

# Clinical Application and Ethical Study of Medical Genetics Testing

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**Abstract:** With the rapid development of genetics technology, medical genetics testing has become an important part of modern medical practice. These tests are able to reveal individual genetic information, which is important for predicting disease risk, guiding treatment choices and promoting health management. However, the sensitivity and complexity of genetic information also raises numerous ethical issues. Types and applications of medical genetics tests are discussed and ethical principles that must be considered when applying these tests in clinical practice are presented, including respect for autonomy, the principle of non-harm, the principle of impartiality and the principle of beneficence.

**Keywords:** Medical genetics testing; Clinical applications; Ethical issues

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## Introduction

Medical genetic tests are increasingly used in clinical applications and provide an important tool for early diagnosis, risk assessment and personalised treatment of diseases. However, these tests also pose a number of ethical challenges, including issues of privacy protection, informed consent, genetic discrimination, and resource allocation. With this in mind, this article aims to provide healthcare professionals, policy makers and patients with insights on how to balance the benefits of genetic testing with ethical responsibilities.

### 1. Definition of medical genetics testing

Medical genetic testing refers to a series of medical practices in which an individual's genetic material (e. g. , DNA, RNA, chromosomes) is analysed to diagnose hereditary disorders, assess genetic risk, guide treatment plans, or conduct genetic counselling through biochemistry, molecular biology, cytogenetics, and other technological means. Such testing can reveal whether an individual carries certain genetic variants that may contribute to the development of a disease or increase the risk of a disease under certain conditions<sup>[1]</sup>. The scope of medical genetic testing is broad and includes, but is not limited to, testing for single-gene disorders, such as cystic fibrosis and Huntington's chorea; testing for chromosomal abnormalities, such as Down's syndrome and Edward's syndrome; and assessing genetic risk for complex diseases (e. g. , cancer, cardiovascular disease).

### 2. Clinical applications of medical genetics testing

#### 2.1 Genetic disease diagnosis

Genetic disease diagnosis is one of the core applications of medical genetics testing, which involves confirming the diagnosis and risk assessment of hereditary diseases. Genetic diseases can be divided into three major categories: single-gene disorders, chromosomal abnormalities and polygenic genetic diseases, and the testing methods and diagnostic strategies for each category have their own characteristics. Testing for single-gene disorders is usually performed for specific genetic mutations that contribute to the development of a genetic disease. Through molecular genetics techniques, such as polymerase chain reaction (PCR) and gene sequencing, variations in disease-causing genes can be precisely identified. Such tests are particularly important for the diagnosis of familial genetic diseases and help to determine the inheritance pattern and risk of the disease. Testing for chromosomal abnormalities, on the other hand, focuses on structural and quantitative changes in chromosomes that may lead to clinical symptoms such as mental retardation and growth retardation. Commonly used tests include karyotyping, fluorescence in situ hybridisation (FISH) and chromosome microarray analysis (CMA), which can detect small deletions or duplications of chromosomes. Risk assessment of polygenic genetic disorders is more complex, as such disorders are usually caused by a combination of multiple genes and environmental factors. Methods such as genome-wide association studies (GWAS) and polygenic risk scoring (PRS) allow assessment of an individual's genetic risk of developing a disease. Such assessments can help in early prevention and intervention, but caution is still needed for specific clinical applications, as the genetic mechanisms of polygenic genetic diseases are not yet fully understood. Genetic disease diagnosis not only provides patients with a basis for confirming the diagnosis, but also provides families with important information for genetic counselling and future fertility planning<sup>[2]</sup>.

#### 2.2 Genetic counselling

Genetic counselling is an important part of the field of medical genetics, which aims to provide individuals or families with information,

risk assessment, prevention strategies and interventions for genetic disorders. Risk assessment for genetic diseases is one of the core components of genetic counselling. By taking a detailed family history and analysing the results of genetic testing, genetic counsellors can assess an individual's or family member's risk of developing a specific genetic condition. This assessment includes not only the risk of known genetic diseases, but may also involve the prediction of genetic risk for future generations. The results of the risk assessment are important in guiding family members' reproductive decisions and lifestyle adjustments. Genetic disease prevention and intervention is another key task of genetic counselling. Based on the results of risk assessment, genetic counsellors will provide personalised prevention advice such as lifestyle changes, regular screening and early diagnosis. For patients with diagnosed genetic diseases, genetic counsellors will also provide advice on treatment options, including medication, surgical intervention or gene therapy. In addition, genetic counselling involves guidance on assisted reproductive technologies (e. g. in vitro fertilisation combined with pre-implantation genetic diagnosis of embryos) to help families at risk of genetic disorders to produce healthy offspring. The goal of genetic counselling is to help individuals and families make informed decisions, reduce the psychological and social burden of genetic disorders, and promote healthy lifestyles<sup>[3]</sup>.

### 2.3 Reproductive medicine

Reproductive medicine is an important field of application of medical genetics, especially in assisted reproductive technologies and pre-implantation genetic diagnosis (PGD) of embryos. The development of these technologies offers couples at risk of genetic disorders the possibility of having healthy offspring. Genetic testing in assisted reproductive technologies includes genetic evaluation of sperm and eggs, as well as genetic screening of embryos produced during in vitro fertilisation (IVF). These tests can help identify gametes or embryos that carry a risk of genetic disease, thereby screening for healthy embryos prior to embryo transfer. For example, by analysing the genome of a sperm or egg, the risk of passing on a genetic disease can be assessed, while at the early stages of embryo development, by sampling the embryo for genetic testing, it can be determined whether or not the embryo carries a specific genetic variant. Preimplantation Genetic Diagnosis (PGD) is an important component of Assisted Reproductive Technology (ART) that allows the genetic analysis of embryos before they are implanted in the mother's uterus. PGD is usually used for couples with a family history of a monogenic genetic disorder or for couples at risk of chromosomal abnormalities. With PGD, embryos can be tested to see if they carry the genetic mutation or chromosomal abnormality that causes the genetic condition. Only those embryos that are genetically normal are selected for transfer, significantly reducing the risk of passing a genetic condition to the next generation. Genetic testing in reproductive medicine not only improves the chances of having healthy offspring, but also provides couples with more reproductive options. Therefore, when promoting and applying these technologies, multiple scientific, ethical and social considerations need to be taken into account to ensure their rational and responsible use.

## 3. Ethical issues in medical genetics testing

Medical genetics testing involves a complex set of ethical issues that centre on how to balance the benefits of scientific advances with the rights and interests of individuals and society. Privacy and confidentiality are key issues, as genetic information may reveal an individual's health risks, family history, and underlying genetic characteristics, and this information needs to be strictly protected<sup>[4]</sup>. Informed consent is necessary, and patients must be fully informed about the purpose of the test, the process, the potential risks, and the implications of the results in order to make autonomous decisions. Genetic testing may raise issues of discrimination, for example, in the work or insurance arena, where individuals may be treated unfairly because of their genetic information. Equity in resource allocation is also a challenge, as not all people have equal access to these testing services. Genetic testing may reveal genetic links between family members, which may affect family relationships and the distribution of responsibilities. Medical genetic testing must therefore be implemented with care to ensure that the clinical benefits of testing are maximized while respecting individual autonomy, protecting privacy, avoiding discrimination and promoting equity.

## 4. Application of ethical principles in medical genetic testing

### 4.1 Respect for autonomy

Respect for autonomy is one of the core principles of medical ethics, which emphasises the right of patients to make autonomous choices about their medical decisions. The importance of patient autonomy in decision-making lies in the fact that it safeguards the fundamental freedom and dignity of the individual and allows patients to decide on treatment options based on their own values, beliefs and preferences. This autonomy is the basis for a trusting relationship between the patient and the doctor and is the key to personalised medicine. To ensure patient autonomy, doctors and healthcare teams should provide adequate information and education to help patients understand their condition, treatment options and their potential risks and benefits. In addition, physicians should respect patients' decisions, even if they may be inconsistent with the physician's recommendations. The informed consent process is an important part of ensuring patient autonomy, and it requires physicians to obtain explicit consent from patients before providing any treatment or intervention.

### 4.2 Principle of Do No Harm

The principle of non-harm is one of the fundamental principles in medical ethics, which requires that medical practice should minimise

harm to patients. This principle is particularly important in the context of genetic testing, where test results may pose psychological and social risks. For example, genetic test results may reveal the risk of future disease in a patient or a member of his or her family, which may lead to anxiety, depression, or strained family relationships. In order to minimise harm, healthcare professionals should fully assess potential psychological and social risks and communicate thoroughly with patients before conducting genetic testing. This includes providing genetic counselling to help the patient understand the significance and possible consequences of the test results, as well as discussing how to cope with the information. In addition, it should be ensured that patients volunteer for testing in a fully informed manner and that the necessary psychological support and social resources are provided<sup>[5]</sup>.

#### 4.3 Principle of Justice

The principle of impartiality is an important component of medical ethics, which requires that fairness be maintained in the allocation of healthcare resources and the provision of services. In the field of genetic testing, this means ensuring that all individuals, regardless of socioeconomic status, race, gender, or other characteristics, have equal access to quality testing services. Equity in resource allocation requires that policymakers take into account the needs and interests of different groups and avoid bias and inequality when allocating limited health care resources. This may involve the development of equitable insurance coverage policies, the provision of financial assistance, or the establishment of publicly funded testing programmes. Universal access to testing services, on the other hand, emphasises removing barriers to accessing genetic testing and ensuring that services are accessible to all. This includes providing testing services in remote or resource-poor areas and raising public awareness of the importance of genetic testing through education and information.

#### 4.4 The Beneficence Principle

The Beneficence Principle is one of the core principles of medical ethics, which emphasises that medical practice should be designed to maximise the benefit to patients. In the context of genetic testing, this principle requires that test results should have clear clinical value, i. e. be able to provide useful information for the diagnosis, treatment or health management of patients. To maximise the benefits of testing, it is first necessary to ensure the accuracy and reliability of the test in order to provide an accurate assessment of genetic risk. Secondly, healthcare professionals should communicate adequately with patients to explain the implications of the test results and develop a personalised healthcare plan based on the results. In addition, consideration should be given to combining genetic testing with other clinical information to improve the accuracy of diagnosis and treatment. Follow-up medical and support services, such as genetic counselling, psychological support and lifestyle guidance, are also needed to ensure that the benefits of testing are maximized.

### 5. Conclusion

In summary, the clinical application of medical genetic testing offers great potential for improving the quality of healthcare and personalising treatment. However, the ethical issues of these tests cannot be ignored and need to be properly addressed in clinical practice. While promoting genetic testing, it is important to ensure that patient privacy is protected, informed consent is respected, genetic discrimination is prevented, and resource allocation remains equitable. In addition, recommendations are made to strengthen ethical education and develop clear ethical guidelines to ensure the responsible and ethical use of medical genetic testing. Through these measures, the clinical benefits of genetic testing can be maximised while protecting the rights of individuals and society.

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### References

- [1] Yang Shuting, Xi Hui, Peng Ying, et al. Reflections on the Competency Training of Resident Physicians in Medical Genetics [J]. Chinese Journal of Graduate Medical Education, 2023, 7 (06): 460-462+466.
- [2] Zhao Shuhua. Analysis of research hotspots and development trends in medical genetics teaching [J]. Basic Medical Education, 2022-24 (03): 161-164.
- [3] Li Zhenhong, Shen Lihua. Application analysis of medical cytogenetic testing technology in the field of maternal and child health [J]. Contemporary Medicine, 2020, 26 (21): 66-68.
- [4] Sun Yanmei. Application of different detection techniques in prenatal genetic diagnosis [D]. Hebei Medical University, 2020. 000459.
- [5] Zhou Ji, Tang Liang, He Qizhi. Research on the construction of a formative evaluation system for medical genetics [J]. Health Vocational Education, 2019, 37 (23): 29-30.

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