Review Article

Empirical Risks of Genetic Counseling in Dental Perspective: An Overview

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Abstract

Since past few years, the science of genetics including human genetics has made rapid progress. Human genetics is much more than the study of mere hereditary diseases. Genetic predisposition may lead to the premature onset of common diseases of adult life such as cancer, coronary heart disease, diabetes, hypertension and mental disorders. Furthermore, it has been proved by studies that genetic factors influence oral conditions such as dental caries and periodontal disease. Genetic counseling is a communication process. The trained professionals help individuals and families deal with issues associated with the risk of or occurrence of a genetic disorder. It is at an exhilarating point in its professional evolution. This paper embraces about the genetic disorders related to dentistry and the role of public health dentist in genetic counseling.

Keywords: Diseases, Genetic counseling, Mutation

INTRODUCTION

Genetics is the study of genes at all levels from molecular to populations. As a basis for relatively rare developmental dysplasias, diseases and syndromes, which show a genetic cause or marked genetic influence becomes known, increasing attention is being paid to those genetic factors that influence more common conditions. An increased appreciation of how genetic factors interact with environmental factors to influence growth and pathology will lead to an increased understanding of pathogenesis and recognition that some groups or individuals may be more susceptible.[1]

The basic principles of genetics were laid down by Mendel and Galton towards the close of the 19th century. However, it is only during the past few years the science of genetics including human genetics has made rapid progress.[2] Human genetics is much more than the study of mere hereditary diseases. It has emerged as a basic biological science for understanding the endogenous factors in health and disease and the complex interaction between nature and nurture. The various branches of genetics are: Cytogenetics, biochemical genetics, clinical genetics, pharmacogenetics, immunogenetics, microbial genetics and so on.[3]

GENETIC PRINCIPLES

The genome contains the entire genetic content of a set of chromosomes present within a cell or an organism. Within the genome the genes that represent the smallest physical and functional units of inheritance that reside in specific sites. A gene can be defined as the entire DNA sequence necessary for the synthesis of a functional polypeptide molecule or RNA molecule. Genotype refers to the set of genes that an individual carries and in particular, usually, refers to the specific pairs of alleles that a person has at a given location of the genome. Phenotype is the observable properties and physical characteristics of an individual, as determined by the individual's genotype and the environment in which the individual develops over a period.[2]

STRUCTURAL CHROMOSOMAL ABNORMALITIES

They can be produced by any of the four mechanisms:

a. Deletion: Breaking away of a portion of a chromosome
b. Inversion: The broken part reattaches itself in reverse orientation
c. Translocation: Two chromosomes break and exchange their broke segments in reciprocal translocations
d. Duplication: An over-representation of a specific chromosomal region
e. Transverse centromeric division: Instead of dividing longitudinally, centromere divides itself into transverse plane forming an isochromosome.[1]

NUMERICAL CHROMOSOMAL ABNORMALITIES

a. Aneuploidy: The diploid chromosome number of the cell is not an exact multiple of its haploid number
b. Polypoidy: Chromosome count exceeds the diploid number and is also an exact multiple of its haploid number.[1]

GENE MUTATIONS

Mutation, is sudden genetic change. It is derived from Latin word "Mutatio" which means any change in form, quality or other characteristics. It can occur at molecular level, substituting one DNA base for another or adding or deleting a few bases, at the chromosomal level, where chromosome can exchange parts and genetic material. It can be classified as:

1. Germ-line mutation (constitutional mutation): The change occurs during the DNA replication that precedes meiosis.
2. Somatic mutation: The change occurs during DNA replication that precedes mitosis.[3]

ROLE OF GENETIC PREDISPOSITION IN COMMON DISORDERS

Although the limits of intelligence, physical ability and longevity are genetically determined, external and environmental influences such as infections, malnutrition and war have long been the main determinants of health and survival. Genetic predisposition may lead to the premature onset of common diseases of adult life such as cancer, coronary heart disease (CHD), diabetes, hypertension and mental disorders.

Cancer

It is not yet certain, but a genetic predisposition may be involved in as many as 10-25% of cases of cancer of the breast or colon. Numerous genes are identified that may affect susceptibility to tumor development.

CHD

It was generally believed that environmental factors alone cause CHD. But investigating family histories often uncover genetic risks. Mapping the human genome will make the genetic predisposition to CHD much easier. High blood pressure and high cholesterol levels, major risk factors in CHD are also genetically influenced.

Diabetes

Evidence for a genetic element in insulin-dependent diabetes mellitus has emerged from studies showing a higher concordance in identical twins than in non-identical twins. About 85% of cases in developed countries are of non-insulin dependent form of disease, which has particularly strong familial tendency.

Mental Disorders

Evidence from family and twin studies demonstrates the existence of genetic predisposition to some common mental diseases.[4]

INFLUENCE OF GENETIC FACTORS ON ORAL CONDITIONS

Genetics and Dental Caries

It is clear from many dietary studies that the variation in susceptibility to dental caries exists even under identical, controlled conditions. This implies that, because of genetic differences, certain environmental factors are potentially more cariogenic for some people than for others. Several investigators have studied the genetic aspects of dental caries in humans, using both the twin and the family pedigree approaches. The family observations by Klein and Palmer and Klein indicated that the children have caries experience remarkably similar to that of their parents when the susceptibility of the two parents is same. While, if the caries susceptibility of the parents is dissimilar the children's susceptibility tends to be more like that of the mother than that of the father. Mothers are the principle source of mutans streptococci to their infants, with a greater rate of transmission to female than male infants. A review of inherited risks for susceptibility to caries found evidence of an association between altered dental enamel development in defined populations and an increased risk of caries, as well as a relationship between host immune complex genes and different levels of cariogenic bacteria and enamel defects. Thus, the individual genotype may influence the

likelihood of intraoral colonization of cariogenic bacteria. Goodman *et al.* reported significant differences in salivary flow, pH and salivary amylase activity between monozygotic and dizygotic twins. Susceptibility to human dental caries is influenced to a significant, but minor degree by heredity. The genetic control is multifactorial in nature and implies considerable environmental influence.

Genetics and Periodontal Diseases

The periodontal disease state is often described as a local inflammatory disease with possible underlying systemic factors. Most genetic studies of a trait make use of families with multiple affected individuals or twins. Michalowicz *et al.*, after conducting studies on 63 monozygotic and 33 dizygotic twins concluded that 38-82% of the periodontal disease identified in these twins was attributable to genetic factors. According to Kornman, association of polymorphisms of inflammation mediating genes and periodontal disease in adult non-smokers indicate interleukin (IL) 1α and 1β genotype may be a risk factor. Progress has been made in the study of rare genetic conditions and syndromes that can predispose to periodontal disease or have periodontal disease as a relatively consistent component of their pleotropic effect. Leukocyte adhesion deficiency Type I and Type II are autosomal recessive disorders of the leukocyte adhesion cascade. Early onset periodontitis is a complex, oligogenic disorder (involving a small number of genes) with IL-1 genetic variation having important but not exclusive influence on disease risk.[3]

ADVANCES IN MOLECULAR GENETICS

DNA Technology

Synthesis of DNA probes with specific sequences that will bind to and identify any complementary DNA sequences that may be present. It is also done for rapid analysis of unknown DNA and the identification of mutations that give rise to disease. Comparisons of different genes and species help elucidate the mechanisms of evolution.

Gene Therapy

It is the introduction of a gene sequence into a cell with the aim of modifying the cell's behavior in a clinically relevant fashion. The gene may be introduced using a virus or by means of lipid or receptor targeting. It may be used in several ways, e.g. to correct a genetic mutation, to kill a cell or to modify susceptibility.

The Human Genome Project

The human genome project is an attempt to systematize the research on mapping and isolating human genes that is already in progress in many countries in order to create a single linear map of the human genome, with each coding gene defined and sequenced. Agencies with a role in coordinating human genome data include UNESCSO, the Genome Data Base, HUGO, the National Institute of Health/Department of Energy (USA) and the European Union.

The Human Genome Diversity Project

The major objective is to define the genetic relationships between human populations and interpret them in terms of natural selection, genetic drift, migration etc.,[4]

FACTORS INFLUENCING GENE FREQUENCIES

a. Mutation: Mutation implies a change in the genetic material of an organism that results in a newly inherited variation. It is now recognized that mutant genes are so widespread in their occurrence that every one of us might be harboring a few or many of them. External influences like ionizing radiation and certain chemicals are capable of producing mutations.
b. Natural selection: It is a process whereby harmful genes are eliminated from the gene pool and genes favorable to an individual tend to be preserved and passed on to the offspring.
c. Population movements: Industrialization, increased facilities for earning, ways of living and education, people are moving-sometimes on a large scale from rural to urban areas. There is also the migration of population between countries, which will lead to changes in the distribution of genes, affecting both the areas of immigration and emigration.
d. Breeding structure: If all marriages were to occur in a random fashion, the effect would be attainment of a genetic equilibrium. In practice. However matings tend to occur selectively within various subgroups based on religion, economic and educational status and family relationships.
e. Public health measures: Advances in public health and medical care services do affect the genetic endowment of people as a whole. The carriers of hereditary diseases, malformations and constitutional weaknesses are able to survive and pass their genes to their progeny.[5,6]

ROLE OF PUBLIC HEALTH PROFESSIONALS

Health Promotional Measures

1. Eugenics: Aims to improve the genetic endowment of the human population
2. Euthenics: Mere improvement of the genotype is of no use unless the improved genotype is given access to a suitable environment, an environment that will enable the genes to express themselves readily. Throughout the course of history, man has been adapting environment to his genes more than the

adapting environment to his genes. The solution of the human race does not lie in contrasting heredity and environment, but rather in the mutual interaction of heredity and environment factors

3. Genetic counseling: Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- Education about inheritance, testing, management, prevention, resources and research
- Counseling to promote informed choices and adaptation to the risk or condition.[6]

Genetic Counseling

Genetic counseling is a communication process in which trained professionals help individuals and families deal with issues associated with the risk of or occurrence of a genetic disorder.[7]

Genetic counseling may be prospective and retrospective:

1. Prospective genetic counseling: This allows for the true prevention of disease. It requires identification of heterozygous individuals for any particular defect by screening procedures and explaining to them the risk of their having effected children if they marry another heterozygote of the same gene. If it can be done, there will be a reduction in the incidence of diseases like sickle cell anemia and thalassemia
2. Retrospective genetic counseling: Most genetic counseling is at present retrospective, the hereditary disorder has already occurred within the family. The methods suggested under retrospective genetic counseling are: Contraception, termination of pregnancy and sterilization depending upon the attitudes and cultural environment of the couples involved.[2]

Genetic counseling is at an exciting point in its professional evolution. The explosion of knowledge and multiple opportunities for patients to learn about their genetic risks have far outpaced advances in understanding the complex psychosocial aspects of genetic counseling practice.

1. Genetic information is key: Providing information about "perceived or known genetic contributions to disease" and engaging in discussion with patients about this information is a particularly unique aspect of genetic counseling
2. Relationship is integral to genetic counseling. The quality of the relationship developed between the genetic counselor and patient is as important as genetic information. Genetic counseling "is a relationally-based helping activity whose outcomes are only as good as the connection established between the counselor and patient."
3. Patient autonomy must be supported. Patients should be as self-directed as possible regarding genetic counseling decisions. The counselor is an active participant, working with the patient's individual characteristics and family and cultural context to facilitate informed decisions. However, an essential aspect of this tenet is that "the patient knows best"
4. Patients are resilient. Most patients have the strength to deal with painful situations. Genetic counselors therefore, encourage patients to draw upon their inner resources (coping strategies) and support systems and resources to make decisions and arrive at the acceptance of their situation
5. Patient emotions make a difference. Patients experience a multitude of emotions that are relevant to genetic counseling. "Patient emotions interact with all aspects of genetic counseling processes and outcomes, for instance, affecting their desire for information, their comprehension of information, the impact of information on their decisions, their willingness and ability to connect with the counselor, their desire for autonomy, and their perceived resilience."[6,8]

Specific Protection

There is increased attention toward the protection of individuals and whole communities against mutagens such as X-rays and other ionizing radiations. Patients undergoing X-rays should be protected against unnecessary exposure of the gonads to radiation. Rh hemolytic disease of the newborn is now preventable by immunizing with anti-D globulin.[2]

Early Diagnosis and Treatment

1. Detection of genetic carriers
2. Prenatal diagnosis
3. Screening of newborn infants
4. Recognizing preclinical cases.[2]

LATEST ADVANCES IN GENETICS

1. DNA vaccination
2. Biochips
3. Human cloning
4. Recombinant DNA technology
5. Stem cell therapy.[10]

CONCLUSION

Genetics is an ever expanding branch of science that will have a major impact on the future health care system. Technologically, it is the most advanced branch of life

sciences till today. In the future, it may be used as an adjunct to standard therapeutic procedures than an independent and self-sufficient treatment system. Although the medical potential is bright, the possibility for misuse of genetic engineering technology looms largely, so society must ensure that gene therapy is used only for the treatment of genetic diseases.

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