Genetic Inheritance Risk Calculation as a Practical Approach in Health Prevention and Management: A Perspective

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ABSTRACT

In the genomic era, understanding the risk of inheriting genetic diseases has become a crucial element in designing effective health prevention and management strategies. The initial step in calculating the risk of genetic inheritance involves gathering information about family health history. Through the analysis of genetic data and family health history, healthcare professionals can determine an individual's potential genetic risk for various diseases. Calculating the risk of genetic inheritance has several important benefits, including early prevention and management, family planning, genetic counseling, targeted therapy and care development, and informed medical decision making. By knowing an individual's genetic risk for a particular disease, they can take appropriate prevention measures or early management actions. These may include lifestyle changes, regular health check-ups, or even advanced genetic testing for early detection. In Family Planning, calculating genetic risk can assist individuals and couples in planning pregnancies and families more effectively. If a high risk is detected, couples can consider options such as genetic counseling, premarital screening, or selecting suitable reproductive methods. Overall, calculating the risk of inheriting genetic diseases can provide individuals and families with valuable information to take preventive measures, make informed medical decisions, and manage their health more effectively.

Keywords: genetic counseling, genetic risk, genomic, inheritance pattern, screening

Perspective

In the genomic era, understanding the risk of inheriting genetic diseases has become a crucial element in designing effective health prevention and management strategies. A practical approach to calculating genetic risk not only helps individuals understand the potential diseases they may experience but also provides a strong basis for appropriate preventive actions (Chen et al., 2016; Grant et al., 2013). Bayes' theorem is frequently utilized to assess genetic risk within intricate family trees and ascertain the likelihood of possessing or

lacking a mutation responsible for a disease subsequent to receiving a negative test outcome. This theorem hinges on two primary elements: the test's specificity and sensitivity, and the initial likelihood of the disease or condition (Weinshilboum & Wang, 2017). The former is contingent on the test's ability to precisely detect and identify mutations, whereas the latter is more complex owing to a multitude of influencing variables (Baptista, 2005).

The initial step in calculating the risk of inheriting genetic diseases is the collection of information about family health history. Through the analysis of genetic data and family health history, healthcare professionals can determine an individual's potential genetic risk for various diseases (Setiawan et al., 2023). This includes diseases such as cancer, diabetes, heart disease, blood disorders, and other genetic disorders that can be passed down from generation to generation. It is recommended to employ standardized symbols and terminology in pedigrees to guarantee accurate communication of data to recipients. Pedigrees aid in assessing patterns of disease transmission within families, covering Mendelian, chromosomal, mitochondrial, or multifactorial hereditary mechanisms. In cases of atypical inheritance patterns, consideration should be given to additional influential factors such as imprinting, uniparental disomy, DNA instability, gene-environment interactions, mosaicism, and synergistic heterozygosity. Understanding the mode of inheritance within familial contexts is beneficial for assessing disease susceptibility in relatives or offspring (Krautscheid & LaGrave, 2016).

Management of health also becomes an integral part of this approach. Patients with higher genetic risk may require closer health monitoring and more focused disease management. This involves collaboration between patients and healthcare professionals consisting of doctors, nurses, genetic counselors, midwives, geneticists, and other healthcare professionals (Setiawan et al., 2022). The primary goal of this interprofessional collaboration is to develop tailored care plans, including the use of appropriate therapies, regular monitoring of health conditions, and appropriate psychological support (Matziou et al., 2014).

The importance of a practical approach in calculating the risk of inheriting genetic diseases lies not only in identifying the risk but also in the actions taken in response to this information. By combining genetic knowledge with proven medical practices, individuals can take proactive steps to improve their health, reduce disease risk, and enhance their overall quality of life (Setiawan et al., 2024). A meta-analysis study has underscored the significant relationship between health knowledge and individual quality of life. Individual knowledge of genetics positively impacts lifestyle, healthy behaviors, and quality of life (Zheng et al., 2018).

Calculating the risk of inheriting genetic diseases has several important benefits, including early prevention and management, family planning, genetic counseling, targeted therapy and care development, and informed medical decision making (*see figure 1*). By knowing an individual's genetic risk for a particular disease, they can take appropriate prevention measures or early management actions. These may include lifestyle changes, regular health check-ups, or even advanced genetic testing for early detection (Yanes et al., 2019).
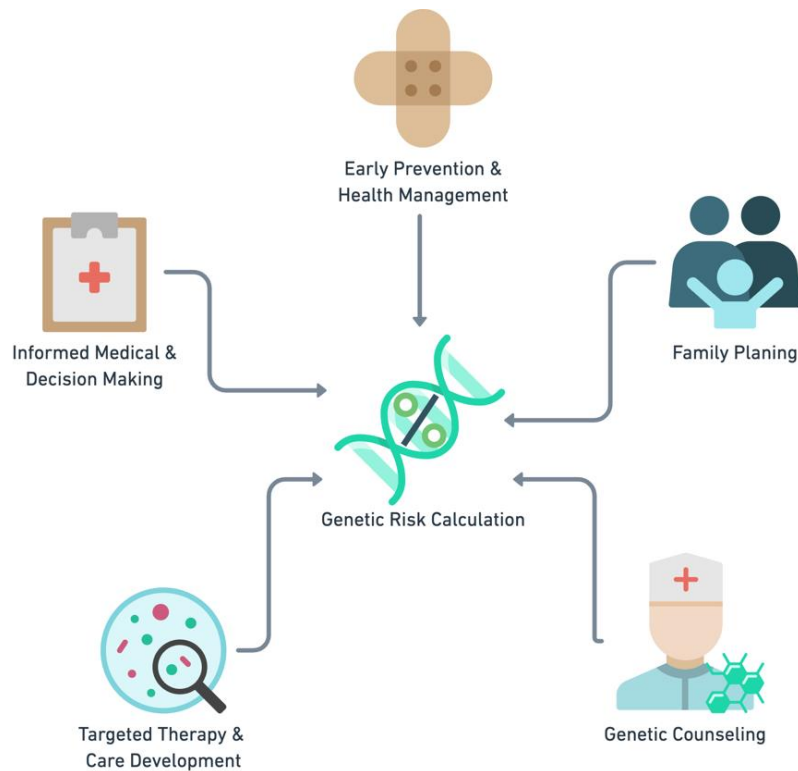
Figure 1. The Benefits of Genetic Risk Calculation
(made with whimsical website-based software: www.whimsical.com)

In Family Planning, calculating genetic risk can assist individuals and couples in planning pregnancies and families more effectively. If a high risk is detected, couples can consider options such as genetic counseling, Premarital screening, or selecting suitable reproductive methods. Premarital screening, introduced since the early 1970s, has shown significant benefits in preventing and controlling the prevalence of inherited genetic diseases such as thalassemia and sickle cell diseases (Alswaidi & O'Brien, 2009; Setiawan et al., 2023). Unfortunately, not all countries have policies mandating couples to undergo genetic testing before marriage due to ethical, religious, cultural, and human rights issues concerning post-diagnostic impact (Alswaidi & O'Brien, 2009; Zhong et al., 2021).

The results of genetic risk calculations can also serve as a basis for obtaining more in-depth genetic counseling. Genetic counselors can provide further information about the risks and offer emotional support as well as practical advice to help individuals and families manage this situation. Research results indicate that psychosocial issues such as anxiety increase due to test results, misconceptions about test results, and delays in receiving test results (Yanes et al., 2019). It is in these genetic counseling sessions where doctors, nurses, or genetic counselors can assist individuals in adapting to genetic issues, including emerging psychosocial responses (Setiawan et al., 2021).

A better understanding of genetic risk can aid in the development of more targeted therapies and personalized care. This means that patients with specific genetic risks can receive more effective treatment and minimize unnecessary side effects or risks.

Pharmacogenomics and precision medicine aim to use individual genetic information to design more targeted treatments (Aronson & Rehm, 2015). Pharmacogenomics studies how genetic variations affect responses to drugs, while precision medicine adapts individually tailored treatment approaches based on genetic information, health history, and other factors. By using genetic information from pharmacogenomics, healthcare practitioners can adjust drug dosages, select the most appropriate therapy, and avoid unwanted drug interactions, thereby enhancing the overall effectiveness and safety of treatment (Ji et al., 2016).

Lastly, by knowing their genetic risk, individuals can make more informed medical decisions. This may involve decisions related to additional genetic testing, preventive surgeries, or even participation in clinical research for the development of new therapies. Overall, calculating the risk of inheriting genetic diseases can provide individuals and families with valuable information to take preventive measures, make informed medical decisions, and manage their health more effectively.

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Conflict of interest
There is no conflict of interest.

Ethical approval
This study was approved by Health Research Ethics Committee, STIKes Muhammadiyah Ciamis in March 24, 2024 with the number 020/KEPK-STIKESMUCIS/III/2024.

Authors' contribution
Each author contributed equally in all the parts of the research. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

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