9.S: Changes in Chromosome Number and Structure (Summary)

- Errors during anaphase in mitosis or meiosis can lead to trisomy and other forms of aneuploidy.
- Errors during the repair of DNA breaks or during meiotic crossing over can lead to chromosome rearrangements.
- Five common forms of aneuploidy in humans are 47,sex,+21 (Down syndrome), 47,XYY, 47,XXX, 45,X (Turner syndrome) and 47,XXY (Klinefelter syndrome).
- Deletion(5) causes a serious condition (cri-du-chat syndrome) because deletions are unbalanced chromosome rearrangements.
- Inversion(9) causes few health consequences because inversions are balanced chromosome rearrangements.
- Bright field microscopy can be used to detect chromosome number abnormalities and some chromosome rearrangements.
- Fluorescence in situ hybridization can be used to detect all types of chromosome abnormalities.
- PCR and DNA chip based techniques can be used to detect chromosome number abnormalities, deletions, and duplications.

Key Terms:

- origin of replication
- meiotic crossover
- telomere
- deletion loop
- centromere
- karyotype
- non-disjunction
- 46,XX
euploid
aneuploid
balanced
unbalanced
first division nondisjunction
second division nondisjunction
double strand break
nonhomologous end joining
DNA repair system
chromosome rearrangement
deletion
inversion
paracentric inversion
pericentric inversion
tandem duplication
translocation
reciprocal translocation
Robertsonian translocation

46,XY
47,sex,+21 (Down syndrome)
trisomy
47,XYY
47,XXX
monosomy
45,X (Turner syndrome)
pseudoautosomal region
47,XXY (Klinefelter syndrome)
46,sex,deletion(5) (cri-du-chat syndrome)
46,sex,inversion(9)
bright field microscopy
Giemsa stain
fluorescence in situ hybridization
fluorescent probe
DAPI stain
amniocentesis