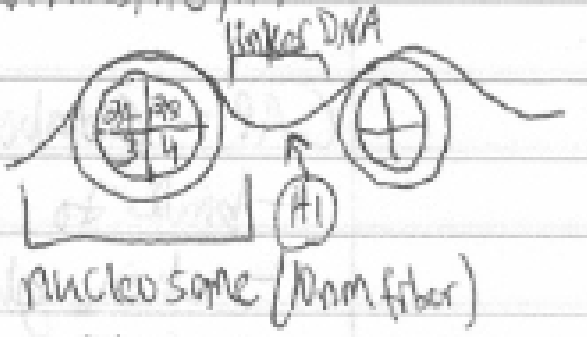
Chromatin = DNA + proteins (Histones)

levels of Organization

① 2nm in diameter = DNA

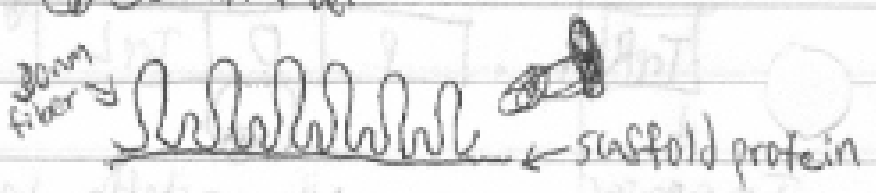
② 10nm fiber → DNA wrapped around core histones (4 types)

- H2A, H2B, H3, H4



③ 30 nm fiber - further coiled by binding of H1 to linker DNA.

④ 300nm fiber



⑤ 8 = 700nm fiber (chromatid)

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Chromosome Structure

Karyotype:
- 1-22 Autosomes
- 1 sex chromosomes

• separated by
- size
- centromere location
  - telocentric - at end of chromosomes
  - acrocentric - off center
  - metacentric - in middle
- p arm = short arm
- q arm = long arm

- G-banding - (giemsa stained) Dark band = heterochromatin, no transcriptional activity
  light bands = euchromatin, transcriptionally active

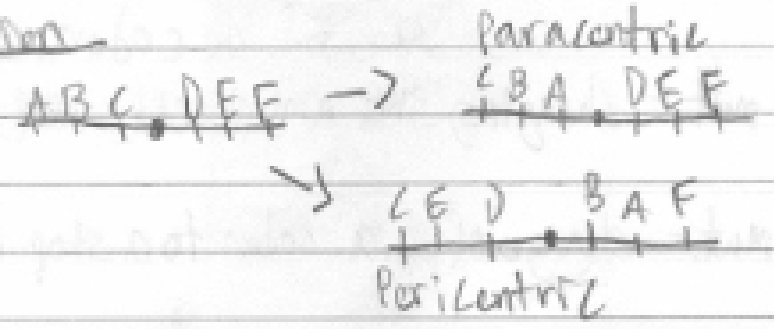
• Heterochromatin
  • constitutive - always heterochromatic
  • facultative - can be heterochromatic or switched to euchromatic

Abnormal chromosome structure + number

• Number - due to non-disjunction
  • referred to as aneuploidy, 2n+1 or 2n-1
  • polyploidy (aberrant Euploidy) - aberrant set #

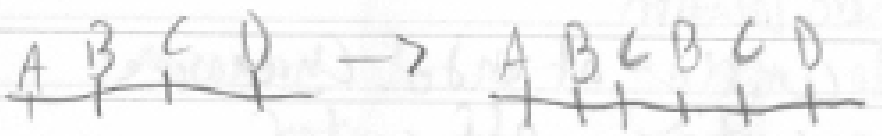
• Structure
  1. double strand break
     - accidental
     - recombination

① Inversion



② Deletion

③ Duplications



Translocation

- reciprocal - exchange b/w non-homologs
- non-reciprocal - one way exchange

Most common ex. 9-22 translocation (Philadelphia chromosome)

Point Mutations

Loss-of-function
Gain-of-function

Types

① base substitution: pyrimidine → pyrimidine
transitions: purine → purine A ↔ G C ↔ T
transversions: pyrimidine ↔ purine or A ↔ C

② base additions or deletions
(a) synonymous mutation - no change in amino acid seq. but does change seq. of a codon.
(b) missense mut. - changing the amino acid seq.

③ nonsense mut. - change of aa. codon to a stop codon