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Treatment of genetic disorders by medical genetics

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DESCRIPTION

Medical genetics is the branch of medicine that encompasses the diagnosis and management of hereditary disorders. Medical genetics contrasts from human genetics, in that human genetics is a field of scientific research that may or may not apply to medicine, while medical genetics refers to the application of genetics to medical care. For example, research on the sources and inheritance of genetic disorders would be considered within both human genetics and medical genetics, while the diagnosis, management, and advising people with genetic disorders would be considered part of medical genetics (Wade, 2015).

In contrast, the study of typically non-medical phenotypes such as the genetics of eye colour would be measured part of human genetics, but not necessarily appropriate to medical genetics (except in situations such as albinism). Genetic medicine is a fresher term for medical genetics and integrates areas such as gene therapy, personalized medicine, and the rapidly emerging new medical specialty, predictive medicine (Pollack, 2014).

Medical genetics comprises many different areas, counting clinical practice of physicians, genetic counsellors, and nutritionists, clinical diagnostic laboratory activities, and research into the causes and inheritance of genetic disorders. Instances of conditions that fall within the scope of medical genetics include birth defects and dysmorphology, intellectual disabilities, autism, mitochondrial disorders, skeletal dysplasia, connective tissue disorders, cancer genetics, and prenatal diagnosis. Medical genetics is increasingly becoming pertinent to many common diseases. Overlaps with other medical domains are beginning to emerge, as recent progresses in genetics are revealing etiologies for morphologic, endocrine, cardiovascular, pulmonary, ophthalmologist, renal, psychiatric, and dermatologic conditions. The medical genetics community is increasingly intricate with individuals who have undertaken elective genetic and genomic testing (Baltimore et al, 2015).

Although genetics has its roots back in the 19th century with the work of the Bohemian monk Gregor Mendel and other pioneering scientists. It started to develop, albeit slowly, during the first half of the 20th century. Mendelian (single-gene) inheritance was studied in a number of chief disorders such as albinism, brachydactyly (short fingers and toes), and hemophilia. Mathematical approaches were also devised and applied to human genetics (Urnov, 2015). Medical genetics was a late developer, evolving largely after the close of World War II when the eugenics movement had fallen into disrepute. The Nazi misuse of eugenics signalled its death knell. Shorn of eugenics, a scientific methodology could be used and was applied to human and medical genetics. Medical genetics saw an increasingly rapid rise in the second half of the 20th century and continues in the 21st century (Kolata, 2015).

CONCLUSION

Genetic studies can custom this admixture linkage disequilibrium to search for disease alleles with fewer markers than would be required otherwise. Association studies also can take advantage of the contrasting experiences of racial or ethnic groups, comprising migrant groups, to search for interactions between particular alleles and environmental factors that might influence health.

Under Medical Genetics, Genetic counselling is the practice of providing information about genetic conditions, diagnostic testing, and risks in the family members, within the agenda of non-directive counselling. Genetic counsellors are non-physician members of the medical genetics team who concentrate in family risk assessment and counselling of patients regarding genetic disorders.

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