RESEARCH ARTICLE

Genetic Testing and Genetic Consultation

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Abstract: The mutation genetic material, including genetic mutations or chromosome aberration, is the source of genetic disease happen, is also different from other diseases of the basic characteristics. Vigorously developing test and genetic screening, timely detection genetic disease patients and virulence genes carriers, is the only feasible method to improve population quality, promote a happy family, social prosperity, prosperous country. Genetic counseling for detection genetic disease patients and virulence genes carriers, and effective and feasible marriage guidance, birth guidance, play an important role in reducing the birth of the sick children and preventing the happening of the disease of children. In prenatal diagnosis of the fetus involved in disease treatment of moral choice in the problem, follow the four basic principles: first, respect the couple's choice; second, don't damage the individual and family; third, reliable prenatal diagnosis results; fourth, voluntary prenatal diagnosis and genetic counseling. These standards in all countries of the world have undoubtedly commonalities.

Keywords: Genetic diseases; genetic testing; genetic counseling; ethic

1. Genetic testing

Hereditary diseases differ from other diseases in that they are caused by changes in genetic material and can therefore be transmitted vertically through heredity between the upper and lower generations in a certain way, and among related individuals such as parents and children, brothers and sisters, there is a certain degree of genetic inheritance. Incidence ratio. Therefore, in a family member, hereditary diseases can not only occur vertically between the upper and lower generations, but also horizontally between brothers and sisters, resulting in the phenomenon of family aggregation of hereditary diseases, thus causing one of the characteristics of the seriousness of hereditary diseases.

In addition to the above-mentioned characteristics different from the general disease, genetic disease itself also has a special severity, manifested as the disease has hereditary, congenital and lifelong, because there is still no effective treatment, patients often worsen the disease, life-long involvement until death, to the patient himself and his ly brought Great pain and heavy burden.

1.1 The incidence of hereditary diseases, the harmfulness to the quality of population and population.

Genetic disease is a disease caused by genetic material, that is, mutation. The material basis of genetic diseases is genetic material mutation, including gene mutation or chromosomal aberration, is the root of genetic diseases, but also different from the basic characteristics of other diseases. In recent years, with the development of medicine, the acute infectious diseases that endangered human health in the past have been effectively controlled. Chronic diseases, such as malignant tumors, hypertension, diabetes, cardiovascular diseases, autoimmune diseases, and monogenic genetic dis-

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eases, which affect human health and quality of life, have become endangering human beings. The important factors of health, these diseases have a certain genetic basis, how to prevent and control the occurrence of such diseases has become the primary task of basic medicine and clinical medicine. With the rapid development of various branches of medical genetics such as cytogenetics, molecular genetics and clinical genetics, the basic genetic principles of many diseases have been clarified, which not only reveal the transmission laws and pathogenesis of many genetic diseases, but also develop some effective genetic methods for diagnosis, treatment and prevention of genetic diseases[1].

Although the incidence of human monogenic diseases is relatively low in the population, ranging from 1/106 to 1/104, the incidence of some diseases is relatively high, for example, the incidence of red-green blindness in men can reach 5%-7%. It is estimated that 3%-5% of the population suffers from various genetic diseases. For polygenic inherited diseases, although only more than 100 diseases have been found, the incidence of each disease is high, such as the incidence of essential hypertension is about 6%, coronary heart disease is about 2.5%, peptic ulcer is about 4%, asthma in China is 0.5% ~ 2%, even up to 5% in the southern region. Therefore, the total estimated population of 15%. People with ~20% suffer from various genetic diseases. The incidence of chromosomal abnormalities was 0.5%~1% in the population. Obviously, if the above three types of genetic diseases are taken into account, the incidence of genetic diseases in the population is as high as 20%-25%. If the incidence of somatic genetic diseases such as malignant tumors and acquired genetic diseases are also taken into account, the incidence of genetic-related diseases in the population is even higher. Therefore, vigorously carrying out genetic testing and screening, timely detection of patients with genetic diseases and carriers of pathogenic genes, and effective and feasible marriage guidance, fertility guidance to reduce or prevent the occurrence and incidence of genetic diseases are the only feasible way to improve the quality of the population, promote family happiness, social prosperity, and prosperity of the country. Law.

1.2 The positive significance and current situation of genetic testing and genetic screening in the prevention and treatment of genetic diseases.

The development of modern medicine and biotechnology has greatly promoted our understanding of birth defects and genetic diseases, but also rapidly increased the possibility of prevention and treatment of birth defects and genetic diseases. These advances not only have a profound impact on human health, but also on a series of social, ethical and legal issues. With the rapid development of genetic knowledge, the scope of genetic testing, screening, diagnosis and counseling is rapidly expanding. For some susceptible groups, through marriage and fertility guidance, changing lifestyle, providing some simple treatment, it is possible to prevent the occurrence of some serious genetic diseases. Thus, the development of modern medicine has played an active and effective role in improving the quality of the population, improving people's health and quality of life, preventing and reducing the occurrence of genetic diseases[2].

In order to prevent the birth of a fetus with congenital defects and genetic diseases, the key is to identify the individual's own risk of genetic disease and/or whether the disease can be passed on to the offspring of the individual. Therefore, genetic risk testing and screening involve genetic family identification and genetic population screening. At the same time, the development of genetic testing and screening, but also with the national or regional public health intervention system and planning, involving the corresponding laws and regulations. Many congenital and genetic diseases can be effectively prevented by providing the public and health workers with genetic risk and population screening techniques, information and basic diagnostic measures. The main measure to determine genetic risk is whether the disease is hereditary or sporadic. The incidence of sporadic diseases, such as chromosomal diseases, especially congenital stupidity, is affected not only by maternal reproductive age, but also by various environmental factors. Therefore, each pregnant woman's fetus, large or small, is at risk of congenital stupidity. Thus, simple and reliable population screening is effective in preventing the occurrence of congenital stupidity. The birth of a fetus with idiocy. At present, prenatal maternal serum screening has been carried out all over the world to assess the risk of fetal congenital stupidity and neural tube defects, which plays a positive role in reducing the birth of such undesirable fetuses, improving the quality of the birth population, relieving family pain and social burden.

With a better understanding of genetic diseases and the development of corresponding public health measures, some genetic metabolic diseases can be prevented from developing pathological symptoms and mental retardation by

early intervention and treatment, and all members of the population at risk should be screened for this known disease. Check. Neonatal screening has been widely carried out for some treatable hereditary metabolic diseases, such as phenylketonuria (PKU), congenital hypothyroidism and other diseases in the world, and corresponding screening policies have been established; however, there is no definite evidence that timely diagnosis of certain hereditary diseases can effectively change the situation. Whether neonatal screening should be carried out for diseases in the bed process, such as cystic fibrosis and progressive muscular dystrophy, remains controversial. For some X-linked recessive inherited diseases, such as hemophilia A, progressive muscular dystrophy and autosomal dominant inherited diseases, such as Hutington's chorea and spinocerebellar ataxia, heterozygosity or pre-symptomatic screening is carried out according to family clues to provide marriage and fertility guidance for the positive women and to provide pregnant women with guidance. Prenatal diagnosis is necessary to prevent the birth of a fetus suffering from this kind of genetic disease. In areas with a high incidence of certain genetic diseases such as thalassemia and glucose-6-phosphate dehydrogenase deficiency, on the basis of identifying the carriers in the family members, check whether the spouse is a heterozygote of the disease, or include the screening of heterozygotes in the pre-marital examination, first check one, if it is a heterozygote, then determine the other. Is it heterozygote? In addition, with the completion of the Human Genome Project, people's awareness of complex genetic diseases, such as hypertension, coronary heart disease, diabetes, myocardial infarction, psychosis and some familial malignancies, which have high incidence and great harm to human health, is also increasing. Pre-screening for the early symptoms of these diseases is necessary. Investigation can provide some effective preventive and early intervention measures to effectively reduce the incidence of such diseases.

These genetic tests and screening have played an active and effective role in reducing the incidence of genetic diseases, improving the quality of the population and public health, and promoting the development of human society. To make genetic testing and screening truly serve the health of the general public, in addition to the orderly and healthy development of testing and screening programs and the protection of corresponding policies and regulations, extensive social publicity and public education should be carried out before the screening project is launched so that the screened public can understand the purpose of genetic screening. And importance, and consciously accept the provision of genetic testing or screening services.

1.3 Ethical issues and principles of genetic testing and genetic screening

As far as the ethics of genetic testing and screening services is concerned, the main issues involved include the formulation of public health strategies, public understanding and acceptance of genetic testing and screening, and accurate analysis and processing of testing and screening results. Therefore, genetic testing and screening should comply with the basic ethical principles of medical genetic services, namely autonomy, benevolence, harmlessness and justice[3].

When formulating policies and strategies for genetic testing or screening, government public health administrations and medical and health service institutions should consider both the social and public welfare effects of preventing and controlling genetic diseases on improving the population quality and public health of social groups and promoting the orderly and healthy development of human society. Efficiency should also take into account the individual and family's own will and action autonomy. The fundamental purpose of medical genetic services is to benefit patients and the general public. Testing or screening should be based on the principle of public benefit. Testing or screening should be able to effectively prevent and control the harm of genetic diseases to human health, thereby reducing or eliminating the risk of genetic diseases to patients. The injuries and pains caused by myself and his family, as well as the heavy financial burden of illness on the family and society of the patients. Any genetic testing or screening that is harmful to the public for commercial gain, regardless of the public interest, or even against ethical norms should be strictly prohibited.

In general, the application of genetic testing or screening must follow the following principles. (1) The technology used must be reliable, and since genetic testing or screening does not equal diagnosis, there must be clear diagnostic techniques for suspicious cases detected through testing or screening to ensure the scientificity and effectiveness of genetic testing or screening, so that testing is truly conducive to improving and promoting public health. Kang, especially for prenatal and neonatal screening, should have strict technical specifications. For testing or screening laboratories, the

government's health administration should examine and certify their qualifications to prevent any individual or enterprise from exaggerating the practical role and significance of genetic testing or screening for commercial profit purposes and over-publicizing it. (2) Genetic testing or screening should be carried out on the basis of the principle of informed consent of the subjects and against compulsory implementation. Genetic testing or screening involves not only the problem of medical adaptation to the population, but also many social problems, such as social culture, moral concepts, customs and habits, and economic affordability. Therefore, in addition to strengthening the positive propaganda and education of testing or screening, the public should be aware of the positive and beneficial results or possible adverse consequences of testing or screening for the population adapted to various tests or screening for themselves and their families, such as prenatal serum of pregnant women with fetal chromosomal abnormalities. Screening can effectively prevent or reduce the birth of fetuses with mental retardation, thus avoiding the birth of these congenital defects of fetuses on the family harm, as well as the family and society brought heavy economic burden; however, the results of prenatal screening can also cause anxiety of pregnant women and their husbands and families, because of the screening technology itself. There are certain limitations, in some cases will be omitted cases, and harm to the families of the subjects. From an ethical point of view, individuals'wishes should be respected for genetic testing or screening, and informed consent should be signed by the medical institutions providing services to the testing or screening subjects. (3) genetic testing or screening should bring obvious benefits to the subjects. If prenatal testing or screening is necessary for the timely diagnosis of fetuses with severe monogenic or chromosomal abnormalities such as Down's syndrome before childbirth, medical termination of pregnancy may be timely taken to prevent these fetuses according to the wishes of pregnant women and their families. The adverse effects of birth on family and society. Although there are many ethical debates about prenatal genetic testing or screening, such as respect for life and the rights of the fetus, these tests or screening are evident in safeguarding the well-being of pregnant women and families, improving the quality of the birth population and ensuring the prosperity of mankind.

Screening for carriers of genetic diseases will help to reduce the incidence of some single-gene recessive genetic diseases in the population, and has a certain practical significance for the guidance of marriage and childbirth. For couples who are both carriers of disease genes, if pregnancy can be diagnosed by prenatal fetal gene, once the fetal disease is identified. Early termination of pregnancy with certain recessive genetic disorders, such as beta-thalassemia, cystic fibrosis, carrier screening for fragile X, and prenatal diagnosis, greatly reduces the incidence of these genetic disorders. For some delayed genetic diseases, such as Huntington's chorea, familial Alzheimer's disease, familial colon cancer, breast cancer, etc., or complex polygenic genetic diseases, genetic testing can predict the risk of disease early, so as to take timely intervention or preventive measures for delaying the occurrence of disease or alleviating symptoms. To improve the quality of life and life has a certain positive significance, but because any testing technology can not achieve 100% accuracy at present, therefore, for the population with positive results of genetic testing, professional personnel should make a scientific and reasonable explanation and corresponding guidance, to avoid the uncertainty of the results of the test results caused by sufferings. The mental harm of[4].

2. Genetic counseling

Genetic counseling is also known as genetic negotiation. According to the scope of genetic counseling, it can be divided into narrow sense genetic counseling and generalized genetic counseling. The narrow sense of genetic counseling is that a consulting physician simply calculates the recurrence risk of a certain genetic disease inquired by a consulting physician, which is seldom used at present, while the broad sense of genetic counseling is that the consulting physician and the consulting physician are concerned with the cause, diagnosis and treatment of a certain genetic disease in the family. A series of answers, discussions and discussions are carried out on the questions of hereditary mode, prognosis, and recurrence risk. Finally, the consultant puts forward corresponding medical suggestions for the consultant to make appropriate choices and decisions, and put them into practice with the assistance of the consultant, so as to achieve the best prevention and treatment effect and avoid the recurrence of the same hereditary disease. Son.

2.1 The role and purpose of genetic counseling

The role of genetic counseling lies in the following questions answered by genetic counselors through careful inquiry and understanding of the relevant counseling content: (1) whether the disease to be diagnosed is a hereditary disease; (2) suggesting the basis for hereditary disease; (3) methods of diagnosis; (4) the occurrence of the hereditary disease in the family, hereditary transmission The process; (5) the probability of onset or recurrence; (6) the countermeasures that can be taken; (7) how to choose the best eugenic measures.

The purpose of genetic counseling is to widely apply modern medical technology to reduce the incidence of genetic diseases, reduce family and social pressure and burden, and constantly improve the quality of the population. Therefore, as far as prenatal genetic counseling is concerned, in the process of taking eugenic measures, it must involve the relationship and interests of the patient himself, the patient's parents and the society. How to make the patient or his parents understand the serious harm of genetic diseases and coordinate and solve their interests is a complicated and difficult problem. . In short, in the process of genetic counseling and processing must be based on legal, social concepts, ethical norms and other principles to effectively solve and implement the problems faced. The general purpose of genetic counseling includes the following three aspects.

2.1.1 For patients themselves

(1) Understanding the causes of genetic diseases, so that patients rationally face the reality, in order to alleviate the psychological and mental pain and stress of patients; (2) to provide information on the disease, development trends and prognosis; (3) to provide possible treatment information. (4) provide genetic risks and the best measures to be taken.

2.1.2 For parents or couples

(1) Providing genetic information to alleviate guilt and anxiety; (2) identifying carriers to guide marriage and fertility planning; (3) helping to formulate fertility plans; (4) proposing feasible fertility plans for consanguineous couples in families at risk of disease; (5) providing advice and formulating feasible measures for couples at high risk Shi (6) to provide parents with children with inherited diseases and congenital malformations.

2.1.3 Social purposes

(1) to reduce the genetic load in the population; (2) to publicize genetic knowledge, so that people understand the serious harm of genetic diseases, improve the sense of good business; (3) to reduce the incidence and incidence of genetic diseases, and constantly improve the quality of the population.

Genetic counseling is an important part of medical genetic services. It plays a very important role in detecting patients with genetic diseases and carriers of pathogenic genes, providing effective and feasible marriage guidance and fertility guidance to reduce or prevent the occurrence and incidence of genetic diseases.

2.2 The principles of logic that should be followed in genetic counseling.

From the form of genetic counseling services, it can be divided into mandatory and non mandatory genetic counseling. What kind of genetic counseling service depends on the relevant laws and regulations of various countries. As far as ethics is concerned, ideal genetic counseling is regarded as non-mandatory counseling. Considering China's specific conditions, non-mandatory genetic counseling should be advocated without violating the relevant laws and regulations of China's "Marriage Law" and "Maternal and Infant Health Care Law".

In non-mandatory genetic counseling, attention should be paid to scientific nature, especially prenatal genetic counseling is a matter of deciding whether the offspring are healthy or not, and should not be rushed. Family history needs to be investigated clearly and accurately. If necessary, it should be examined and visited. It should not be judged and dealt with according to inaccurate materials. It is not easy to judge that a consultant should not give birth or terminate pregnancy. In short, genetic counseling should be considered from all aspects of society, family and individual. As a genetic counseling physician, more humanistic care should be provided to the counselors. We should pay attention to the influence of social and psychological factors on the promotion of eugenics. Eugenics is a social work. We need to publicize and popularize scientific knowledge to the public. The propaganda should be scientific and reasonable, avoid overstepping words, and make the masses neither too nervous nor too careless. In addition to understanding the physiological and pathological factors, genetic counselors should also understand the impact of social and psychological factors on diseases and eugenics in order to do a better job in genetic counseling. Good genetic counseling will lay a good

foundation for eugenics.

In the process of genetic counseling, genetic counseling physicians should strive to provide accurate, comprehensive and impartial information for counselors and family members so that they can make decisions. The counselors should be sympathetic and tolerant, and provide guidance to help people make their own decisions. In the process of consultation, the consulting doctor should avoid purposefully biased information, and prevent misleading or inducing people to make certain decisions that the consulting doctor considers to be the best. Genetic counseling does not tell people what to do, but helps counselors understand and express their own values and ultimately make their own decisions.

The basic principles of genetic counseling are: to inform couples about the nature and severity of fetal malformations; whether there are currently medical treatments and possible sequelae after birth; possible genetic patterns; and to alleviate couples'anxiety and guilt psychologically. Couples are encouraged to make their own choices as to whether to continue or terminate pregnancy; couples who require termination of pregnancy should respect their wishes and, on the basis of relevant legal and medical ethical principles, hospitals may take abortion measures to terminate pregnancy. For some cases with slight fetal structural abnormalities but unclear diagnosis, such as simple pyelonectasia, femoral shortening, enhancement of intestinal echo, choroidal cyst, etc., the cases were analyzed and the possible causes and prognosis of the couple were informed. If necessary, karyotype analysis and other examinations were performed to arrange the cases. In addition to serious diseases that may affect the quality of life of the fetus. On the basis of respecting the wishes of couples, prenatal follow-up should be strengthened to avoid blind induced abortion.

In prenatal genetic counseling and diagnosis, the medical staff concerned should strictly follow the principle of informed choice, and the doctors engaged in prenatal genetic counseling and prenatal diagnosis have the responsibility and obligation to obtain prenatal diagnosis results based on existing laboratory tests and clinical analysis, and to inform both spouses of the condition of the fetus and the possible prognosis. However, doctors should not have any suggestion or inducement to make further choices. On the basis of informed choice and according to the above principles, the husband and wife have the right to make their own choice. If both husband and wife wish to terminate pregnancy, they shall, with their consent and signature, terminate the pregnancy by artificial abortion according to Article 19 of Chapter III of the Law on Maternal and Infant Health Care, if they are incapable of doing so, with the consent and signature of their guardian. Due to the limited development of medical science itself, prenatal diagnosis based on laboratory and clinical data may be inconsistent with the actual situation. Medical staff have the right and obligation to make necessary explanations to both husband and wife when they choose to terminate pregnancy.

The ethical issue of induced abortion has always been a matter of concern and controversy. It involves the interests of ethics, women themselves, fetuses, families and society. In the early 1970s, L.R. Kass and P. Ramrey pointed out that the purpose of prenatal diagnosis was to abort a congenitally defective fetus, which violated the basic purpose of saving lives in medicine and the principle of equality for all; A. Milunsky and A. G. Motulsky believes that prenatal diagnosis can abort congenital defects, ensure the birth of more healthy babies and relieve the suffering of couples. It can be seen that relying solely on a certain principle can not adapt to the complexity of human society and the differences of existing moral standards. In fact, it is impossible to be totally divorced from the belief in certain special interests, such as family, occupation, region, nationality, religion, culture or ideal, or from the moral traditions of a particular society to talk about the ethics of induced abortion[5–7].

The fetus as a living body has its right to survival, but when we are faced with a seriously defective fetus, whether to allow it to be born, or to take appropriate measures to terminate pregnancy, is often a dilemma. Defected fetuses have the right to live, but after all, the fetus is different from adults, they lack the ability to think and choose. For example, patients with advanced cancer have the right to choose euthanasia in order to avoid further suffering. As far as ethics is concerned, although there are still different views, it has been recognized by law in some countries. For fetuses with congenital defects, the choice between birth and death seems to be decided by the parents of the fetus. It is difficult to establish a unified criterion for abortion of severely malformed fetuses. Generally speaking, it depends on the severity of the fetal malformation, the method of treatment, the economic status of the family and the quality of life of the fetus after birth, as well as the wishes and religious beliefs of the parents of the fetus[8].

As far as the current medical development is concerned, there is still no effective treatment for most fetuses with severe deformities. It is not against ethical and moral standards to terminate pregnancy by selective induced abortion. If a seriously defective fetus is not allowed to be born without abortion, it may cause great harm to pregnant women, families and children themselves. Imagine that if a couple gave birth to a severely defective baby, and medicine was at a loss, the child's life would be a painful ordeal. This situation will leave a heavy burden on parents'mind, life and economy. It will also bring about lifelong regrets in morality and ethics. Once a child has grown up and is able to think independently, he or she is likely to accuse his or her parents of letting him or her be born into the world, but of being unable to cope with their physical and psychological pain. To this end, the British "induced abortion act" enacted in 1967 allows abortion of seriously defective fetuses. The Chinese government enacted the "Mother and Child Health Care Law of the People's Republic of China" in 1994, which provides legal protection for the health and legitimate rights and interests of women and children. On the issue of moral choice in prenatal diagnosis, Mirensky proposed four basic criteria: first, respecting the choice between husband and wife; second, not harming the individual and family; third, the results of prenatal diagnosis are reliable; fourth, the voluntary nature of prenatal diagnosis and genetic counseling[9]. Despite differences in culture,[10] social system and moral standards, these norms are undoubtedly common in all countries of the world.

With the progress of society and the development of science, perhaps in the near future, when people can make early and safe prenatal diagnosis of genetic diseases or fetal congenital defects, and have the ability to carry out effective prenatal treatment, such as fetal intrauterine treatment, human somatic cell gene therapy, human embryo gene therapy, strict The principle of handling the abnormal fetus will also change accordingly. However, the norms of improving the quality and quality of life of the births,[11] respecting the rights and interests of couples and fetuses, and safeguarding the interests of individuals, families and society will not change.

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