

Pediatric Genomic Medicine in Genetic Diseases of Children

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Abstract

Pediatric genomic medicine has revolutionized healthcare by providing insights into the genetic basis of diseases in children. In recent years, the use of genetic testing for ancestry information has become increasingly common in pediatric genomics. These tests can provide valuable insights into a child's genetic heritage and can be used to identify potential disease risks, inform treatment decisions, and facilitate better healthcare outcomes. However, the integration of genetic ancestry findings into pediatric genomic medicine also raises complex ethical, legal and social issues. In this article, we will explore the role of genetic ancestry findings in pediatric genomic medicine, its applications, benefits, and the associated ethical considerations. Genetic ancestry findings, often referred to as genetic ancestry testing or DNA ancestry testing, involve the analysis of an individual's DNA to determine their genetic heritage and ancestry.

Keywords: Pediatric genomic medicine • DNA ancestry testing • Genetic ancestry • Pharmacogenomic

Introduction

These tests examine specific markers or regions of a person's DNA to trace their ancestral origins to various populations or geographical regions. Ancestry tests analyze specific genetic markers, such as single nucleotide polymorphisms which can vary among different populations. By comparing these markers to reference databases, the tests estimate an individual's genetic ancestry. Genetic ancestry findings rely on large databases that contain genetic information from diverse populations worldwide. These databases serve as reference points for comparison. Ancestry tests often provide estimates of an individual's ancestral origins, specifying percentages of genetic heritage associated with different geographic regions or ethnic groups. Genetic ancestry findings can provide insights into a child's predisposition to certain genetic conditions that are more prevalent in specific populations. For example, sickle cell anemia is more common in individuals of African descent, while cystic fibrosis is more common in individuals of European descent. Ancestry information can influence treatment decisions. Some medications may be more or less effective or may have different side effect profiles based on an individual's genetic ancestry. Pharmacogenomic testing can help optimize medication selection and dosing. In some cases, genetic ancestry information can aid in diagnosing rare genetic diseases that are more prevalent in certain populations. This knowledge can guide clinicians in considering specific genetic tests when evaluating a child's symptoms. Genetic ancestry findings can be valuable for research purposes and can inform genetic counseling for families, especially those with diverse or mixed ancestry backgrounds [1].

Literature Review

By considering an individual's genetic ancestry, healthcare providers can

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assess disease risks more accurately, leading to more targeted preventive measures and early interventions. Understanding a child's genetic ancestry can help healthcare providers deliver culturally sensitive care and consider cultural factors that may influence healthcare decisions. Genetic counselors can use genetic ancestry findings to provide families with tailored information and guidance about their child's genetic heritage and potential health risks. Genetic ancestry testing often requires sharing genetic data with commercial testing companies, raising concerns about data privacy, security, and the potential misuse of genetic information. Obtaining informed consent from parents or guardians for genetic ancestry testing in children is essential. Parents may need to fully understand the implications of such testing, including the potential discovery of non-paternity or other unexpected genetic information. Genetic ancestry information can be emotionally charged, and healthcare providers must approach discussions about ancestry with cultural sensitivity and empathy, considering the potential impact on the child and family. There is a risk of genetic determinism, where individuals may interpret genetic ancestry findings as determinants of their identity or abilities. Healthcare providers must emphasize that ancestry is just one aspect of a person's genetic makeup. Ensuring equitable access to genetic ancestry testing is essential to prevent disparities in healthcare. Not all families may have the means or awareness to access such testing. In pediatric cases, the decision to undergo genetic ancestry testing should involve the informed consent of the child's parents or guardians. As the child grows older, they may also become involved in the decision-making process [2].

Discussion

Healthcare providers should weigh the potential benefits of genetic ancestry information against potential harms. It is crucial to ensure that the information obtained will have a meaningful impact on the child's healthcare. Healthcare providers should receive cultural competency training to communicate genetic ancestry findings effectively and sensitively. They must understand the potential emotional significance of such information for families. Careful consideration should be given to the selection of reputable testing companies and secure data storage to protect the privacy of genetic information. Continued advancements in genetic testing technologies will provide more accurate and detailed genetic ancestry information. Professional organizations may develop guidelines for the responsible use of genetic ancestry findings in pediatric care, helping to standardize practices and address ethical concerns. Healthcare providers and the public will benefit from increased education and awareness about the potential benefits and limitations of genetic ancestry testing. As genetic testing becomes more common, healthcare providers will need to

enhance their cultural competence to effectively communicate and interpret genetic ancestry information. Genetic ancestry findings have found their place in pediatric genomic medicine, offering valuable insights into a child's genetic heritage and potential health risks. By incorporating genetic ancestry information, healthcare providers can enhance disease risk assessment, optimize treatment decisions, and provide more personalized care. However, the integration of genetic ancestry findings also raises complex ethical, legal and social considerations, particularly regarding consent, privacy, and cultural sensitivity [3].

As this field continues to evolve, healthcare providers, researchers and policymakers must work collaboratively to navigate these challenges and ensure that genetic ancestry information is used responsibly and ethically in the best interest of pediatric patients and their families. In the rapidly advancing field of genomic medicine, understanding an individual's genetic ancestry has become a valuable component of comprehensive genetic analysis. Genetic ancestry findings not only provide insights into a person's heritage but also have important implications for pediatric genomic medicine. In this article, we will explore the role of genetic ancestry findings in pediatric genomics, its applications, ethical considerations, and potential benefits in improving healthcare outcomes for children and their families. Genetic ancestry information can inform clinical diagnosis and treatment decisions. Certain genetic variations are more prevalent in specific populations, which can influence disease risks and treatment responses. Understanding a child's genetic ancestry can help identify potential disease risks based on their ethnic or geographic background. This information can guide preventive measures and early screenings. Ancestry can impact drug metabolism, and certain populations may have unique pharmacogenomic profiles. This knowledge is crucial for determining optimal drug dosages and reducing the risk of adverse drug reactions in pediatric patients. Genetic ancestry findings contribute to our understanding of genetic diversity and population genetics, which can inform research studies and clinical trials in pediatric genomics [4].

In some cases, parents may want to know their child's genetic ancestry as part of informed consent for genetic testing. Understanding the child's genetic background can influence their decision-making process. Parents or guardians may make decisions about genetic testing and ancestry information on behalf of their children. Ensuring informed and voluntary consent while respecting a child's autonomy is crucial. Genetic ancestry information may reveal unexpected or sensitive familial or historical details. Healthcare providers and genetic counselors must handle this information with care and sensitivity. Genetic ancestry findings can have psychosocial implications for the child and their family. It may affect their self-identity and perception of ethnicity, potentially leading to complex emotional responses. Cultural and ethical aspects of discussing genetic ancestry should be considered. Healthcare providers should be culturally sensitive and provide support when discussing these findings. Protecting the privacy and security of genetic ancestry data is paramount. Ensuring that this information does not lead to discrimination or misuse is an ethical responsibility. Diagnosis and Disease Management: Genetic ancestry can influence the interpretation of genetic test results. For example, specific genetic variations may be benign in certain populations but pathogenic in others. Understanding ancestry can refine the diagnosis and guide treatment decisions. Carrier screening for genetic conditions often involves assessing the presence of specific genetic variations prevalent in certain populations. Genetic ancestry information can help identify which conditions are most relevant for screening in a given individual or family. Genetic ancestry may impact how a child metabolizes medications [5].

Incorporating genetic ancestry information into pediatric genomic medicine allows for more precise and personalized healthcare interventions. This can result in improved diagnosis and treatment outcomes. Tailoring drug treatments based on ancestry can enhance therapeutic effectiveness and reduce the risk of adverse drug reactions. In research studies and clinical trials involving pediatric genomics, understanding the genetic ancestry of participants can help ensure diverse representation and generalize findings to broader populations. Genetic ancestry findings can inform family planning decisions, particularly when parents belong to different ethnic or geographic backgrounds. It can help anticipate potential genetic risks in offspring. Many

individuals have complex ancestral backgrounds with contributions from multiple populations. Interpreting genetic ancestry can be challenging in such cases. Genetic reference databases may have limited representation of certain populations, leading to less accurate ancestry estimates for individuals from underrepresented backgrounds. Genetic ancestry findings may not align with an individual's cultural or self-identified ethnicity, leading to potential identity conflicts. Ancestry testing provides only a partial view of an individual's genetic heritage and cannot capture the richness of their cultural and family history. Medicalizing genetic ancestry by using it solely for clinical decision-making can oversimplify the complexities of identity, heritage, and culture. Advances in genomics and ancestry testing technologies are expected to enhance the accuracy and granularity of genetic ancestry estimates, especially for individuals with complex ancestries [6].

Conclusion

Genetic ancestry information may become a standard component of electronic health records allowing healthcare providers to access this information alongside medical history and genetic data. Genetic counselors and healthcare providers will play a critical role in educating families about the implications of genetic ancestry findings and providing support for any emotional or identity-related challenges. Healthcare providers will need to develop cultural competency and sensitivity when discussing genetic ancestry to ensure effective communication and support for diverse patient populations. Ongoing discussions and the development of ethical frameworks will help guide the responsible integration of genetic ancestry information into pediatric genomics. Genetic ancestry findings have found their place in pediatric genomic medicine as a valuable tool for clinical diagnosis, treatment, and disease risk assessment. However, their integration into healthcare raises complex ethical considerations, including issues related to privacy, consent, and psychosocial impact. It is essential to approach genetic ancestry information with sensitivity and cultural awareness while respecting individuals' autonomy and privacy. As technology and understanding of genetic ancestry continue to evolve, the responsible use of this information can contribute to improved healthcare outcomes for children and their families while acknowledging the richness of their identities and heritage.

Acknowledgement

None.

Conflict of Interest

None.

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