Bunnik et al., 2013). These considerations are especially relevant in this population, given that adolescents are in a process of cognitive and emotional development that conditions their ability to understand complex information and make autonomous decisions (Ross et al., 2013).

2. Autonomy and decision-making capacity

2.1 Progressive autonomy

The principle of autonomy in adolescents must be understood as a dynamic and progressive process. As the individual matures, he or she acquires a greater capacity to understand genetic information and participate in decisions related to his or her health (Alderson, 2007).

However, in Latin American clinical practice, decision-making is usually mediated by parents or guardians, which can limit the active participation of the adolescent in his or her own care process.

2.2 Informed consent

Informed consent in genetics presents particular complexities due to the probabilistic nature of the information and its family implications. In adolescents, it is recommended to obtain the patient's assent, accompanied by the consent of the parents or guardians, adapting the information to the child's level of cognitive development (McCullough & Chervenak, 2008).

3. Right to know and right not to know

One of the most relevant ethical dilemmas in genetics is the balance between the right to know and the right not to know genetic information. This conflict is particularly important in late-onset diseases or with no available treatment (Biesecker, 2001).

In adolescents, the disclosure of genetic information can generate anxiety, affect the construction of identity and modify the perception of the future, so it is recommended to individualize the communication of these results.

4. Genetic confidentiality and privacy

Genetic information is unique because of its predictive and familial nature. This poses ethical challenges when a patient's outcomes have implications for other family members (Offit et al., 2004).

The health professional must balance patient confidentiality with the potential benefit of informing at-risk family members, always maintaining an approach focused on respect for individual rights.

5. Psychological impact and the principle of non-maleficence

Access to genetic information can have significant psychological effects, including anxiety, guilt, or stigmatization. In adolescents, these effects may be more intense due to their stage of development (Wakefield et al., 2016).

The principle of non-maleficence obliges professionals to minimize these risks, through empathetic communication, psychological support and continuous evaluation of the emotional impact.

6. Justice and equity in access

From the principle of justice, it is essential to guarantee equitable access to genetic services. In Latin America, there are significant inequalities related to socioeconomic, geographical, and educational factors (Penchaszadeh, 2001).

In contexts such as Ecuador, these inequities limit access to genetic counseling and affect adolescents' ability to understand and use genetic information in decision-making.

7. Ethical implications in adolescent gynecology

7.1 Reproductive decisions

Genetics in gynecology introduces additional ethical dilemmas related to fertility and reproduction. Adolescents with conditions such as Turner syndrome face complex decisions about their reproductive capacity, which requires an ethical approach that combines clear information, emotional support, and respect for autonomy (McCullough & Chervenak, 2008).

7.2 Prenatal diagnosis

Prenatal diagnosis raises ethical questions related to termination of pregnancy, disability, and quality of life. These decisions are strongly influenced by cultural, social, and legal factors, especially in Latin America (Lyerly et al., 2009).

8. Role of the health professional

The health professional plays a fundamental role as an ethical mediator in genetic communication. Their responsibilities include providing clear information, assessing patient understanding, promoting informed decisions, and respecting cultural and personal values (Resta et al., 2006).

9. Applied ethical framework

An approach based on the classical principles of bioethics is proposed:

- **Autonomy:** promoting the active participation of the adolescent
- **Charity:** Maximizing the Benefits of Genetic Knowledge
- **Nonmaleficence:** minimizing psychological damage
- **Justice:** Ensuring equitable access to services

This framework allows clinical practice to be oriented towards a more comprehensive and patient-centred model.

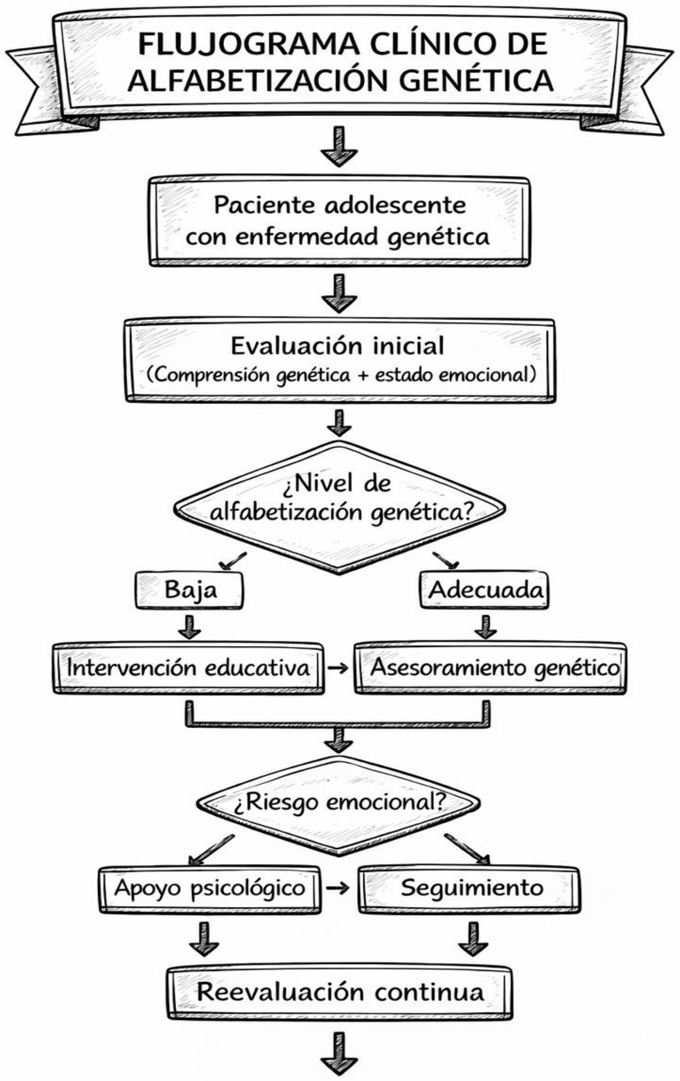
10. Conclusions

Ethical issues in adolescent genetic literacy are critical to appropriate clinical care. The complexity of genetic information, together with the characteristics of adolescent development and Latin American sociocultural particularities, requires a comprehensive ethical approach.

The integration of bioethical principles in clinical practice will improve the quality of care, promote patient autonomy and reduce health inequities. (FIG 4)

Figure 4

*The flowchart shows a **structured clinical process** for genetic literacy in adolescents with genetic diseases. It begins with the evaluation of the patient (genetic understanding and emotional state), followed by the classification of the level of literacy, which guides educational or reinforcement interventions. Subsequently, genetic counseling and psychological evaluation are integrated, allowing emotional risk to be identified. According to this, psychological support or follow-up is established, culminating in a **continuous reevaluation** as the axis of comprehensive care*



REFERENCES

Abrams, L. R., McBride, C. M., Hooker, G. W., Cappella, J. N., & Koehly, L. M. (2015). The many facets of genetic literacy.

Alderson, P. (2007). Competent children? Minors' consent to health care treatment and research. Social Science & Medicine, 65(11), 2272–2283. https://doi.org/10.1016/j.socscimed.2007.07.007

McAllister, M. (2003). Genetic counseling outcomes. Journal of Genetic Counseling, 12(3), 171–184.

McAllister, M., et al. (2011). Psychological adaptation scale. Clinical Genetics, 79(3), 303–311.

McAllister, M., & Payne, K. (2011). Adaptation framework. Patient Education and Counseling, 82(2), 204–212.

McBride, C. M., et al. (2010). Genetic risk communication. Annual Review of Genomics and Human Genetics, 11, 343–369.

McGill, B. C., Wakefield, C. E., Vetsch, J., et al. (2019). Understanding inherited conditions. European Journal of Human Genetics, 27(3), 445–454. https://doi.org/10.1038/s41431-018-0294-2

Nutbeam, D. (2000). Health literacy as a public health goal. Health Promotion International, 15(3), 259–267. https://doi.org/10.1093/heapro/15.3.259

Nutbeam, D. (2008). Health literacy concept. Social Science & Medicine, 67(12), 2072–2078.

O'Connor, S., Hanlon, P., O'Donnell, C. A., et al. (2016). Digital health adoption. BMC Medical Informatics and Decision Making, 16, Article 120. https://doi.org/10.1186/s12911-016-0351-0

Paasche-Orlow, M. K., et al. (2007). Literacy pathways. American Journal of Health Behavior, 31(Suppl 1), S19–S26.

Paz-y-Miño, C., et al. (2015). Genetic services in Ecuador. Journal of Community Genetics, 6(3), 223–230. https://doi.org/10.1007/s12687-014-0210-6

Penchaszadeh, V. B. (2000). Genetic services in Latin America. Revista Panamericana de Salud Pública, 8(1–2), 84–92. https://doi.org/10.1590/S1020-49892000000700013

Pleasant, A., et al. (2016). Global health literacy. International Journal of Environmental Research and Public Health, 13(12), Article 1230.

Resta, R., Biesecker, B. B., Bennett, R. L., et al. (2006). Genetic counseling definition. Journal of Genetic Counseling, 15(2), 77–83. https://doi.org/10.1007/s10897-005-9014-3

Rolland, J. S. (2005). Chronic illness and family systems. Families, Systems, & Health, 23(4), 431–445.

Sanderson, S. C., et al. (2012). Genetic knowledge gaps. Genetics in Medicine, 14(2), 223–230.

Sentell, T. (2012). Literacy disparities. Journal of Health Communication, 17(Suppl 3), 82–99.

Sørensen, K., et al. (2012). Health literacy model. BMC Public Health, 12, Article 80.

Stellefson, M., Hanik, B., Chaney, B., et al. (2011). eHealth literacy. Journal of Medical Internet Research, 13(4), Article e102. https://doi.org/10.2196/jmir.1673

Stellefson, M., et al. (2013). Mobile health interventions. Preventing Chronic Disease, 10, Article E94. https://doi.org/10.5888/pcd10.120310

Tercyak, K. P., et al. (2001). Genetic counseling adolescents. Journal of Clinical Oncology, 19(1), 34–40.

von Wagner, C., et al. (2009). Health literacy model. Social Science & Medicine, 69(10), 1530–1537.

Wakefield, C. E., Hanlon, L. V., Tucker, K. M., et al. (2016). Psychological impact of genetic information. European Journal of Human Genetics, 24(4), 495–499. https://doi.org/10.1038/ejhg.2015.130

Wiener, L., et al. (2015). Adolescent coping. Journal of Adolescent Health, 56(1), 6–14.

World Health Organization. (2002). Genomics and world health. WHO.