NUMERICAL CHANGES IN CHROMOSOME

Chromosomes are simple linear DNA molecules on which genes are located. For example, largest human chromosome, chromosome 1, has about 3536 genes. To ensure that each cell possesses these genes the chromosome has features that allow it to be passed on during cell division. **Origins of replication** found along its length provide places for DNA replication to start, **telomeres** protect each end of the chromosome, and a single **centromere** near the middle provides a place for microtubules to attach and move the chromosome during mitosis and meiosis.

If something goes wrong during cell division, an entire chromosome may be lost and the cell will lack all of these genes. The primary cause behind these chromosome abnormalites and is **Nondisjunction During Mitosis or Meiosis.**

Segregation of chromosomes occurs in anaphase. In mitosis and meiosis II, sister chromatids (of replicated chromosomes) are normally pulled to opposite ends of the cell (see Figure 2.12). In Meiosis I, it is homologous chromosomes, which are synapsed at that time, that segregate and move apart.

In rare cases, the sister chromatids (or paired chromosomes in Meiosis I) fail to separate, or dis-join. This failure to segregate properly is called **nondisjunction** and it can happen during mitosis, meiosis I, or meiosis II. This nondisjunction results in both chromatids (or chromosomes) moving to one pole and none at the other. One cell will have an extra copy and the other will lack a copy. Thus failure to segregate properly leads to unbalanced products.

Consequence of non-disjunction

The result of a non-disjunction event is daughter cells that have an abnormal number of chromosomes. Cells, such as the parent cell in Figure 1, which have the proper number of chromosomes, are said to be **euploid**. The daughter cells have one too many or one too few chromosomes and are thus **aneuploid**. Even though both product cells have at least one copy of all genes, both cells will probably die. The reason is due to the loss or gain of a large number of genes. Genes produce an standard amount of product - either functional RNAs or proteins. The parent cell shown has a **balanced** genotype because it has two copies of all of its genes. Each of its genes produces half of the products needed by the cell. But if one of these cells suddenly had only one copy (or three copies) of an important gene, the amount of product would be either 50% (or 150%) of what was required. Such a change for a single gene could probably be tolerated by the cell and it would probably survive. But the sudden change to one copy (or three copies) of the hundreds or thousands of genes on an entire chromosome the results would be more than tolerable and be catastrophic for the daughter cells. They have what’s called an **unbalanced** genotype, which usually decreases their viability.

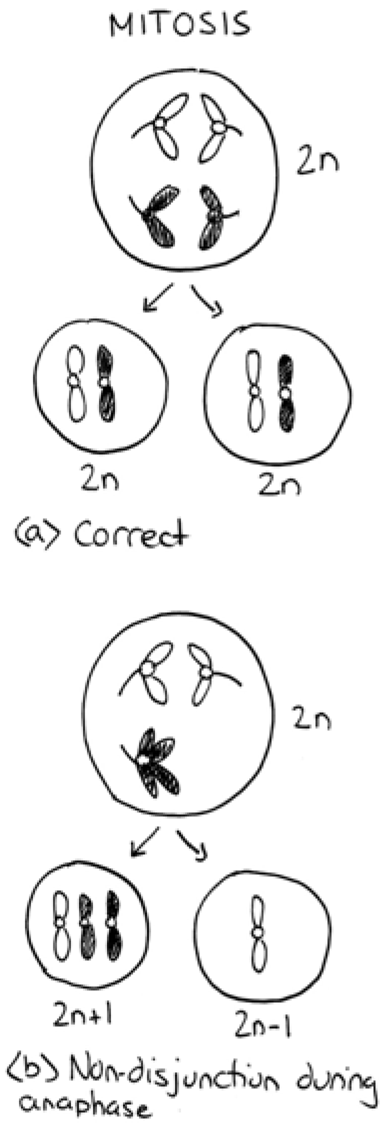

Figure 1: *Mitosis done successfully (a) and unsuccessfully (b). The cell is diploid and the homologs of one chromosome are shown in white and black. (Original-Harrington-CC:AN)*

If a **first division** or **second division nondisjunction** event occurs during meiosis the result is an unbalanced gamete (Figure 2b and c). The gamete will often be functional, but after fertilization the embryo will be genetically unbalanced. This usually leads to the death of the embryo. There are some exceptions to this in humans and these will be presented later in this chapter.

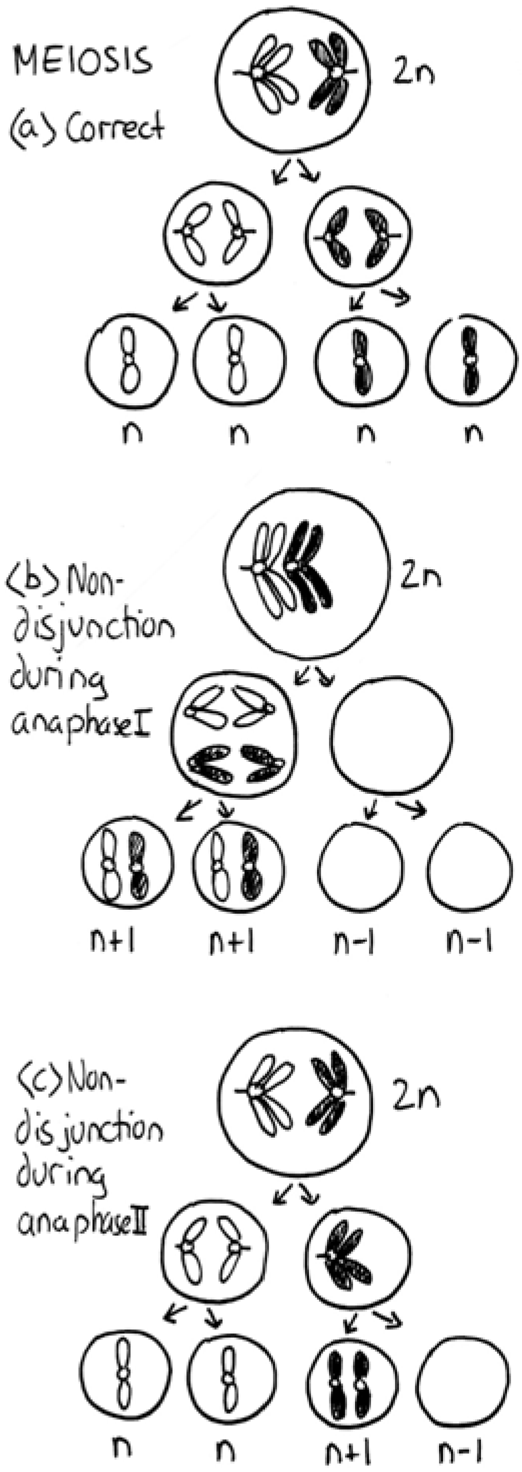

Figure 2: *Meiosis done successfully (a) and unsuccessfully (b and c). (Original-Harrington-CC:AN)*

p.s. The note has been prepared for students and does not have any original content