Aneuploidy

Term refers to cytogenetic abnormalities in which all or part of one are more chromosomes are duplicated or deleted.

Autosomal aneuploidy: refers to all such abnormalities that do not involve the se chromosomes. These can be either numerical or structural.

Autosomal aneuploidy:

1- Autosomal Trisomy

Trisomy 21

The phenotype was delinated by John Down 1828 and referred to today as Down syndrome.

Down syndrome is more frequent in males ,

Phenotype:

1- Craniofacial appearance
2- Flat nasal bridge
3- Small mouth and thick lips
4- Small and overfloded ears.
5- Hands and feet are smaal
6- Cardiac anomalies.

Ghenotype

Prescencs of third copy of chromosome 21.

47, XX or XY , +21

Trisomy 18: first described by John Edwards in 1690.

The disease called Edwards syndrome.

This disease more frequent in females. In addition, caused by the prensence of third copy chromosome 18. Babies with disease are often born small and have heart defects. Named by Edwards in 1960.

Genotype 47, XX or XY, + 18

2- Autosomal monosomy:

Monosomy 21: phenotype

1- Infant have growth restriction
2- Facial dysmorphism
3- Redundant skin in the neck
4- Congential heart diseas
5- Genotype : 45, XX or XY , -21

Monosomy 22

1- Infant died after birth
2- Intracranial hemorrhage
3- More frequent in male.
4- Genotype : 45, XX or XY , -22

Polyploidy: A numerical chromosome abnormalities with changes in the number of complete sets of chromosomes.

Triploidy: chromosome number in triploidy (3n) 69 chromosomes

69 XXX or XXY

Most patients died shortly after birth.

Tetraploidy: chromosome number (4n) 92 chromosomes