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Prospective Review on Genetic Disorders

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Abstract

A genetic disorder is a genetic condition which is a result of one or more abnormalities in the genome, which is mostly observed from birth (congenital). Majority of the genetic disorders are rare and affect one in every several thousands or millions. These disorders may be hereditarily passed down from the parents' genes or may be resulted by new mutations or changes in the DNA. In the latter case, the disorder will be passed down unless it occurs in the germ line. A same disorder may be caused due to an inherited genetic condition or by new mutations and mainly due to environmental factors e.g. some types of cancers. A person with a genetic defect or abnormality will actually suffer from the disease generally depends on environmental factors and events in his development.

Keywords: ***Genetic disorders, Mutations, Mendelian disorders, Non-Mendelian disorders, Multifactorial and polygenic disorders***

Introduction

Genetic disorders are either hereditary disorders or a result of mutations. Some disorders may render advantageous, in certain conditions. There are numerous pathways for genetic defects, at present about 4,000 genetic disorders are known, with more being discovered. Cystic fibrosis is one of the most common genetic disorders; about 5% US population carry at least one copy of the defective gene.

Recent advances and understanding had revealed that almost all diseases have a genetic component. These mutations may not always manifest as disease and may only manifest as symptoms in the presence of environmental factors.

Genetic Disorders have different etiologies and result in the following type of disorders

1. Single gene disorders which include Mendelian Disorders and Non-Mendelian disorders [1-3]
2. Multifactorial and polygenic disorders
3. Disorders with variable modes of transmission
4. Cytogenetic disorder which include autosomal disorders and X or/and Y chromosome linked disorders

The severity of genetic disorders may range from minor deformation to risk of mental retardation or even result in death in few cases. Hence it is essential to have proper knowledge and precise understanding of mechanism of these disorders at molecular level and also throws light on the importance of prenatal diagnosis of these lethal, incurable and chronic disabling genetic diseases[4-12. People can be educated through literature, internet, family medical practitioners and genetic counselors etc. Open access journals provide access to the ongoing research all over the world, which is being published in them, without any restriction in the form of subscription charges. These open access journals help readers gain knowledge

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and understand about various genetic disorders on the other hand they also help the authors and researchers to globalize their research activity and gain popularity in the field [13-24].

In order to create awareness among the public and fight against these genetic disorders, physicians, geneticists, genetic counselors and researchers unite to form societies or associations. The main aim of these societies is to counsel and create awareness among the pregnant women and their family, parents and family members of the children with genetic disorders. Major societies like American Society of Human Genetics aims to improve public awareness about genetics they also support and promote researches in the field of genetics. Parkinsons On Move Foundation focus on educating people suffering with parkinson's and those dealing with parkinson's to improve their lives and give them a quality life, Macedonian Scientific Society for Autism is established by citizens-special educators, medical doctors, psychiatrists, allergologists, immunologists and other professionals from different scientific disciplines, and deals with investigation of mechanisms associated with autism, • evaluation of effects of rehabilitation on the people with autism, treatment of autism patients, supporting the parents of the children affected with autism, coordinating governmental and non-governmental organizations dealing with autism in Macedonian Republic**.** Autism community network works to serve the members of the autism community in South Texas**,** Thalassemia International Federation established in 1987 with a mission to develop as well as establish National Control Programs for the prevention and quality treatment of thalassemia and their vision is to establish equal access to excellent health care for patients suffering with thalassemia. These societies help in dissemination of recent scientific advancements among the researchers, physicians, geneticists and caretakers of people affected with genetic disorders so that they can provide better service to the patients and work together to improve their quality of life. The society enables the understanding of the disorders by creating awareness among the global communities[25].

International peer-reviewed scholarly journals like Journal of Genetic Disorders and Genetic Reports are publishing valuable research papers on topics like genetic syndromes and gene therapy, mutations and functional consequences, chromosomal aberrations and their effects, Genetic Counseling & Education , Multifactorial and polygenic (complex) disorders etc. Kazuaki Matsumoto and Masayasu Ohta had published a case report entitled 'A Case of Wolf-Hirschhorn Syndrome and Familial Mediterranean Fever' which discussed the difficulties in the diagnosis of these two genetic disorders.

Many conferences like 2nd World Congress on Human Genetics & Genetic Disorders provide a suitable platform for the enthusiastic upcoming researchers and students to present their research and ideas and get suggestions from the experts in the field, these types of interactions will help them flourish and excel in their field. Alireza Haghighi presented a poster entitled 'Next generation sequencing: New hope for patients with genetic disorders' in 2nd International Conference on Translational & Personalized Medicine.

Some Genetic Disorders Discussed

Huntington's disease (Autosomal dominant inherited disease [26-33]**)**

Huntington's disease (HD) is earlier called as Huntington's chorea. It is a rare inherited neurological disorder with an occurrence of upto to 8 people per 100,000. It's occurrence ranges from 1 in 20,000 people of Western European origin to 1 in one million in people of Asian and African origin. It is an autosomal dominant inherited disease. It is a result of trinucleotide repeat expansion in a gene called Huntingtin (Htt) gene. This expansion results into production of an altered form of the Htt protein, this mutant Huntingtin (mHtt), leads to neuronal cell death in select regions of the brain and is a terminal illness. Major symptoms of this disease are abnormal body movements which is also called chorea and a lack of coordination, in many cases it also affects a number of mental abilities and personality aspects. A lot of research is on the go

for treating this disease. Blair R. Leavitt has published an interesting paper entitled "Feasibility and Safety of an Aquatherapy Program in Mid- to Late-Stage Huntington Disease" where he discussed about the safety and feasibility of aquatherapy for HD.

Cystic fibrosis (Autosomal recessive inherited disease [34-39])

Cystic fibrosis (CF), is otherwise called mucoviscidosis, is a autosomal recessive hereditary disease which can affect the entire body, leading to progressive disability and early death. It is one of the most common life-shortening, childhood-onset inherited diseases with an occurrence of about 1 in every 3900 children in the United States. It is most common among Ashkenazi Jews and Europeans; one among every twenty-two Europeans carry one gene for CF. Individuals with cystic fibrosis can be diagnosed before birth by genetic testing CF is a result a mutation in a gene called the cystic fibrosis transmembrane conductance regulator (CFTR). This gene helps to regulate sweat, digestive juices, and mucus. Only one CFTR gene is required to prevent cystic fibrosis. CF develops when both the genes doesn't work normally. Hence, CF is considered an autosomal recessive disease. Elias Matouk et al. described about C-Reactive Protein in Stable Cystic Fibrosis which can act as an indicator of Clinical Disease Activity and Risk of Future Pulmonary Exacerbations

Aicardi syndrome (X-linked dominant inherited disease [40-42])

Aicardi syndrome is a rare congenital disorder which is the result of an abnormality of the X chromosome and is characterized by (partial or complete) agenesis of the corpus callosum, seizures (often infantile spasms) and retinalabnormalities.

Colour blindness (X-linked recessive inherited disease)

Colour blindness also known as colour vision deficiency or Dyschromatopsia, is a condition where one cannot differentiate between some or all colours that others can distinguish. It is mostly of genetic origin, but in cases may also occur due to eye, nerve, or brain damage.

Leber's hereditary optic neuropathy (Mitochondrial inheritance disease [43,44])

Leber's hereditary optic neuropathy (LHON) is also called as Leber optic atrophy. It is a mitochondrially inherited disease which is characterised by degeneration of retinal ganglion cells (RGCs) and their axons which results in acute or subacute loss of central vision. It is inherited from mother to offspring as it is primarily due to mutations in the mitochondrial genome and only the egg contributes mitochondria to the embryo. It is resulted from one of three pathogenic mitochondrial DNA (mtDNA) point mutations.

Gout (Multifactorial and polygenic disorder [45-54])

Gout is a genetic/acquired disorder of uric acid metabolism which results into hyperuricemia and consequent acute and chronic arthritis. The recurrent attacks of acute arthritis are due to the precipitation of monosodium urate crystals in the joints leading to inflammation. Enzyme defects leading to overproduction of uric acid like partial deficiency of hypoxanthine guanine phosphoribosyl transferase (HGPRT) enzyme (Lack of genes to produce this enzyme) or high dietary intake of purines as in pulses results to high levels of uric acid. Hence gout has both a genetic and environmental predisposition. Aurea Lima has presented a keynote on Moving towards personalized medicine in rheumatoid arthritis: Methotrexate cellular pathways as pharmacogenetic predictors of Methotrexate therapeutic outcome in 5th International Conference on Predictive, Preventive and Personalized Medicine & Molecular Diagnostics .

Ehlers-Danlos Syndrome (Disorders with variable modes of transmission [55-60])

Ehlers-Danlos Syndrome is characterized by defects in collagen synthesis and structure which is a result of deficiency of lysyl hydroxylase enzyme or mutation in the coding genes resulting in deficient synthesis of type 3 collagen or mutation in

the type 1 collagen gene leading to deficient conversion of procollagen type 1 to collagen due to. These abnormal collagen fibres exhibit inadequate tensile strength and results in hyper extensible skin and the joints are hypermobile.

Fragile X chromosome (Cytogenetic disorders [61-71])

Fragile X chromosome disorder is characterized by mental retardation which is resulted by cytogenetic alteration of the x chromosome.

Cytogenetic disorders are characterised by either alteration in the structure or number of the chromosomes.

Conclusion

Genetic diseases can be well controlled through an integrative approach of population screening, genetic counseling, community education, carrier identification and neonatal screening and systematic treatment. A lot of information is shared trough open access journals which can be accessed and used by various professionals like physicians, geneticists and genetic counselors for controlling and treating the genetic diseases [72-87].

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