

Advantages and Disadvantages of Anthropogenetic Methods in Diagnosing Hertine Diseases

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Abstract: In this article, the causes of genetic diseases, factors of origin, diagnosis of the disease and their advantages and disadvantages in the use of anthropogenetic methods in this process are discussed.

Keywords: hereditary diseases, anthropogenetics, chromosome, mitochondria, mutation, fibrosis, hemophilia, diagnosis, offspring, benefit, deficiency, genetics, diagnosis.

Introduction. There are many types of hereditary diseases, some of which are:

1. Genetic diseases: This is a group of hereditary diseases that occur as a result of changes in the genes responsible for the proper functioning of the organism. Examples: cystic fibrosis, hereditary hemophilia, cerebral palsy.
2. Chromosomal abnormalities: These are changes in the number or structure of chromosomes that can lead to various hereditary diseases. Examples: Down syndrome, Klinefelter syndrome, Turner syndrome.
3. Mitochondrial diseases: This is a group of diseases that occur as a result of mutations in mitochondrial genes. Mitochondria are organelles responsible for energy production in cells. Examples: Lesch-Nyher disease, mitochondrial dystrophy, mitochondrial encephalopathy. [1]

Factors that influence the occurrence of hereditary diseases include:

1. Genetic predisposition: The presence of certain genes passed from parents to offspring can increase the risk of developing certain hereditary diseases.
2. Mutations: Changes in genes can occur by chance or due to environmental influences such as radiation or chemicals.
3. Adverse conditions during fetal development: The exposure of the developing fetus to external factors (such as infections, toxins, or poor nutrition) can lead to hereditary diseases.
4. Family history: The presence of hereditary diseases in the family can increase the likelihood of their passing on hereditary genetic mutations.
5. Consanguineous marriages: Marriages between close relatives (such as cousins) increase the likelihood of hereditary diseases, because relatives can share a number of genes.

It is important to note that hereditary diseases can have varying degrees of heritability and occur to different degrees in different individuals, depending on their genetic makeup and interaction with the environment.[2]

Methods. Anthropogenetics is the scientific study of heredity and the genetic makeup of humans. Anthropogenetics uses a variety of methods and approaches in the diagnosis of hereditary diseases. Some of these include:

1. Genetic testing: This is the primary diagnostic method that can detect the presence of genetic changes associated with hereditary diseases. This can also be a DNA test to detect specific mutations or changes in the genome that may be associated with the disease.
2. Cytogenetics: This method studies chromosomes and chromosome structures using a microscope. It can be used to detect numerical or structural abnormalities of chromosomes that may cause hereditary diseases. [3]
3. Molecular genetics: This is the study of specific genes and DNA to detect the presence of mutations or changes that may cause hereditary diseases. This can be DNA sequencing or other methods that look for specific changes in the genome.
4. Genetic mapping of the family tree: Anthropogenetics can use genetic mapping to study the inheritance of a particular disease across generations. Researchers analyze family trees and study the transmission of a disease from generation to generation.
5. Genomics: This is the study of the entire human genome, including all genes and DNA. Genomics allows us to identify important genes that may be associated with hereditary diseases and to study their function and effect on disease.

These techniques help to identify the presence of genetic changes associated with hereditary diseases and provide information that can be used to diagnose, predict, and treat these diseases. [4]

Results. Types of results of anthropogenetic methods in the diagnosis of hereditary diseases. Types of results of anthropogenetic methods in the diagnosis of hereditary diseases may include:

1. Determining the presence or absence of a specific genetic mutation or variant in the patient's genome. This can be useful in diagnosing hereditary diseases associated with specific genetic changes.
2. Assessing the risk of developing a hereditary disease based on the patient's genetic profile. This helps to predict the likelihood of developing a particular disease and take preventive measures or plan treatment.
3. Identifying heterozygous carriers of genetic mutations. This is important for assessing the risk of transmitting the disease from generation to generation.
4. Determining the patient's clinical and genetic phenotype. This allows you to clarify the diagnosis, predict the prognosis, and recommend the most effective treatment.
5. Identifying possible genetic factors that affect the patient's response to drugs. This allows for individualized therapy planning, taking into account the genetic characteristics of each patient.
6. Identifying family relationships between patients. This can be useful for identifying hereditary risk and for joint management of diseases in families.
7. Predicting the possible consequences of hereditary diseases for the health of the patient and his descendants.
8. Assessing the effectiveness of treatment based on monitoring changes in the patient's genetic profile.

These are some examples of the results of anthropogenetic diagnostics of hereditary diseases, and the specific results depend on the purpose and scope of the method.[5]

Discussions. Advantages and disadvantages of anthropogenetic methods in the diagnosis of hereditary diseases. Advantages of anthropogenetic methods in the diagnosis of genetic diseases:

1. Early detection of diseases: Anthropogenetic methods allow you to determine the presence of hereditary diseases even before the appearance of clinical symptoms. This allows you to start treatment or take preventive measures immediately after the birth of a child.
2. High diagnostic accuracy: Anthropogenetic methods for diagnosing genetic diseases have high sensitivity and specificity, which allows you to reliably determine the presence or absence of genetic defects.
3. Prevention of the spread of the disease: Anthropogenetic diagnostics allows you to determine the presence of genetic defects in parents and prevent the transmission of the disease to offspring. This is especially useful if the genetic disease is hereditary.

Disadvantages of anthropogenetic methods in the diagnosis of genetic diseases:

1. High cost: Some anthropogenetic diagnostic methods are expensive and not available to all patients. This can be a limitation for those who cannot afford to pay for such a study.
2. Technical limitations: Some anthropogenetic diagnostic methods require specialized equipment and expertise to perform. This may be difficult in settings where access to such resources is limited.
3. Key issues: Anthropogenetic diagnosis may raise a number of ethical or moral concerns, particularly when it comes to interfering with a person's genetics or making decisions about early pregnancy loss and sex selection.
4. Psychological consequences: Learning that you or your child has a genetic disorder can cause negative emotions and psychological distress for patients and their families. This may require additional psychological support and resources.[6]

Conclusion. In conclusion, it can be noted that there are certain requirements and rules for obtaining the results of any analysis or medical examinations, some of which are as follows:

1. Knowledge of the basics of genetics: it is necessary to understand the basic principles of heredity and mutations of genetic material, the use of anthropogenetic methods in the diagnosis of hereditary diseases.
2. High specialization: Diagnosis of genetic diseases requires highly qualified and specialized specialists, as this requires the correct identification and interpretation of genetic data.
3. Availability of special equipment: to implement anthropogenetic methods for diagnosing genetic diseases, special equipment such as DNA sequencers, polymerase chain reactions, etc. is required.
4. Ethical standards: when using anthropogenetic methods in the diagnosis of hereditary diseases, it is necessary to adhere to ethical standards and ensure the confidentiality of the patient and his genetic data.
5. Availability of sufficient samples: To conduct research and diagnose genetic diseases, it is necessary to have a sufficient number of samples of genetic material to ensure reliable results.
6. Reliability and accuracy of methods: Anthropogenetic methods must be reliable and accurate to correctly diagnose hereditary diseases.
7. Availability of standardization and quality control: The use of anthropogenetic methods in the diagnosis of genetic diseases requires the presence of standards and a quality control system to minimize the likelihood of errors and obtain reliable results.
8. Compliance with legislation: When using anthropogenetic methods in the diagnosis of hereditary diseases, it is necessary to comply with the legislation of the country where the research is being conducted, including on obtaining patient consent for genetic diagnosis.

References:

1. Hereditary diseases: what they are, types, characteristics and examples. <https://uz.warbletoncouncil.org/enfermedades-hereditarias-278>
2. Causes of hereditary diseases. <https://azkurs.org/irsiy-kasalliklaring-kelib-chiqish-sabablari.html#:~:text=Irsiy%20kasalliklar%20%E2%80%93%20genetic%20informatsiya%20%28irsiy%20akborot%29ning%20buzilishi,mutatsiyalar%20lardir%20tufayl%20paydo%20bo%E2%80%98lib%2C%20nasldan%20naslga%20o%E2%80%98tadi.>
3. Human genetics. https://uz.wikipedia.org/wiki/Human_genetics
4. The main methods of anthropogenetics: genealogical, twin, cytogenetic, biochemical, population-statistical, dermatoglyphic. <https://azkurs.org/taqdimot-mavzu-genetika-tadykilot-uslublari.html?page=2>
5. Biochemical Anthropogenetic Method as a Basis for Diagnostics and Prevention of Hereditary Pathology. T. I. Shevchuk. 2019.
6. Analysis of anthropogenic impact on the environment, measures to reduce it, and waste management. Nadezhda A. Eremeeva, Olga A. Savoskina, Lyudmila M. Poddymkina, Khamzat A. Abdulmazhidov, Oshibka! Nedopustimyy obekt hypersylki. and Abdurachman G. Gamidov. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10089261>