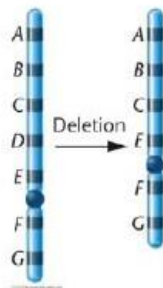


Lecture 16 & 17: Chromosome Rearrangements

Thursday, 25 May 2017 5:27 pm

Categories of Chromosomal rearrangements

1. Deletion - segment missing



2. Duplication - segment is repeated

And there are more....

The notes also include:

An abnormal chromosome only survives mitosis/meiosis if it has one centromere and two telomeres.

Rearrangements can be balanced or unbalanced:

- **Balanced**
 - No gain or loss of genetic material
 - Often harmless
 - Heterozygotes are at risk for producing offspring with unbalanced complement
- **Unbalanced**
 - Gain or loss of genetic material
 - Serious effect on organism

Deletions

- Loss of part a chromosome, resulting in effective 'monosomy' for just this segment
- Usually detrimental, deletion of greater than 2% of total haploid genome will be lethal
- Defects are due to haplo-insufficiency of one or more genes

Responsible for Cri-du-chat syndrome, segment of chromosome 5 is deleted.

Duplications

- Mostly caused by abnormal crossing over of the chromosomes mispair due to the presence of repeated sequences in genome. Called unequal crossing over.
- It can between non-sister chromatids during meiosis
- Or unequal crossing over can occur between sister chromatids during mitosis

Many small duplications have no phenotypic effect.

Leads to copy number variation. So some individuals can have more copies of a given gene than others

This section is further continued....

Another section includes:

Robertsonian Translocations

Unique to acrocentric chromosomes.

Involves the fusion of long arms of two acrocentric chromosomes to form one new metacentric chromosome

Short arms get lost. Chromosome number is reduced

