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EDITORIAL

Atención al riesgo genético preconcepcional desde la medicina preventiva personalizada

Attention to Preconception Genetic Risk from Personalized Preventive Medicine

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The preconception period constitutes a critical stage for the prevention of genetic diseases, birth defects, and neurodevelopmental disorders. In the current context of preventive medicine, the early identification of genetic risk before conception represents a strategic opportunity for early, effective, and ethical intervention. The progressive integration of community genetics, Primary Health Care (PHC), and the principles of personalized medicine has enabled the transformation of the traditional approach, throughout history, towards a proactive, predictive, and preventive model centered on the individual, the family, and the community.[1,2]

Preconception genetic risk is defined as the increased probability that a couple will conceive a child affected by a genetic disease or birth defect of monogenic, chromosomal, or multifactorial origin. This risk is conditioned by multiple factors, including personal and family history, population or ethnic background, reproductive age, consanguinity, the presence of chronic diseases, and exposure to environmental or teratogenic factors. Its assessment before pregnancy allows action at a time of maximum preventive effectiveness, when it is still possible to modify risks, optimize health conditions, and offer informed reproductive alternatives.[1,3]

Community genetics emerges as a field oriented towards applying genetic knowledge in populations, with the aim of improving collective health through prevention, promotion, and equity. Unlike traditional clinical genetics, which focuses on the affected individual, community genetics prioritizes the identification of risks in apparently healthy individuals and their management from the family and social environment. This approach is compatible with PHC, which is characterized by its accessibility, continuity of care, and deep understanding of the community context.[4]

From the PHC perspective, preconception genetic risk assessment can be systematically incorporated using simple, high-impact clinical tools, such as structured genetic history-taking and the creation of family trees spanning at least three generations. Identifying histories such as recurrent miscarriages, perinatal deaths, intellectual disability, birth defects, or known hereditary diseases allows for risk stratification and the definition of timely preventive measures, without initially requiring complex technologies.[4-6]

Personalized medicine strengthens this approach by enabling a more precise characterization of individual and familial genetic risk. Far from being limited to advanced genomics, personalized medicine integrates clinical, genetic, environmental, and social information to tailor preventive decisions to the specific characteristics of each individual or couple. In the preconception realm, this approach makes it possible to rationally select genetic screening tests, prioritize interventions based on the risk profile, and personalize preventive recommendations, all while respecting reproductive autonomy and ethical principles.[7,8]

The application of personalized medicine in PHC enables the identification of population

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subgroups with a higher probability of being carriers of relatively frequent autosomal recessive diseases, such as hemoglobinopathies, cystic fibrosis, or specific inborn errors of metabolism. The early detection of carriers offers the possibility of implementing preventive measures before conception, which include: genetic education, specific supplementation, control of maternal chronic diseases, and timely referral to clinical genetics services, when indicated.[2,3,9]

The timely diagnosis of genetic risk during the preconception stage presents substantial advantages over prenatal diagnosis. While the latter is performed when the pregnancy is already established and intervention options are limited and complex, preconception assessment allows for a truly preventive approach, with a greater margin for informed decision-making and risk modification. This difference underscores the importance of strengthening preconception care as an integral part of Primary Health Care (PHC).

The adoption of preventive measures constitutes one of the fundamental pillars of the preconception approach. These measures include the elimination or reduction of teratogenic exposures, the optimization of nutritional status, particularly the supplementation with 400 mg of folic acid starting three months prior to conception for both partners, adequate metabolic control of chronic diseases such as diabetes, epilepsy, collagenopathies, or thyroid disorders, and updating the immunization schedule, among others. Personalizing these interventions increases their efficacy by adapting them to the biological and social characteristics of each couple.

Various recent studies have demonstrated that preconception care programs that integrate genetic assessment and personalized medicine principles are associated with better reproductive outcomes, a reduced incidence of serious genetic diseases, and more efficient use of healthcare resources. These benefits reinforce the need to consolidate PHC as the ideal setting for the progressive and sustainable implementation of these preventive strategies, with the establishment of well-defined and personalized protocols.[1]

The full integration of genetics and personalized medicine into primary care faces significant challenges. Among these, the insufficient genetic training of PHC professionals and the need to improve the population's genetic literacy stand out.[10] Addressing these barriers requires health policies that recognize the strategic value of preconception diagnosis and promote the continuous training of healthcare personnel.

In countries with health systems based on PHC, such as Cuba, the integration of community genetics at the primary level represents an exceptional opportunity to strengthen prevention and equity in reproductive health. The accumulated experience in the maternal-child program, along with the territorial organization of services, provides a solid foundation for systematically incorporating preconception genetic risk assessment as part of the comprehensive care for couples of reproductive age.[11]

From an ethical and social perspective, preconception genetic counseling must be conceived as a dynamic process of risk communication, aimed at supporting free and informed decision-making. The goal is not to direct reproductive choices, but to offer truthful, understandable, and contextualized information, respecting cultural values and individual preferences. This approach is essential to ensure the acceptance and sustainability of preventive strategies.[6,8]

The integration of community genetics, primary health care, and personalized medicine in preconception genetic risk assessment constitutes a key strategy for advancing towards a truly preventive medicine. Identifying risks early, personalizing interventions, and acting from the first level of care protects the health of future generations and reaffirms the role of PHC as the central axis of health systems oriented towards prevention, equity, and quality of care.[1,2,8]

In this context, Mayabeque province has favorable conditions to begin consolidating, starting in 2026, a territorial model for addressing preconception genetic risk. Among the priority actions are: the systematic incorporation of preconception genetic risk assessment in family planning consultations, using standardized protocols with the participation of genetic counselors from each municipality; the strengthening of continuous training for family doctors and nurses in basic aspects of community genetics and personalized medicine; the timely referral of couples with elevated risk from the primary level to clinical genetics consultations; and the development of strategies for genetic education and literacy aimed at the population.

The progressive and evaluable implementation

of these actions will strengthen the primary prevention of genetic diseases and birth defects, optimize the use of healthcare resources, and contribute sustainably to the improvement of reproductive and primary health in the province.

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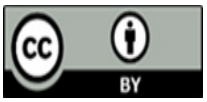
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