Summary and conclusion

The genetic disorders are divided into chromosomal disorders and gene disorders .

A) Chromosomal disorders are divided into two classes .

1 - Abnormalities of chromosomal number: These arise from non disjunctions that is from failure of two homologous chromosomes in the first division of meiosis or of two sister chromatids in mitosis or the second division of meiosis to pass to opposite poles of the cell .

2- Abnormalities of chromosome structure :

Result from chromosome breakage and reunion . When a chromosome breaks it can rejoin in its old form (restitutions) or it can rejoin with another broken chromosome (reunion) . Reunion leads to a structural rearrangement that can be balanced or unbalanced . If balanced , the amount of genetic material is identical to that of the normal cell . Types of balanced rearrangements include balanced reciprocal translocation , Robertsonian translocations and inversions . If the rearrangement is unbalanced this indicates loss or gian of chromosomal material **.**

B) Gene disorders are either mendalian monogenic or non mendalian disorders .

1 - Mendalian monogenic disorders are caused by single mutant gene whitch show on of four simple mendelian patterns of inheritance .

1- autosomal dominant 2- autosomal recessive

3- x-linked dominant 4- x-linked recessive

1 - Autosomal dominant triats :

Are fully manifested in the presence of a gene in the heterozygous state that is when only one abnormal gene is present .

2 - Autosomal recessive disorder :

Autosomal recessive conditions are clinically apparent only in the homozygous state i.e , when both alleles at a particular genetic locus are mutant alleles .

3 - X-linked inheritance :

Genes located on the x- chromosome are termed x-linked because the female has tow x chromosomes she may be either heterozygous or homozygous for the mutant gene and the trait may exhibit recessive or dominant expressions . The male has only one x chromosome and therefore is homozygous for x linked traits.

A- X-linked dominant traits :

This mode of inheritance is uncommon . Its characteristic features are as follow :

(1) females are affected about twice as often as males .

(2) heterozygous females transmit the trait to both genders with a frequency of 50% .

(3) homozygous affected males transmit the triat to all of their doughters and non of their sons .

B- X-linked recessive triats :

This mode of inheritance is relatively common . Its characteristic features are as follow :

(1) the disorder is fully expressed only in homozygous affected male .

(2) heterozygous females are normal occassionaly they may exhibit mild features of the disorder

11 - Non mendilian inheritance :

1 - Polygenic inheritance .

Most phenotypic traits are determined by the collaboration of many genes at different loci rather than by single gene . polygenic inheritace is suggested for traits that show continous variations in the form of a normal distribution curve **.** In multifactorial genetic disease there is both a polygenic component and an environmental component of causative factors .

2 - Nontraditional mode of inheritance :

A) Mitochondrial disease are a group of disorders relating to the mitochondria .

B) Trinucleotide repeat disorder are a set of genetic disorders caused by trinucleotide repeats in certain genes exceeding the normal, stable, threshold, which differs per gene.

C) X-inactivation is a process by which one of the two copies of the X chromosome present in female mammals is inactivated.

D) Uniparental disomy refers to the situation in which two copies of a chromosome come from the same parent, instead of one copy coming from the mother and one copy coming from the father.

E) Somatic mutation is a mutation that occurs in the somatic (i.e. non-reproductive) cells of an organism **.**

Genetic and chromosomal disorders may lead to diseases .

A) - Chromosomal disorders :

1) -Chromosome number abnormalities : Refer to Table (2)

1 – Down syndrome . 2 - Turner syndrome .

3 – Klineflter syndrome . 4 –Trisomy 13 .

5 – trisomy 18 .

2) - Chromosomal structure abnormalities : Refer to Table (3)

1 -Prader-willi Syndrome . 2 - Wolf-Hisrchhorn Syndrome.

3 - Digeorge Syndrome. 4 - Del, 18p.

5 - Aniridid syndrome .

B) – gene disorders :

1) – Monogenic mendelian disorders

A –Autosomal dominant diseases :Refer to table (4)

1) – MEN 2) – Pheochromocytoma

3) - Pseudohypoparathyriodism 4) - MODY

B) –Autosomal recessive diseases : Refer to table (5)

1) – Pendred syndrome 2) –Laurance Moon Biedle syndrome

3) –Growth hormone deficiency 4) – congenitalal adrenal hyperplasia

C)- Sex linked recessive diseases : Refer to table (6)

1) – Diabetes insipus 2) – Androgen insensitivity syndrome

3) –Kennedy syndrome 4) – Adrenoleukodystrophy

2) – Nonmendelian inheritance :

A) - Multifactorial inheritance :Refer to table (7)

1) – D.M 2) - PCOS

B) – Nontraditional mode of inheritance :

1) – Beckwith – Wiedemann syndrome .

2) – Partial paternal uniparental disomy of chromosome 6 in infant .

3) – Deafness with diabetes