OUTLINE 18

VII. Mutation

E. Examples: Human genetic abnormalities

F. Chromosomal abnormalities

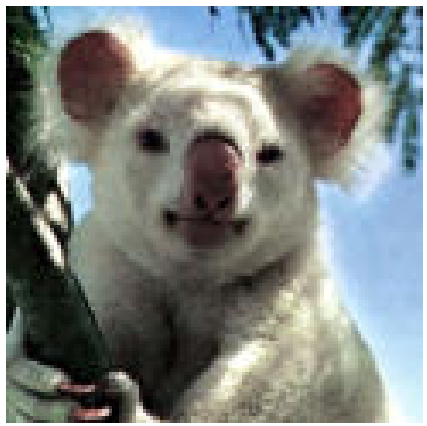
1. Chromosome breakage
2. Aneuploidy and non-disjunction

H. Characterization of Human Genetitc abnormalities

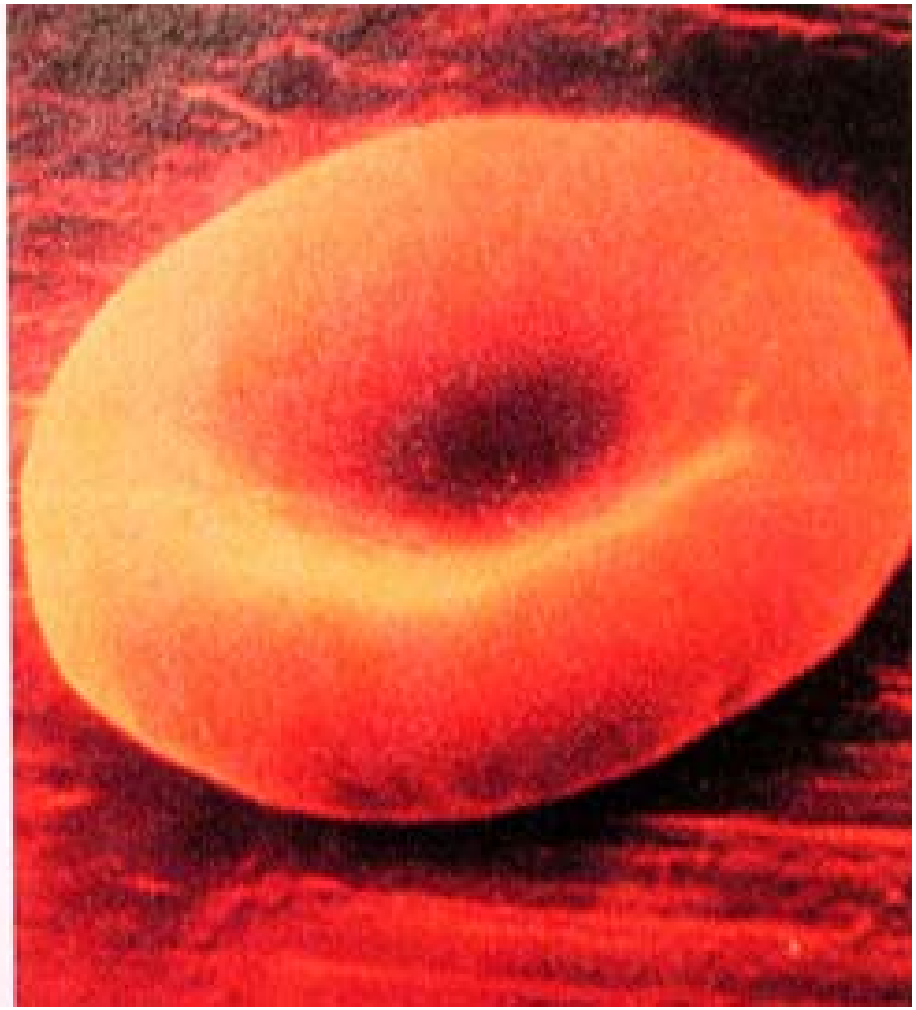
1. Pedigree analysis
2. Sex chromatin analysis
3. Karyotype analysis
4. Gene sequencing and marker genes


M
Erbgutverändernd

Albinism



Sickle cell



Normal red blood cell

Sickle cell anemia: an autosomal co-dominant

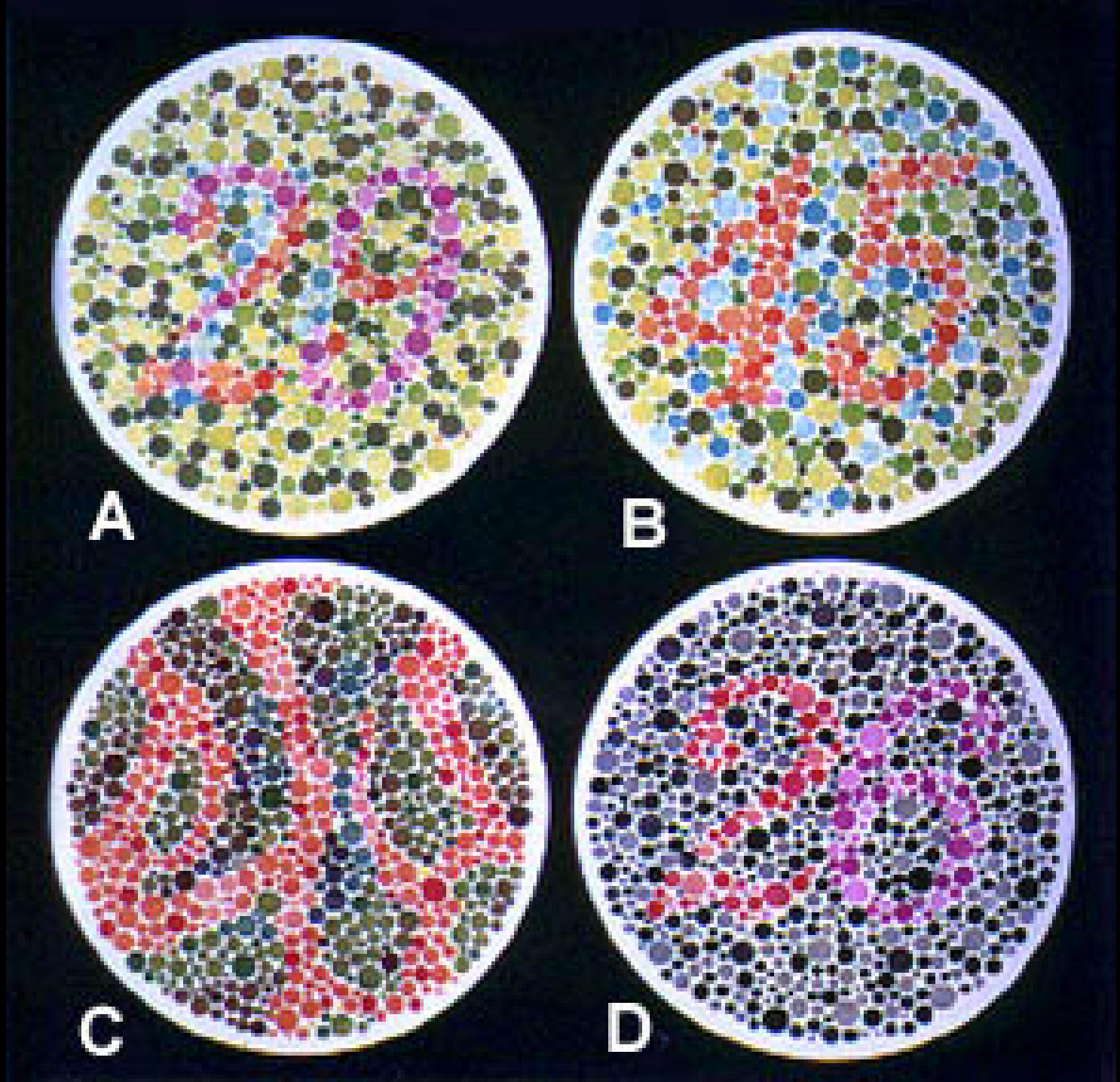
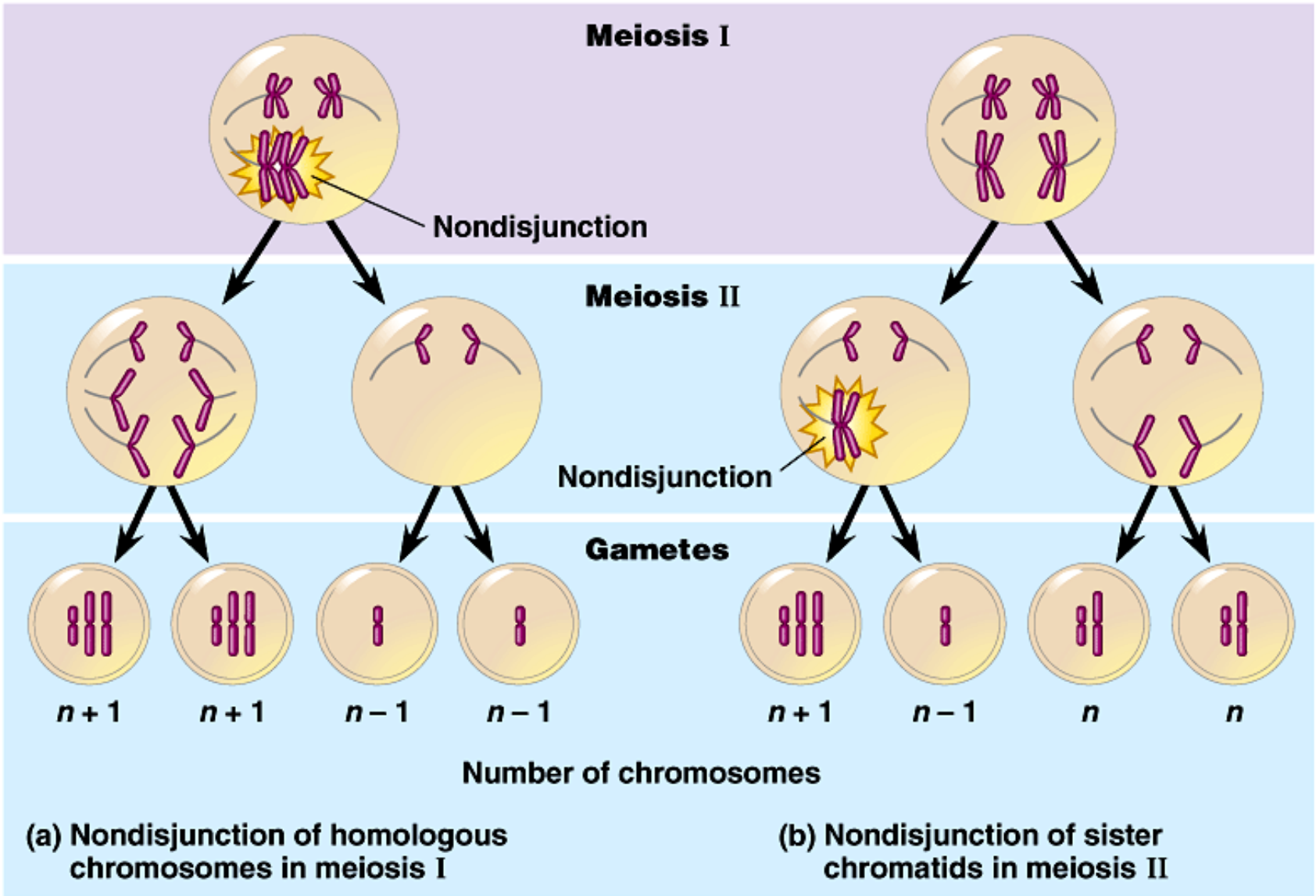
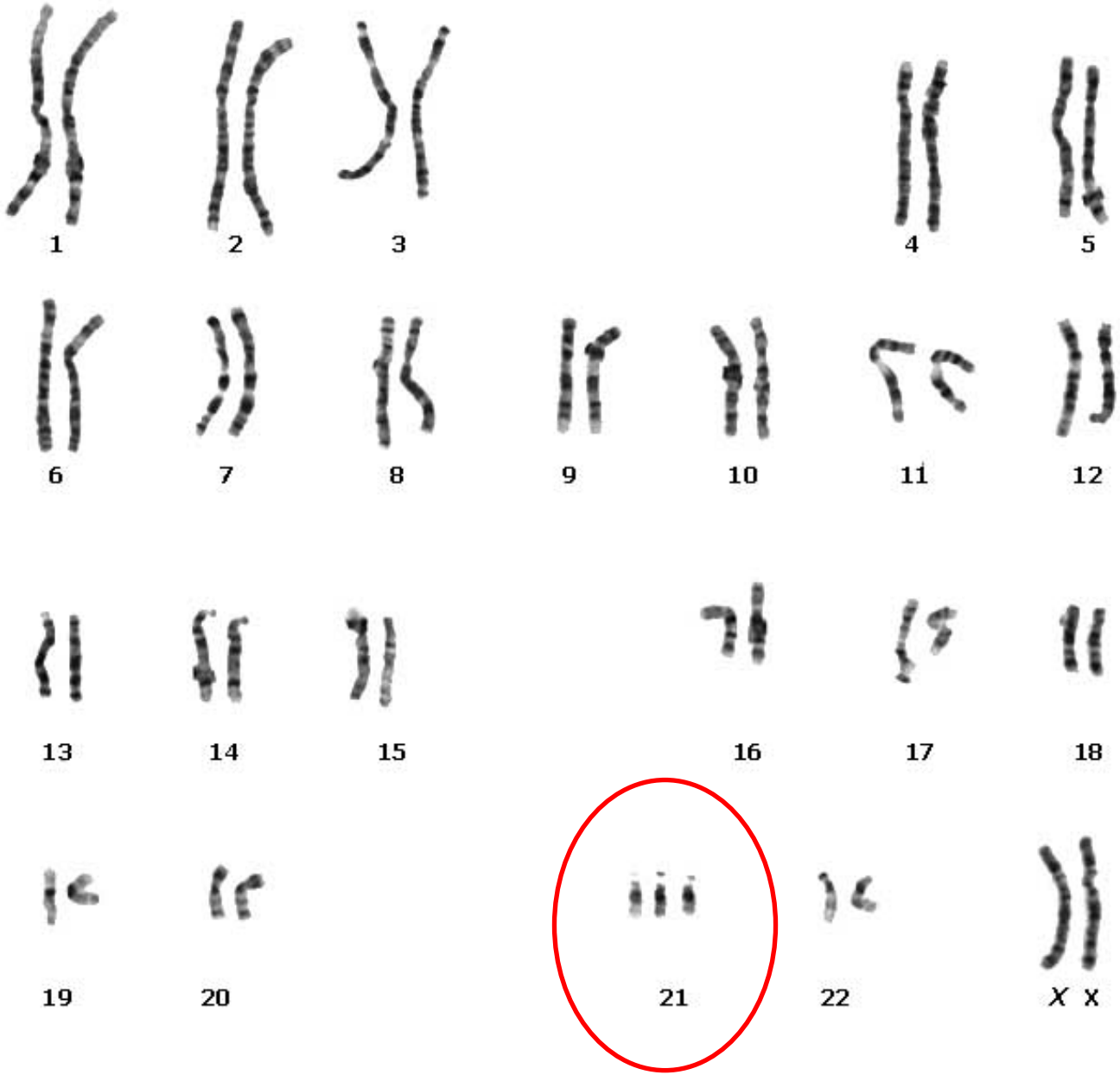
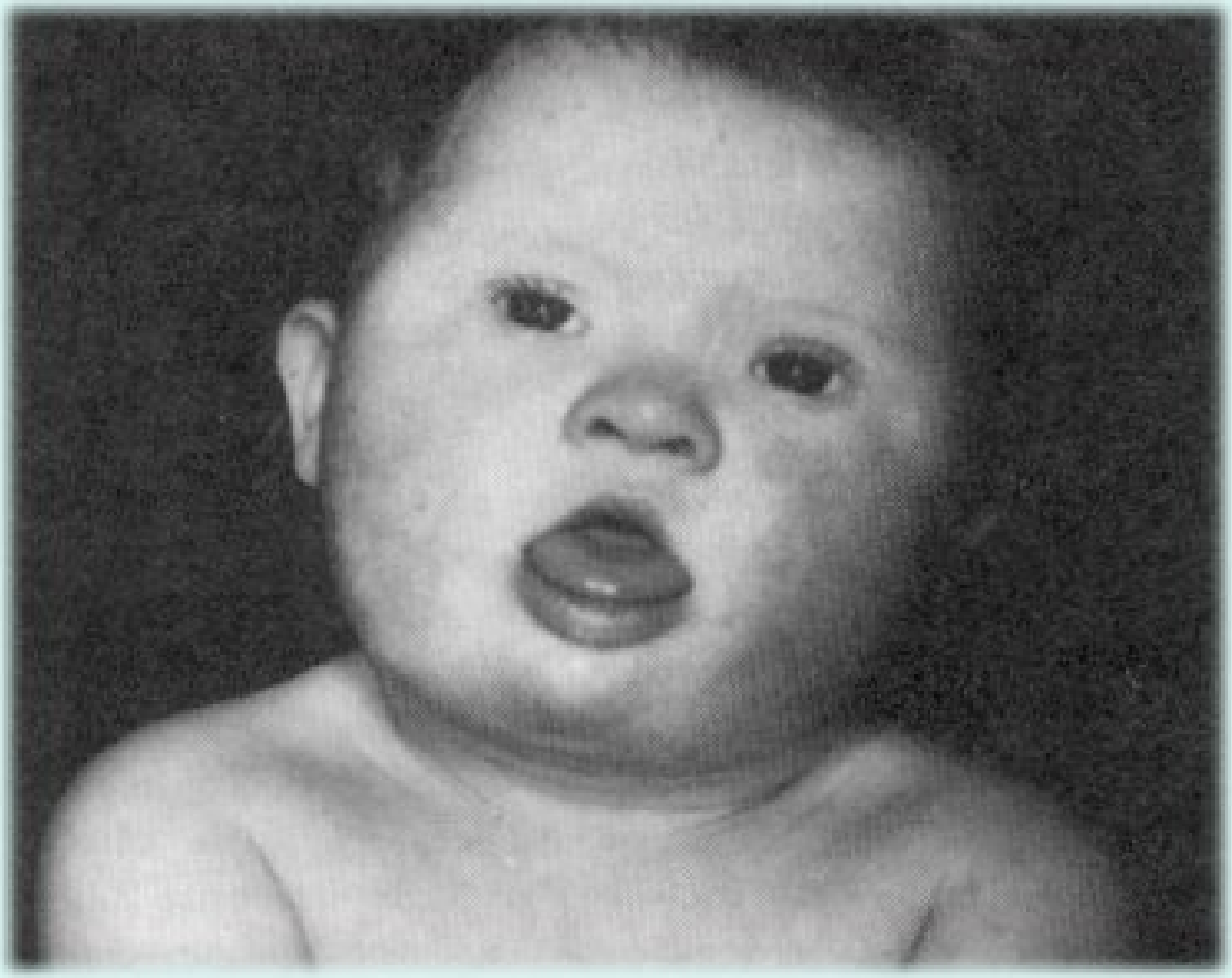

A
B
C
D

Fig. 15.11: Non-disjunction leading to aneuploid gametes

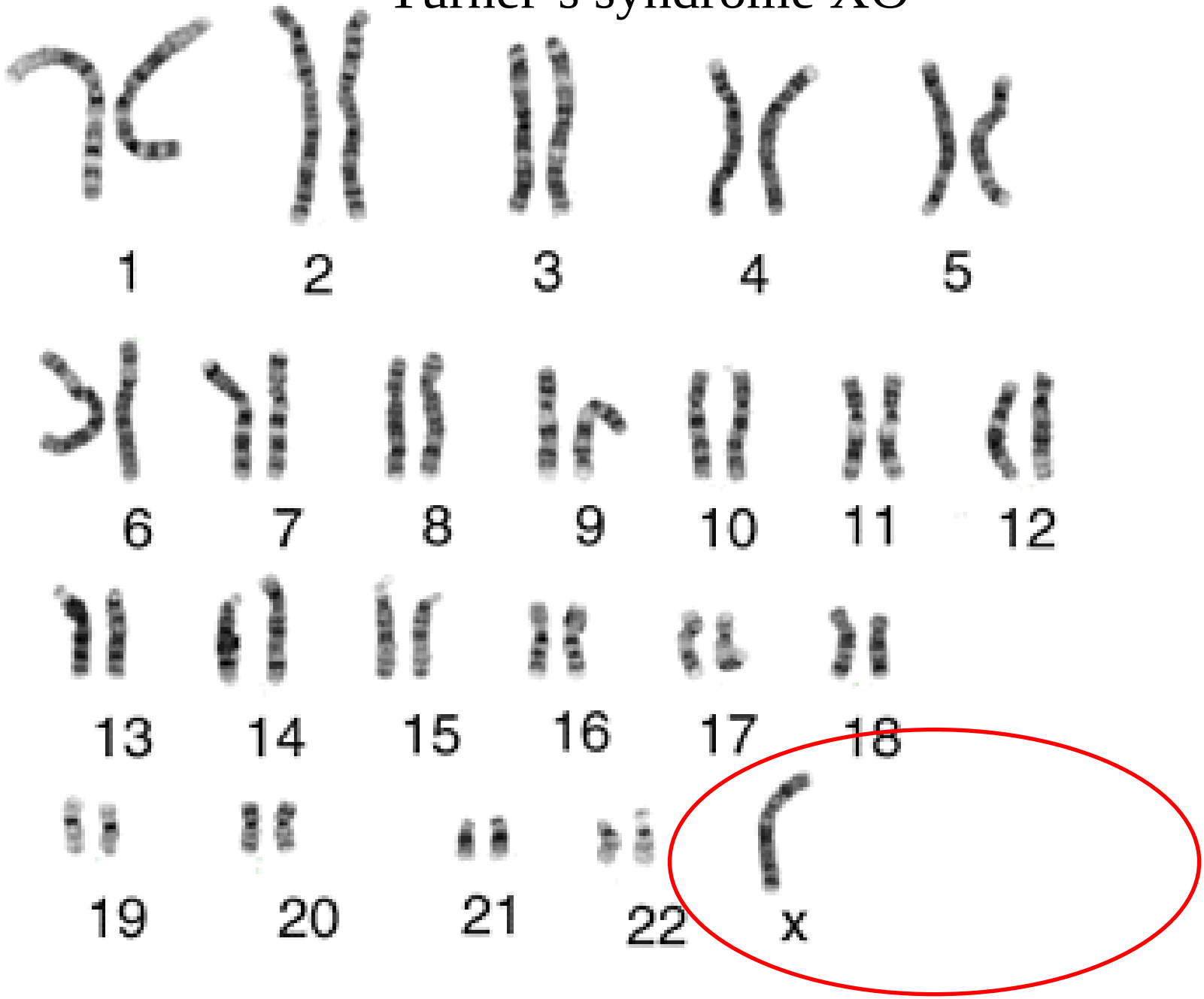


Trisomy 21

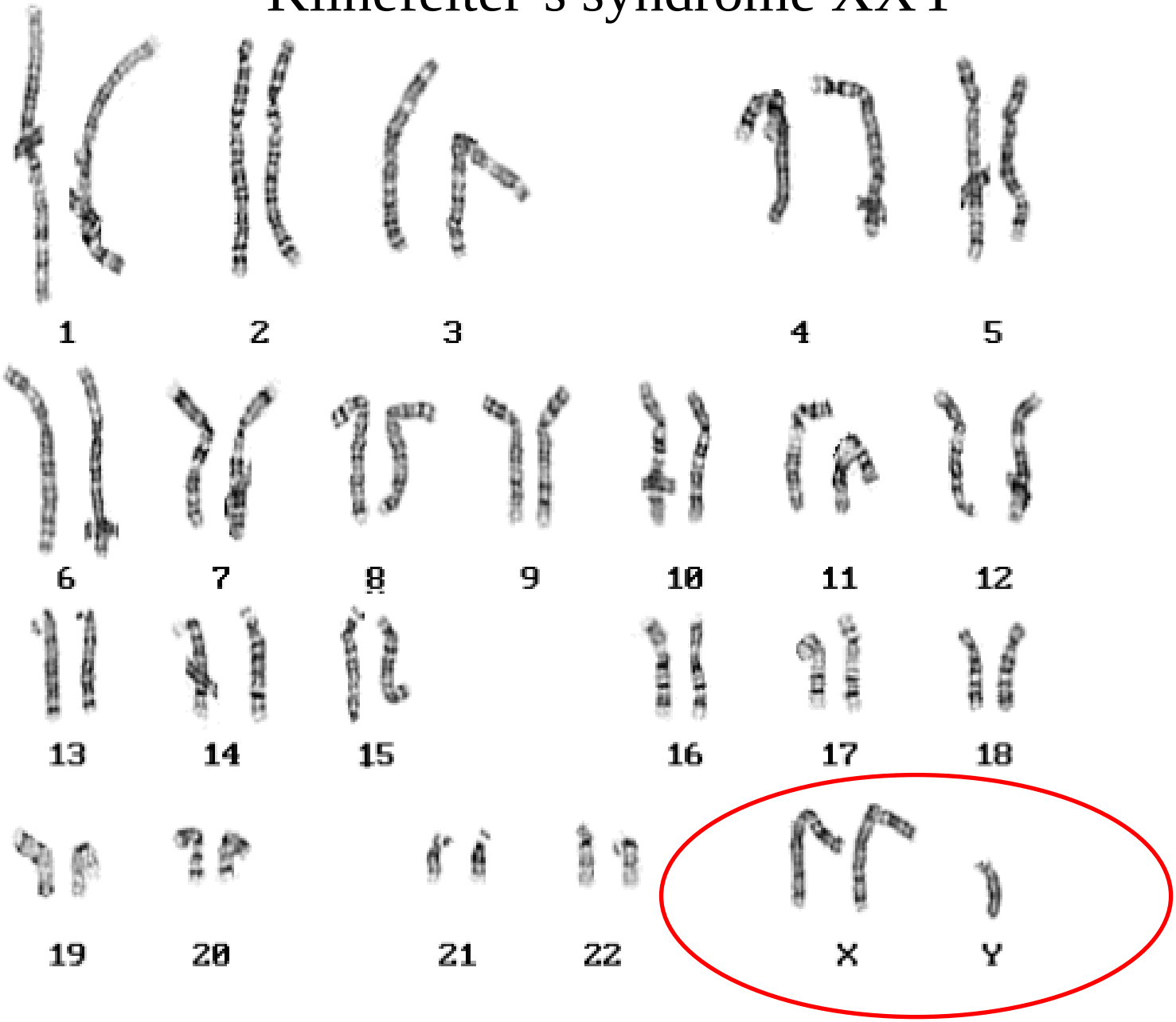




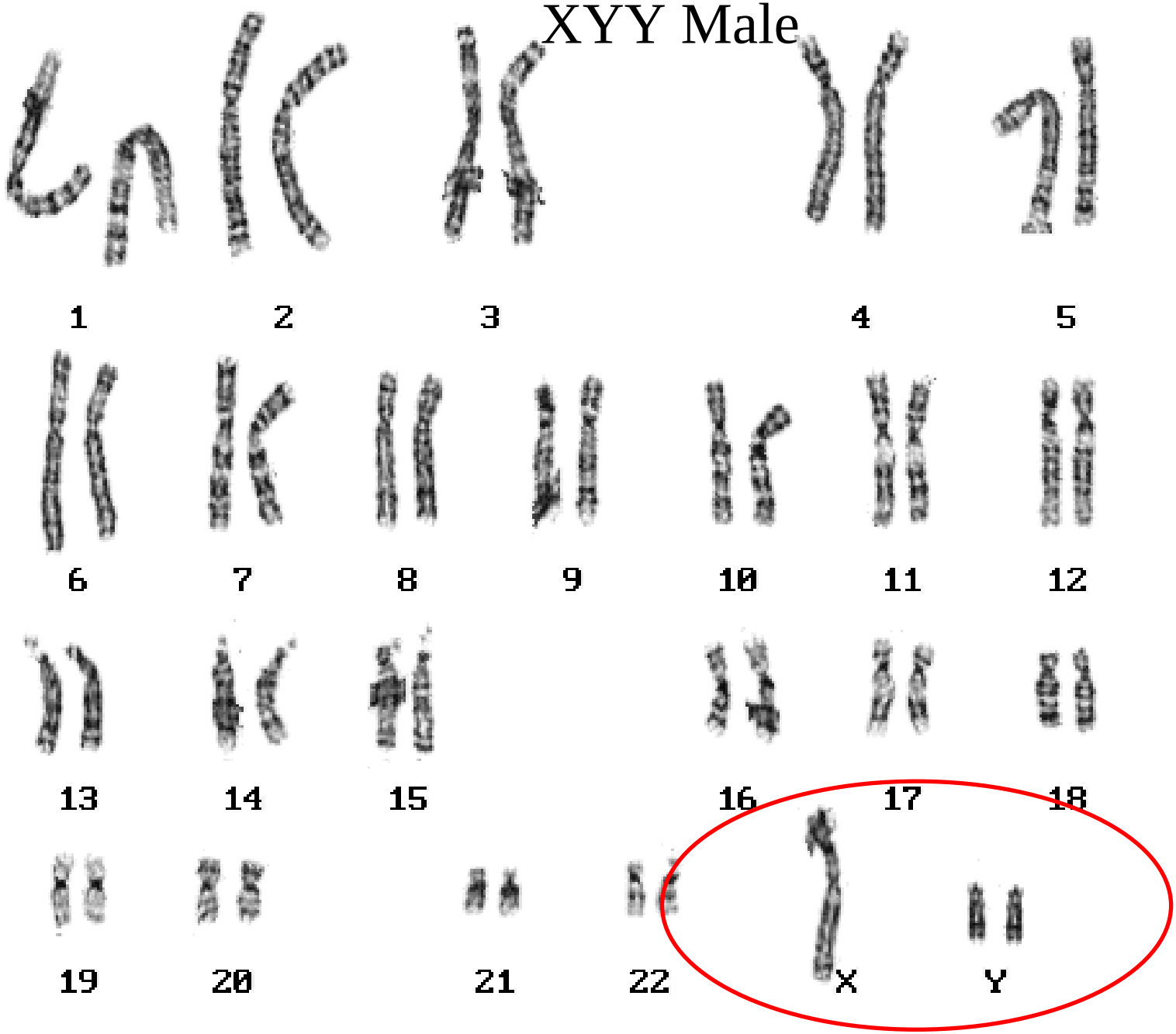
Turner's syndrome XO



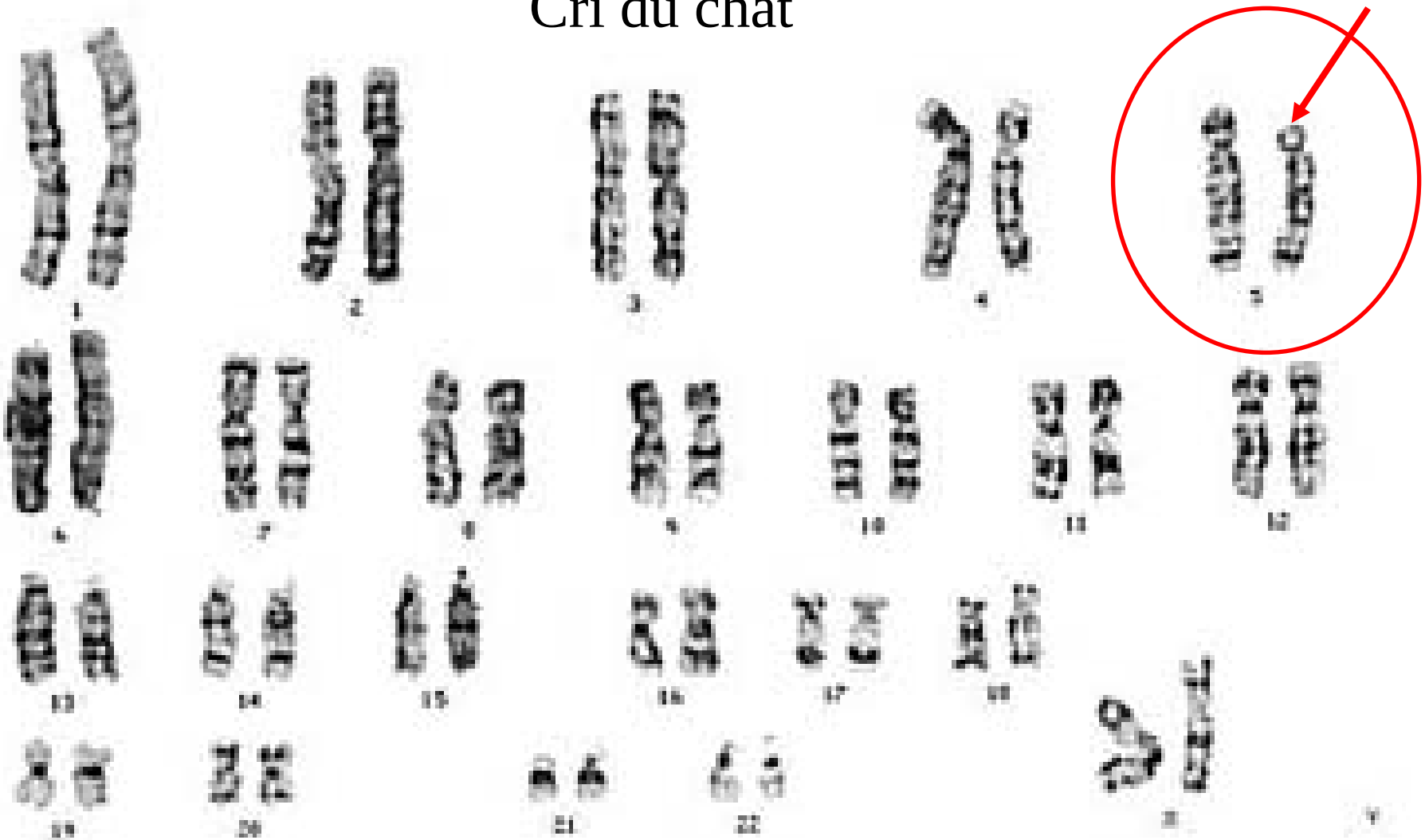
Klinefelter's syndrome XXY



XYY Male



Cri du chat



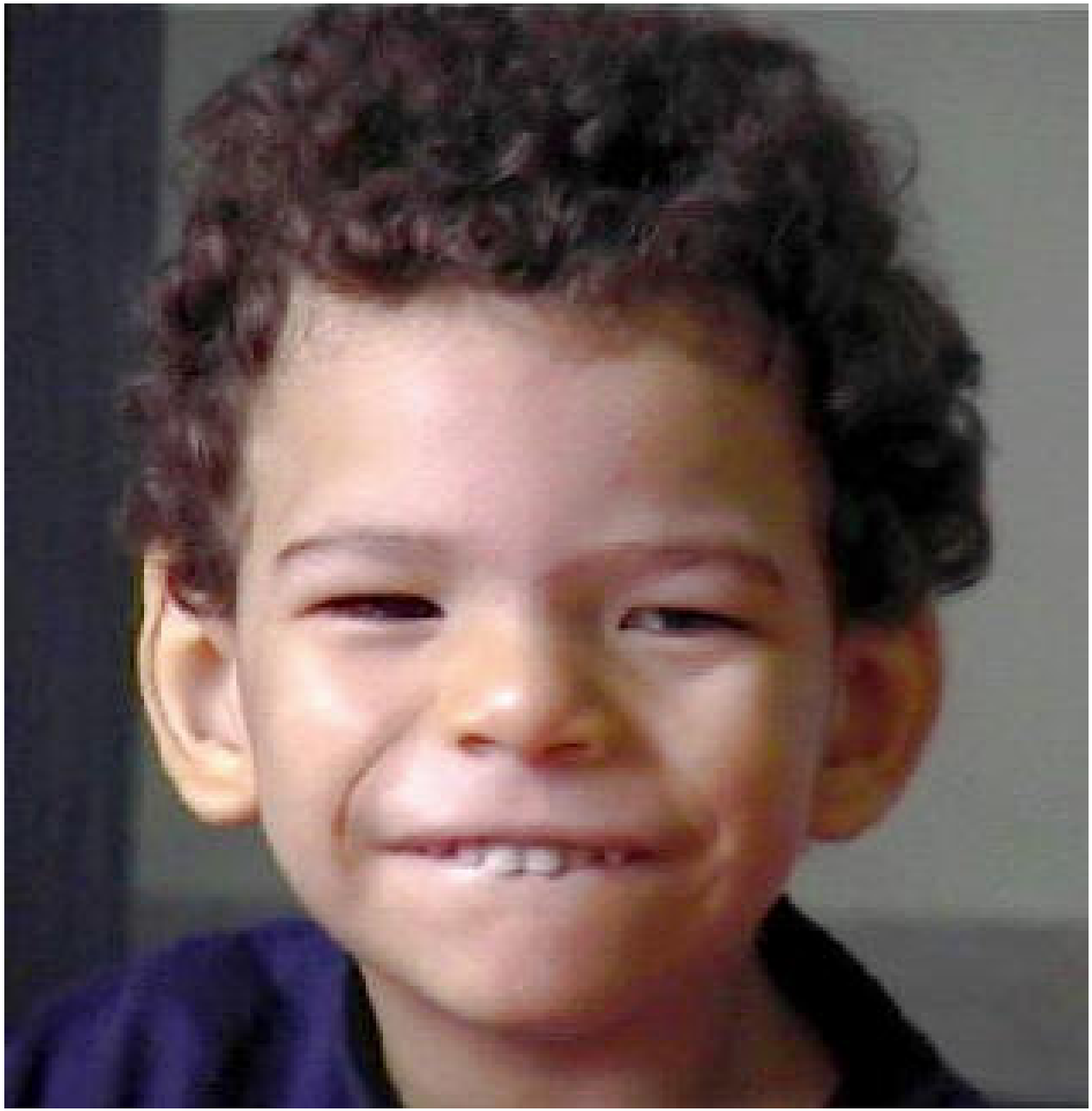
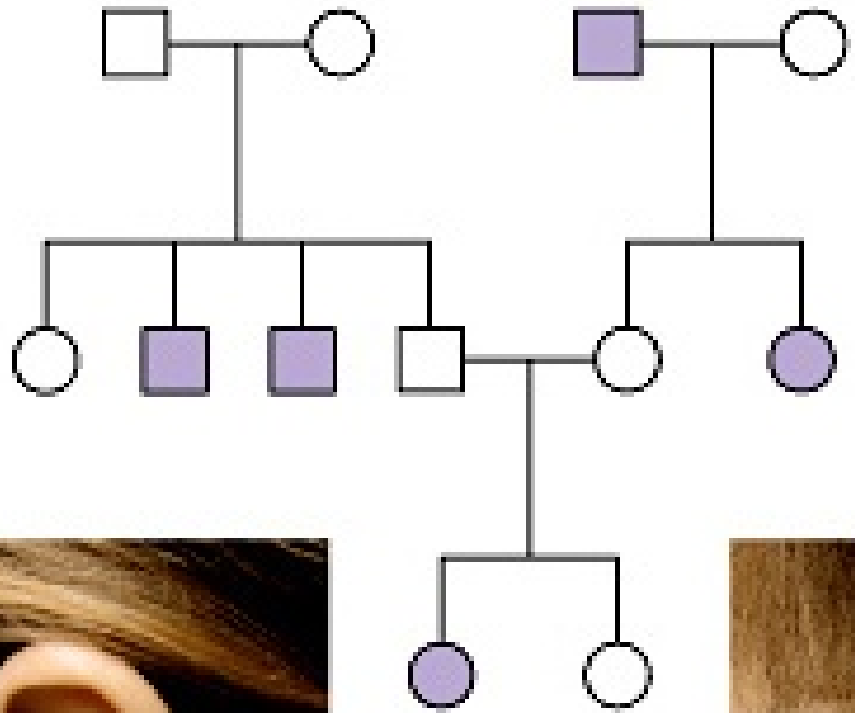


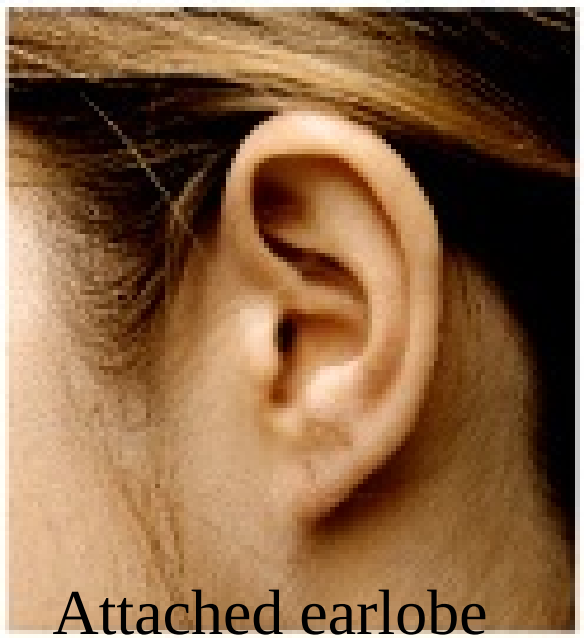
Fig 14.14 Use of pedigree analysis



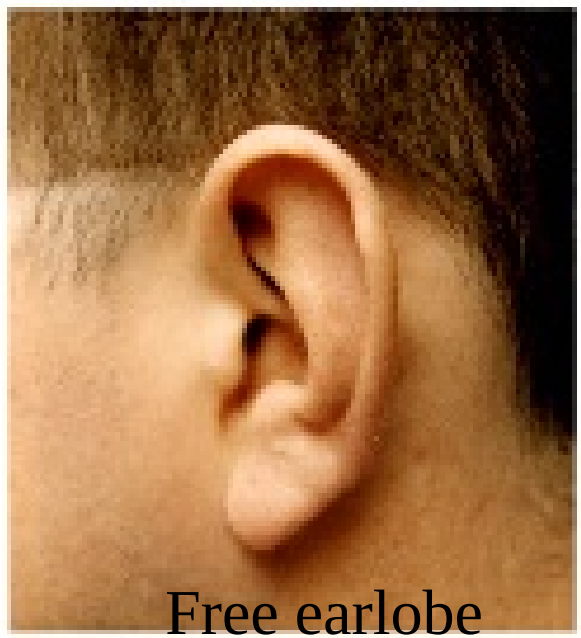
= male

= female

= attached

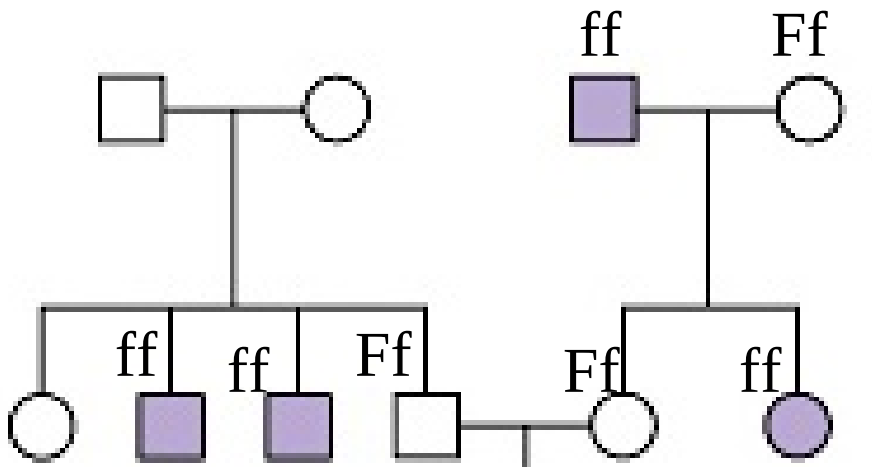


Attached earlobe



Free earlobe

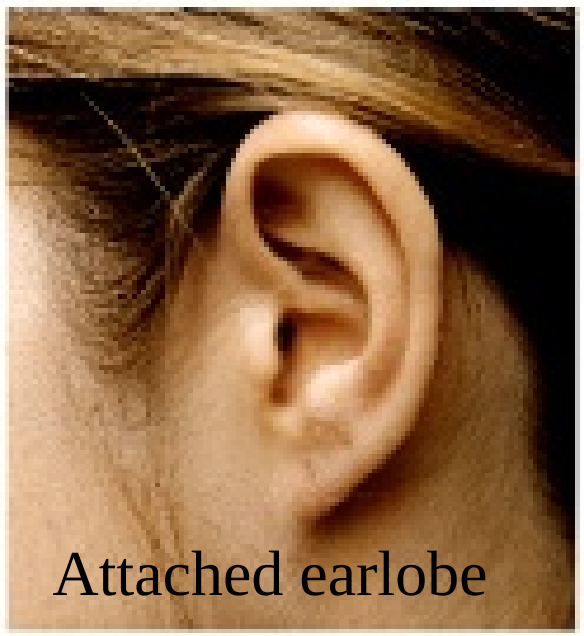
Fig 14.14 Use of pedigree analysis

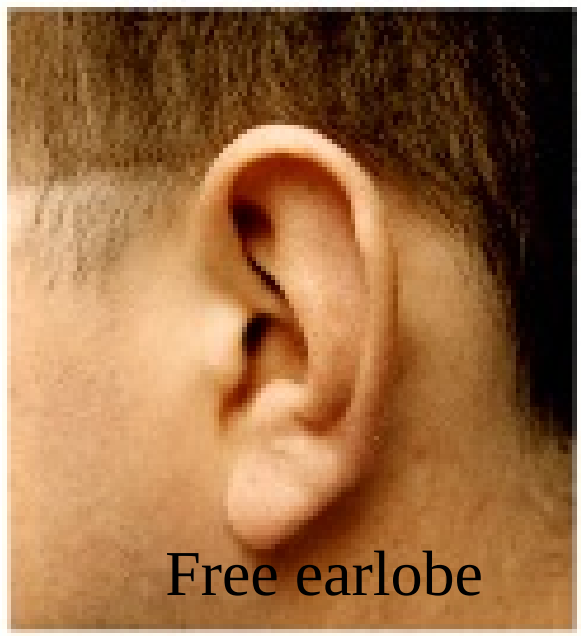


= male

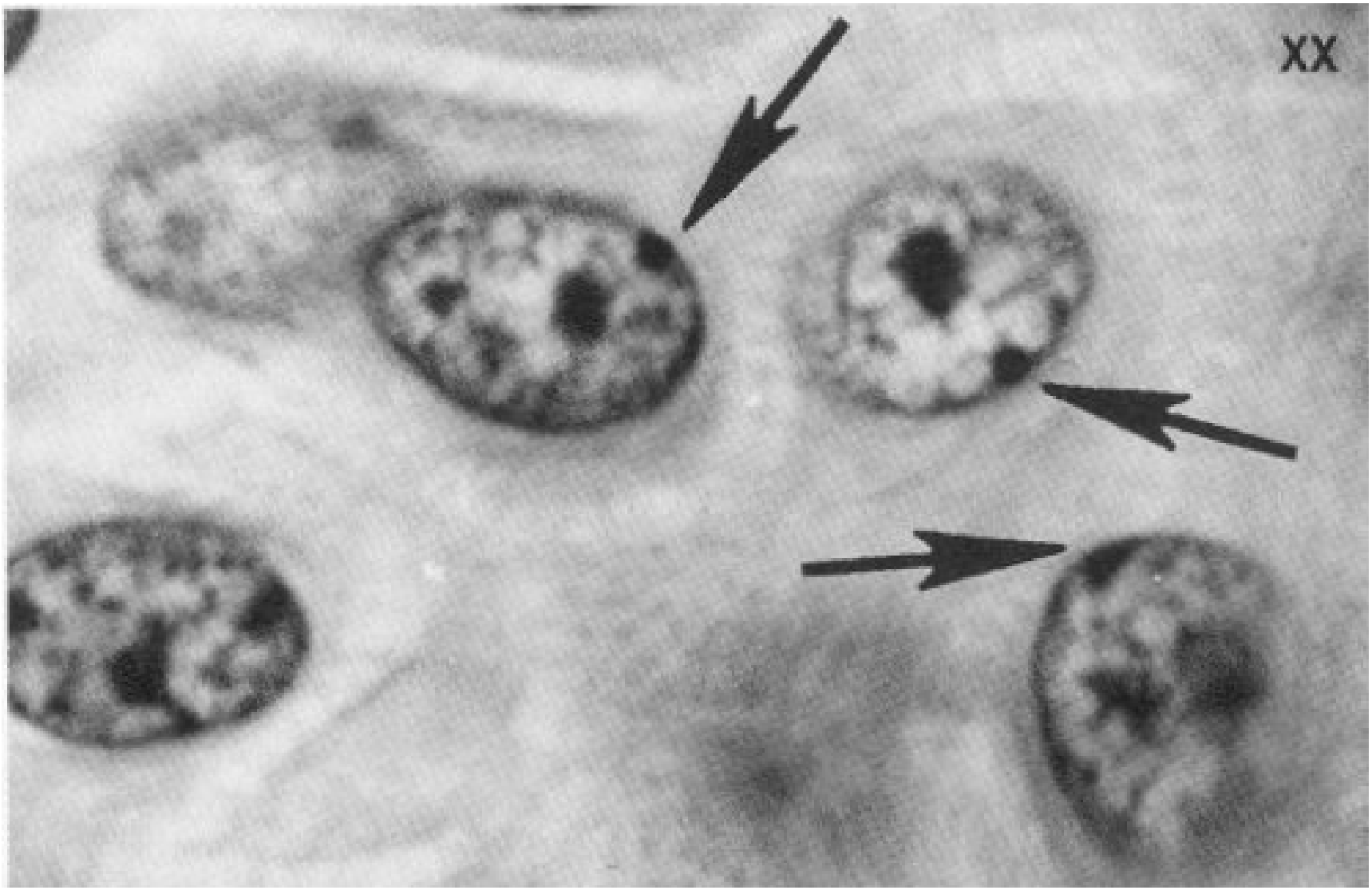
= female

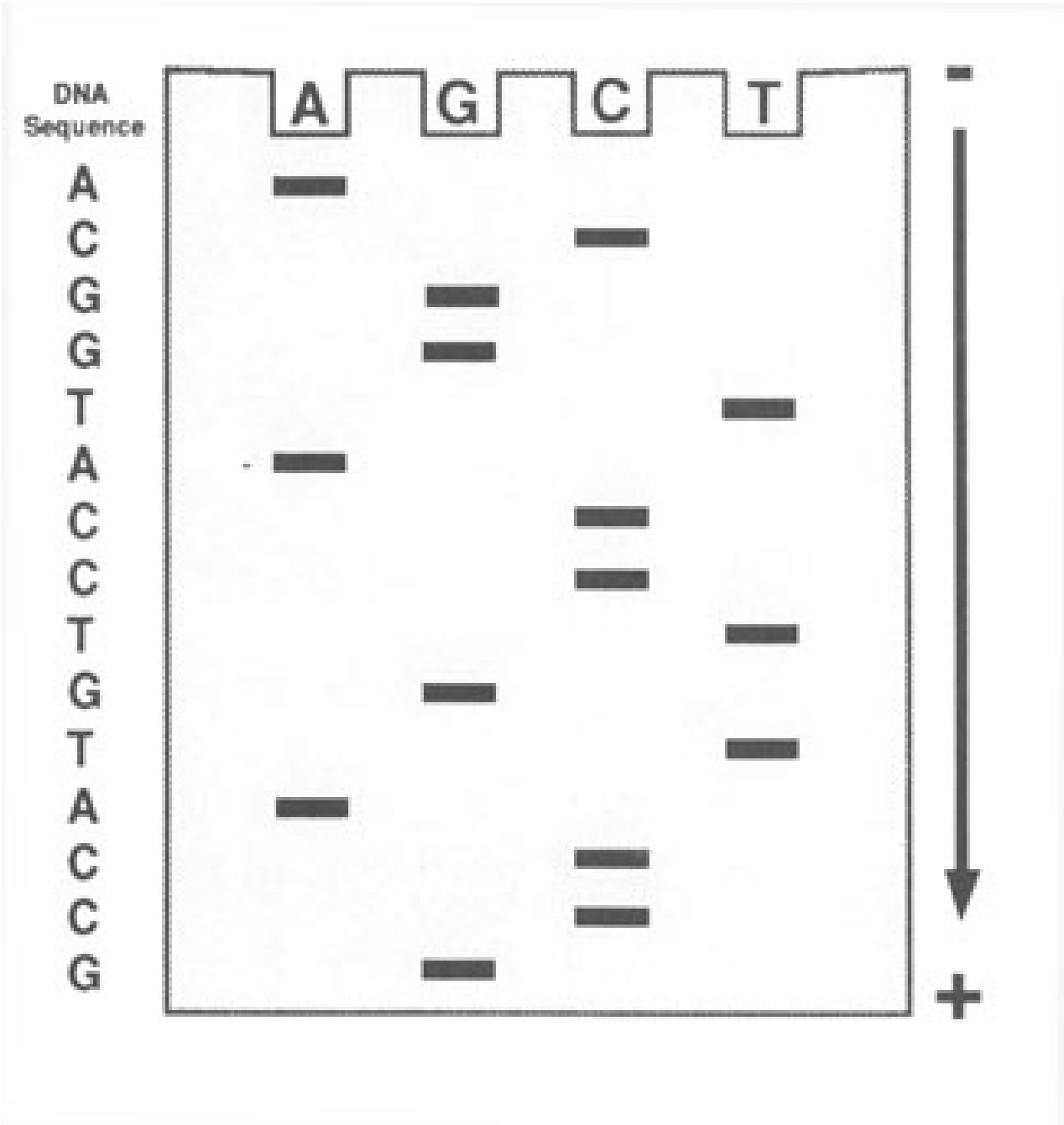
= attached





Sex chromatin: Barr body




DNA Sequence
A
C
G
G
T
A
C
C
T
G
T
A
C
C
G
A
G
C
T
-
+