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Short Communication

Perception regarding Chromosomal Disorders

Yujing Li

Department of Human Genetic, Emory University School of Medicine, USA

INTRODUCTION

Chromosomal disorder, any syndrome characterized by malformations or malfunctions in any of the body's systems, and caused by abnormal chromosome number or constitution. An abnormal condition due to something unusual in an individual's chromosomes [1]. For example, Down syndrome is a chromosome disorder caused by the presence of an extra copy of chromosome 21, and Turner syndrome is most often due to the presence of only a single sex chromosome: one X chromosome. A chromosomal disorder, chromosomal anomaly, chromosomal aberration, or chromosomal mutation is a missing, extra, or irregular portion of chromosomal DNA. It can be from a typical number of chromosomes or a structural abnormality in one or more chromosomes. Chromosome mutation was formerly used in a strict sense to mean a change in a chromosomal segment, involving more than one gene. The term "karyotype" refers to the full set of chromosomes from an individual; this can be compared to a "normal" karyotype for the species via genetic testing. A chromosome anomaly may be detected or confirmed in this manner. Chromosome anomalies usually occur when there is an error in cell division following meiosis or mitosis [2]. There are many types of chromosome anomalies. They can be organized into two basic groups, numerical and structural anomalies. Some chromosomal abnormalities occur when there is an extra chromosome, while others occur when a section of a chromosome is deleted or duplicated. Examples of chromosomal abnormalities include Down syndrome, Trisomy 18, Trisomy 13, Klinefelter syndrome, XYY syndrome, Turner syndrome and triple X syndrome [3].

STRUCTURAL ABNORMALITIES

When the chromosome's structure is altered, this can take several forms

Deletions: A portion of the chromosome is missing or deleted. Known disorders in humans include Wolf-Hirschhorn syndrome, which is caused by partial deletion of the short arm of chromosome 4; and Jacobsen syndrome, also called the terminal 11q deletion disorder.

Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material. Known human disorders include Charcot-Marie-Tooth disease type 1A, which may be caused by duplication of the gene encoding peripheral myelin protein 22 (PMP22) on chromosome 17.

Translocations: A portion of one chromosome is transferred to another chromosome. There are two main types of translocations:

Reciprocal translocation: Segments from two different chromosomes have been exchanged.

Robertsonian translocation: An entire chromosome has attached to another at the centromere - in humans these only occur with chromosomes 13, 14, 15, 21, and 22.

Inversions: A portion of the chromosome has broken off, turned upside down, and reattached, therefore the genetic material is inverted.

Insertions: A portion of one chromosome has been deleted from its normal place and inserted into another chromosome.

Rings: A portion of a chromosome has broken off and formed a circle or ring. This can happen with or without loss of genetic material.

Isochromosome: Formed by the mirror image copy of a chromosome segment including the centromere.

Chromosome instability syndromes are a group of disorders characterized by chromosomal instability and breakage. They often lead to an increased tendency to develop certain types of malignancies.

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Correspondence to: Dr. Li Y, Department of Human Genetic, Emory University School of Medicine, USA; E-mail: yli@genetics.emory.edu

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