

GENETIC DISORDERS

- Genomics - Study of Human Genome.
- Genome - Sum total of Genetic Information of an individual which is encoded in the Structure of DNA.

Human DNA consist of 3 Billion Base Pairs.

Founder Effect - describes distribution of diseases on the basis of genetics

CLASSIFICATION OF GENETIC DISORDERS.

- Chromosomal Disorders.

1. Relating to Sex Chromosomes.
- Klinefelter's Syndrome

Abnormal males having 2 or more X Chromosomes - XXY, XXXY.

Non - Functional Testis .

Spermatozoa are ABSENT in their ejaculations .

Scanty Hairs on Face, Axillae + Pubis .
Gynecomastia + Mental Retardation .
most common Cardiac Abnormality -
mitral Valve Prolapse

• ii. XYY Syndrome (Super male) male with an extra Y Chromosome .
Exceptional Height (6 feet or over) Personality disorders + Behavioral disturbances

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Anti - Social, Aggressive + Criminal Behavior .

iii. Turner's Syndrome

They have 45 chromosomes instead of 46 - XO instead of XX (O represents the missing Chromosome viz. due to Nondisjunction of sex chromosomes) .
most common Chromosomal Disorder in Humans .

Females with under - developed Sex Glands (Streak Ovaries) .

Infertile + Primary Amenorrhea (most common cause for primary amenorrhea)

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Short Stature + Webbed Neck .
Pulmonary Stenosis + Renal
malformation .

most common Cardiac Anomaly -
Bicuspid Aortic Valve .

most common cause of Death -
Coarctation of Aorta

• iv. Super Females . Females with 3 to
5 X Chromosomes - XXX, XXXX, XXXXX .
Underdeveloped External Genitalia,
Uterus + Retardation .

2. Relating to Autosomes . 2.
mongolism/Down's Syndrome Trisomy .
Short Stature + Small Round Head .
Narrow Tilted Eye - Slits + malformed

Ears .

Short Broad Hands + Lax Limbs .
mental Retardation .

Atresia of the Alimentary Tract . most
common Cardiac Defect - Endocardial
Cushion Defect . Frequency of mongolism
- increases with increasing maternal Age

• ~~del~~. Unifactorial Disorder (Single
Gene or mendelian Disease) .
mendelian Diseases are inherited
according to the mendelian Laws .

These are the Dominant, Recessive +
Sex - Linked Diseases .

1. Autosomal Diseases Autosomal
Dominant Traits ($m = f$) Autosomal
Recessive Traits ($m = f$)

Achondroplasia Albinism Huntington's
Chorea Phenyl Ketonuria
neurofibromatosis Alkaptonuria
Retinoblastoma Cystic Fibrosis
Hereditary Spherocytosis Galactosemia

• 2. Sex Linked Disease Sex Linked
Dominant Traits (100% Daughters, No
Sons) Sex Linked Recessive Traits (m
> f) Vitamin D Resistant Rickets
Hemophilia Type A + B Blood Group Xg
Duchenne muscular Dystrophy (DMD)
familial Hypophosphatemia Color
Blindness Alport Syndrome G6PD
Deficiency Hypophosphatemic Rickets
Fragile X

• ~~labeled~~. multifactorial Disorders •
essential Hypertension • Schizophrenia •
mental Retardation • Duodenal Ulcers •
Ischemic Heart Disease • Diabetes •
Familial Polypsis Coli • Familial Non -