

## A Review on Genetic Disorders and Syndromes

Srilatha B\*

Department of Biotechnology, Presidency College, Bangalore University, Bangalore

\***Corresponding author:** Srilatha B, Department of Biotechnology, Presidency College, Bangalore University, Bangalore, E-mail: srilatha.biotech09@gmail.com

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### Abstract

Genetics is the study of heredity and heredity is a biological process where a gene from a parent passes into their children or offspring. Every off spring inherits genes from both of their biological parents and these genes in turn express specific traits of diseases which are hereditary from parents to offspring's.

A genetic syndrome is a genetic problem caused by one or more abnormalities in the genome; especially the condition arises from birth. Sometimes the genetic disorders are quite rare and affect one person in every thousands or millions. Most of the genes are the building blocks of heredity. They are mainly passed from parent to offspring's. They are the carriers of DNA and proteins. Proteins do most of the work in cells. The main function of the proteins they move molecules from one cell to another.

**Keywords:** Genetics; Genetic Syndrome; Genetic Disorders

### Introduction

Sometimes Genetic disorders may be hereditary, passed down from the parents' genes to off spring genes. In Some genetic disorders, defects may be caused by new mutations or changes in the Chromosomal or DNA. In some cases or in some forms of cancer, it may be caused by an inherited genetic condition in some people, by new mutations in other people, and mainly by environmental causes in some people. Whether, when and to what extent a person with the genetic disorder or syndrome will actually suffer from the disease and is almost always affected by the some external factors and events in the person's development [1-10].

In a mutation there will be change in a gene or genes modeling. The mutation changes the gene's structure for making a protein, so the protein does not work properly and it may leads to a medical condition called a genetic disorder or Genetic Syndrome [11-15].

People should be educated and obtain knowledge on various functions of the genes and these diseases so that the right preventive measures can be taken during their occurrence. People can gain awareness through literature, internet sources, family physicians and consultants. Open access journals provide more visibility and accessibility to the readers in gaining the

required information. The on-going researches all over the world, which are being exhibited through open access journals, serve as the main source of information in various fields [16-20].

In order to create awareness among the people, group of physicians and professionals unite to form a society or an organization. The main aim of these societies or organizations is to counsel or spread awareness among the victims of genetic disorders as well as healthy professionals, geneticist, and scholar researchers. Major societies like Genetics Society of Vietnam aims to improve public awareness in genetic related disorders like Down's syndrome. Down's syndrome is a genetic disorder caused when abnormal cell division takes place results in extra genetic material from chromosome 21. Genetic Society of America is being organized by OMICS Group International. OMICS Group International is an amalgamation of Open Access publications and worldwide international science conferences and the scientific events conducted in the field of Genetics. American Society of Human Genetics is the leading society of America associated with the OMICS International the Society's nearly 8,000 members include researchers, academicians, clinicians, laboratory practice professionals, genetic counsellors, nurses and others who have a special interest in the field of human genetics. Association of Biotechnology and Pharmacy will be useful to form a forum for scientists so that they can bring together to discuss and find scientific solutions to the problems of society. The annual meetings will help the members to share their knowledge and publish their research knowledge particularly by members and fellows of the Association and special care will be taken to provide an opportunity for young scientists. Autoimmunity network of Switzerland associated with the OMICS is a community of immunologists, physicians, rheumatologists & researchers who share and exchange the knowledge about autoimmune diseases and relevant areas. Members of the autoimmunity network have the opportunity to develop a network with colleagues, attend well established and successful annual congresses, and take advantage of the added benefit of being a part of a vibrant online social network [20-40].

Open Access literature plays a key role in proving the information and current researches across the globe. Journal of Genetic Disorders & Genetic Reports provides information on latest technologies related to Genetic Disorders & Genetic Reports and also many conferences like World Congress on Human Genetics organised by OMICS the main theme of the conference is Genomic Revolution: A debate on Human Genetic Disorders & Diseases [40-45].

Journal of Neurological Disorders Journal studies improve the knowledge and provide cutting-edge research strategies for the development of new therapeutics. Human Genetics & Embryology is a leading provider of information on genetics and embryology and novel methods of treatment followed. The above mentioned Open access journals on Genetics are the peer-reviewed journals that maintain the quality and standard of the journal content, reviewer's agreement and respective editor's acceptance in order to publish an article. These journals ensures the barrier-free distribution of its content through online open access and thus helps in improving the citations for authors and attaining good journal impact factors [45-49].

### **Genetic Disorders or Syndromes**

The most common Genetic syndrome or disorders include Cystic Fibrosis, Down syndrome, Severe Combined Immunodeficiency Disorder (SCID), Tay-Sachs, Jackson-Weiss Syndrome, Fragile X, Huntington's disease, Duchenne muscular dystrophy, Sickle-cell disease, Turner syndrome etc [50-61].

Down syndrome is a genetic disorder caused by the presence of all or part of a third copy of chromosome. It is typically associated with physical growth delays, characteristic facial features, and mild to moderate intellectual disability. In every

cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Typically, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. An article Evaluation and Efficient DNA Extraction Protocol for Single Nucleotide Polymorphisms Genotyping in Down Syndrome written by, Marcia R Amorim explains Down syndrome (DS) is the most frequent genetic form of intellectual disability. Nondisjunction is the leading cause of DS but molecular events underlying this mechanism remain unknown. The identification of genetic alterations that may influence DS etiology is essential. Single nucleotide polymorphisms (SNPs) in folate metabolizing genes as maternal risk factors for DS have been studied in populations throughout the world. Cystic fibrosis (CF) is the most common genetic disorder among Caucasians, with over 10 million people – or one out of every 31 Americans - carrying the defective gene. This autosomal recessive disease, caused by a mutation on chromosome 7 that leads to an alteration in the function of the cystic fibrosis trans membrane conductance regulator (CFTR) protein, affects 30,000 people in the United States, with half of this population being age 18 or older. Ladores S clearly explained Fertility and Reproductive Health Implications of Targeted Therapeutics for Cystic Fibrosis. In stable adult cystic fibrosis (CF) patients, we assessed the role of baseline high sensitivity C-reactive protein (hs-CRP) on CF clinical variables and frequency of intravenous (IV) treated pulmonary exacerbations (PEs) 1-year post- baseline. Elias Matouk clearly explained the C - reactive protein in Stable Cystic Fibrosis: An Additional Indicator of Clinical Disease Activity and Risk of Future Pulmonary Exacerbations [62-80].

Turner syndrome is a chromosomal disorder characterized by the presence of a single normal X chromosome in women. Additionally to the X chromosome monosomy, other cell lines can co-exist, containing the Y chromosome or part of it. The presence of Y chromosome in patients with Turner syndrome represents an increased risk (15- 30%) of developing gonadoblastoma. In a manuscript entitled Y-Chromosome Detection in Turner Syndrome authored Susana Fernandes MSW of USA provides Turner Syndrome (TS) with karyotype 45, X is one of the most common cytogenetic abnormalities (1:2500 among newborn females). It is compatible with life, even though the great majority (more that 99%) of the conceptuses with karyotype 45, X are a spontaneously loss, usually before 28 weeks of gestation. 2nd World Congress on Human Genetics & Genetic Disorders held in 2017 in USA, the mail theme of the conference is Human Genetics & Genetic Disorders [81-90].

Apart from the articles, presentation at conferences, symposiums, workshops also yield a better exposure to health information and advanced technologies that are being invented in the present generation. In 11th International Conference on Pediatric Pathology & Diagnosis going to held in March 2017 in UK, where the renowned speakers are going to discuss on the Pediatric Pathology & Diagnosis.

### **Novel Technologies in Genetics**

As the Genetic disorders and diseases have become more prevalent, there are many scientific professionals are trained especially in detecting, treating and counseling the prevention of genetic syndromes and diseases. Kiley J Johnson is a scientific professional with research expertize in Genomic testing. Kiley J Johnson has written an article The Genomic Novel and Priority Mapping Tool: Using Empathic Design to Develop Innovative Patient-Centered Decision-Making Tools for the Genomic Testing Experience which clearly explains about the Genomic testing is now being conducted in clinical practice.

There is a deficit of patient-oriented tools for learning about and making decisions regarding results from whole exome/whole genome sequencing (WES/WGS). Empathic, novel, and multi-disciplinary design approaches to creating new products or services were utilized to develop our materials. Interventions to promote articulation of personal priorities and hopes related to WES/WGS included a: (1) Patient Results Priority Mapping Tool; and (2) Genomic Novel, which included a worksheet called Spectrum of Influences.

Fatima Zohra Sediki clearly explained about the Spectrum of CFTR Mutations in the Algerian Population: Molecular and Computational Analysis little has been reported on the occurrence of cystic fibrosis in Algerian population. In order to contribute to the few existing data we undertook this study. The aim was in first instance to detect genetics alteration in the CFTR gene of 21 CF Algerian patients by sequencing. 14 different mutations were detected one of them has never been described. Among these mutations the c.680T>G (L227R) which seems to be specific to the Algerian population, it was in silico studied to determine its impact at a molecular level. This is the first study that combined a molecular and computational analysis. These findings will assist in genetic counseling, prenatal diagnosis and future screening of CF in Algeria [91-100].

## **Conclusion**

Genetically disorders are most prominent and prevalent in many highly developed countries. Many innovative technologies have been developed to reduce the mortality due to genetically Problems. Genetically Disorders caused by one or more abnormalities in the genome; especially the condition arises during birth itself. The goal of this session is to understand the Causes, Origin, Genesis and Source of various types of genetically disorders. All this information can be accessed in open access health care literature which exhibits the novel techniques and innovative researches taking place in the research field. Many professionals share their views; suggestions through the open access literature which can be accessed by all in order attain knowledge on brain diseases. Although neurological disease is the major problem in USA, the developed technologies, awareness through the literature have given hope to the patients for reducing the mortality rate.

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