Study of Human karyotypes, normal and abnormal (Down syndrome, Klinefelter syndrome, Turner syndrome, Cri-du-Chat syndrome, Chronic Myeloid Leukaemia Karyotypes) (from Photographs)

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Aim

To study normal human karyotypes (46,XX and 46,XY) and to identify selected chromosomal abnormalities (Down syndrome, Klinefelter syndrome, Turner syndrome, Cri-du-Chat syndrome and Chronic Myeloid Leukaemia) from photographs of karyograms.

Principle

A karyotype is the complete set of chromosomes of an individual arranged in homologous pairs according to size, position of centromere and banding pattern.

In humans, the normal somatic karyotype is diploid, with 46 chromosomes, comprising 22 pairs of autosomes and one pair of sex chromosomes (XX in females, XY in males).

Photographs of metaphase chromosomes are cut and arranged as karyograms, which allow detection of numerical and structural chromosomal abnormalities responsible for various genetic syndromes and some malignancies.

Materials Required

- Photographs or printed plates of: normal male (46,XY) and normal female (46,XX) human karyotypes.
- Photographs of abnormal human karyotypes:
    - Down syndrome (Trisomy 21)
    - Klinefelter syndrome (47,XXY and variants)
    - Turner syndrome (45,X)
    - Cri-du-Chat syndrome (5p deletion)
    - Chronic Myeloid Leukaemia (CML) with Philadelphia chromosome, t(9; 22).
- Human chromosome reference chart/ideogram
- Pencil, eraser, scale and notebook for recording observations and drawing diagrams

Procedure

A. Study of Normal Human Karyotypes (from photographs)

1. Observe the photograph of a normal human metaphase spread and its corresponding arranged karyogram.
2. Identify and count the total number of chromosomes; confirm that there are 46 chromosomes arranged in 23 pairs.

3. Locate autosomes (chromosome pairs 1 to 22) arranged in decreasing order of size and grouped according to centromere position.
4. Identify the 23rd pair of sex chromosomes and determine the genetic sex: XX for female and XY for male.
5. Note the diploid formula and write it in standard cytogenetic notation (e.g., 46, XX for normal female; 46, XY for normal male).

B. Study of Abnormal Human Karyotypes (from photographs)

For each provided abnormal karyotype:

1. Observe the overall chromosome number and arrangement.
2. Compare autosomes and sex chromosomes with the normal karyotype.
3. Identify the specific numerical or structural abnormality (extra chromosome, missing chromosome or segment, or translocation).
4. Write the standard karyotype notation and name of the syndrome/condition

Observations

Normal Human Karyotypes (Fig. 1)

Type	**Chromosome number and sex**	**Key observation**	**Inference**
Normal female	46, XX	22 pairs of autosomes and a pair of large X chromosomes.	Normal human female karyotype.
Normal male	46, XY	22 pairs of autosomes, one X and one smaller Y chromosome.	Normal human male karyotype.

Abnormal Human Karyotypes

Syndrome / Condition	**Standard karyotype notation (example)**	**Chromosomal change (from photograph)**	**Salient features (brief)**
Down syndrome (Fig. 2)	47,XX,+21 or 47,XY,+21	Extra chromosome 21; three copies of chromosome 21 are visible.	Intellectual disability, characteristic facies, hypotonia, increased

Syndrome / Condition	Standard karyotype notation (example)	Chromosomal change (from photograph)	Salient features (brief)
			risk of cardiac defects. microbenotes+4
Klinefelter syndrome (Fig. 3)	47,XXY (classical form)	Presence of two X chromosomes and one Y chromosome in a phenotypic male.	Tall stature, small testes, gynecomastia, infertility, usually male phenotype.
Turner syndrome (Fig. 4)	45,X	Only one X chromosome; absence of the second sex chromosome (no second X or Y).	Short stature, primary amenorrhoea, streak ovaries, webbed neck, female phenotype.
Cri-du-Chat syndrome (Fig. 5)	46,XX,del(5p) or 46,XY,del(5p)	Deletion of short arm of chromosome 5 (5p) seen as missing segment.	High-pitched cat-like cry in infancy, microcephaly, developmental delay.
Chronic Myeloid Leukaemia (Fig. 6)	46,XX,t(9;22)(q34;q11) or 46,XY,t(9;22)(q34;q11)	Presence of Philadelphia chromosome: shortened chromosome 22 due to reciprocal translocation between long arms of chromosomes 9 and 22.	Myeloproliferative disorder with leukocytosis, splenomegaly, presence of BCR-ABL fusion gene.

Discussion

Human karyotyping helps detect chromosomal abnormalities responsible for genetic diseases and cancers. Numerical abnormalities arise due to nondisjunction during meiosis, while structural abnormalities occur due to chromosomal breakage and rearrangement.

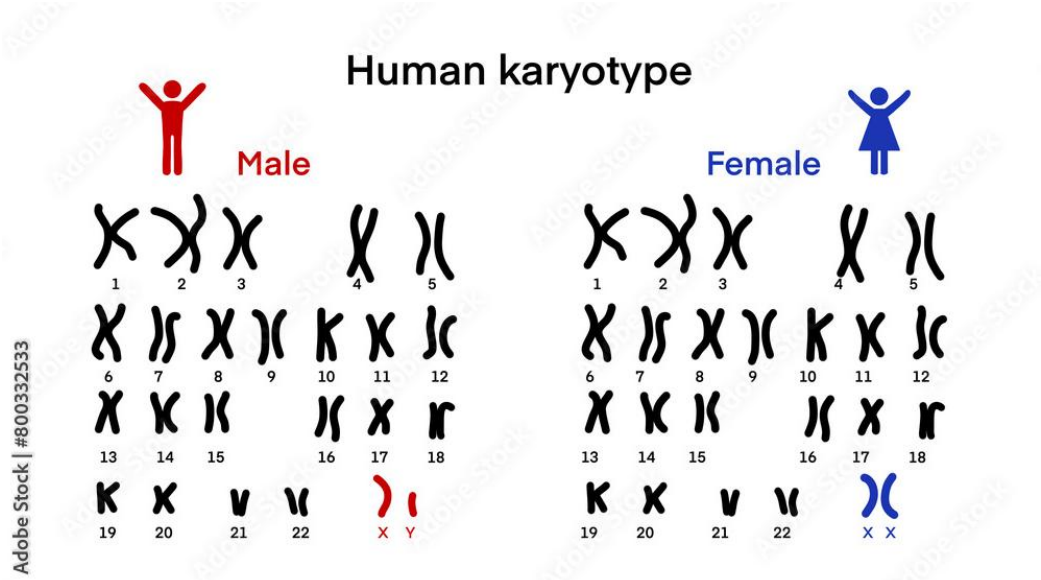

Figure 1: Normal human male and female karyotype.

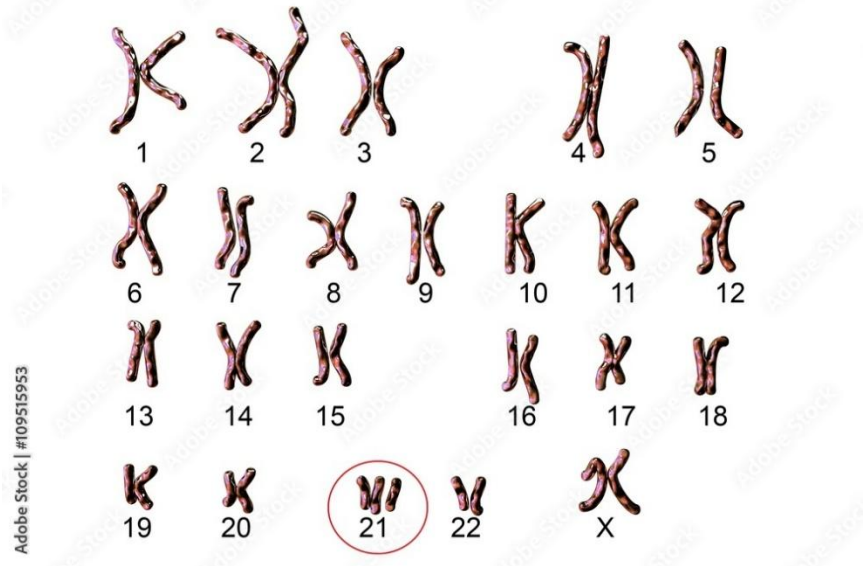

Figure 2: Karyotype of down syndrome (47, XX, +21)

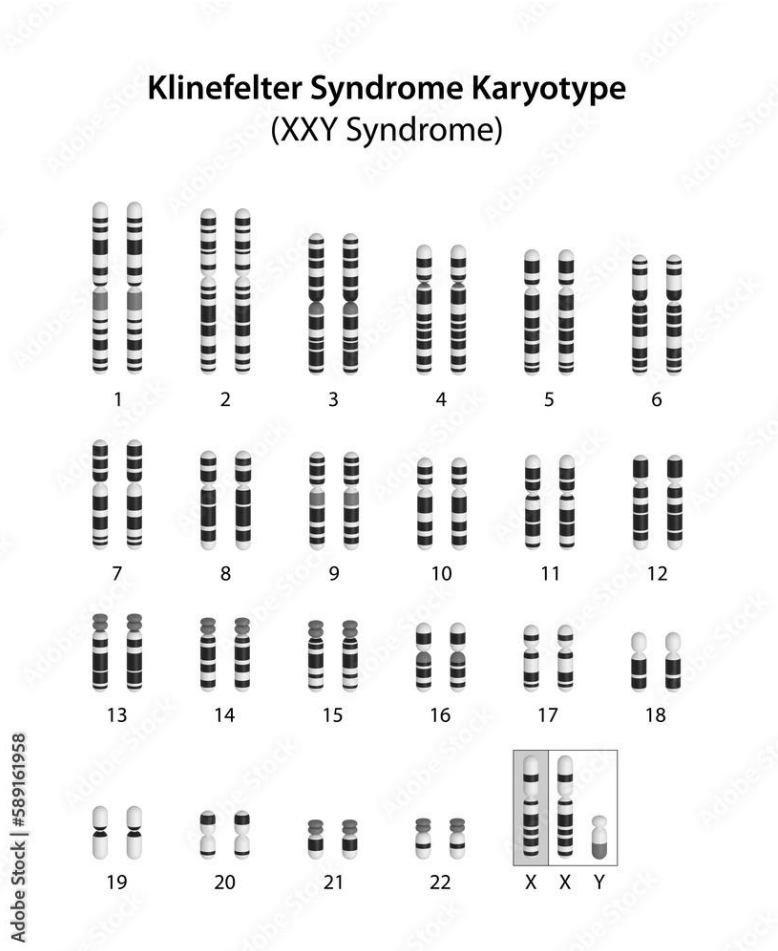

Figure 3: Karyotype of Klinefelter syndrome (47, XXY).

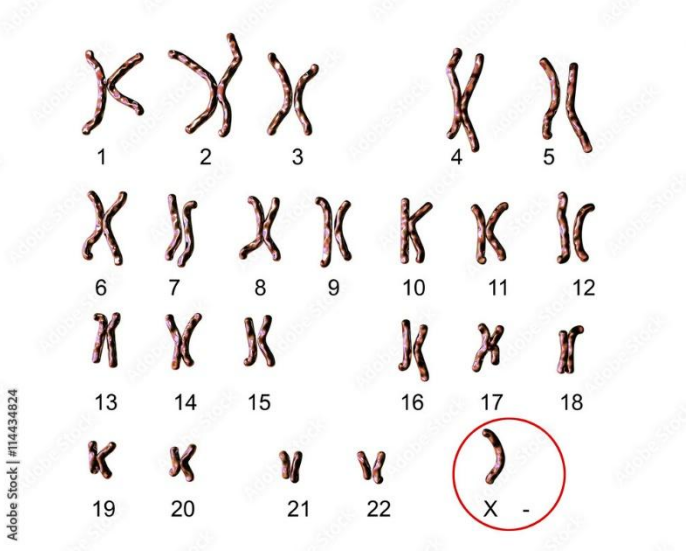

Figure 4: Karyotype of Turner syndrome (45, X)

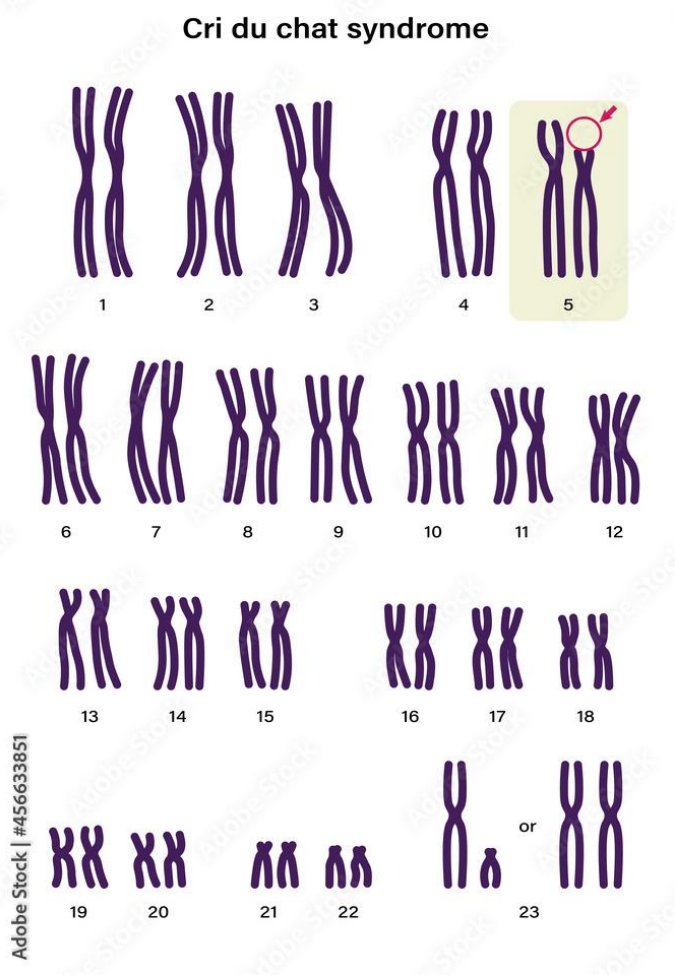

Figure 5: Karyotype of Cri du Chat syndrome [46, XX, del(5p)]

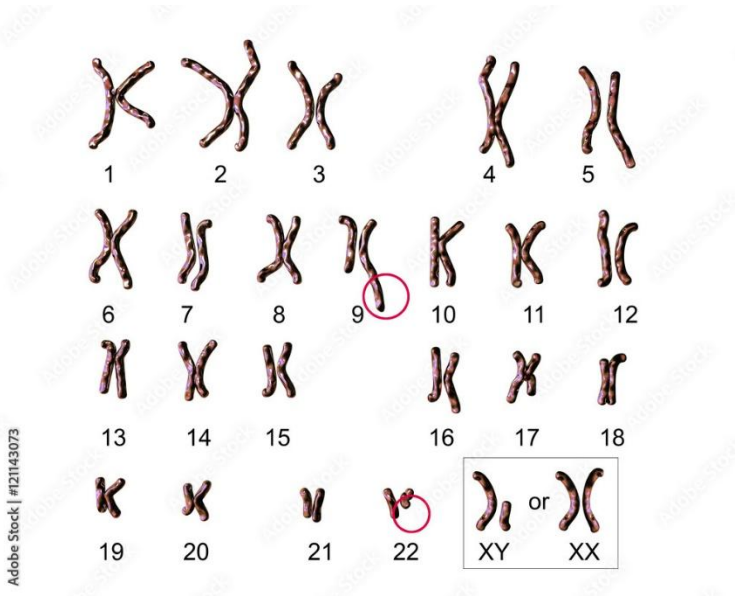

Figure 6: Karyotype of Chronic Myeloid Leukaemia [46,XX,t(9;22)(q34;q11) or 46,XY,t(9;22)(q34;q11)]