Genetics and Evolution
Pamela Diggle (pamela.diggle@colorado.edu)
Office: Ramaley C287
Office hours: Thursday and Friday 1-2 or by appointment

INTRODUCTORY MATERIAL

An example of the genetics and evolution of two inter-related diseases

Sickle cell disease is a form of anemia
It is the most common inherited blood disorder in the US
It affects 72,000 Americans
It affects 1 in 500 African Americans

Fig 5.21 Sickle-cell disease. These sickled cells become stuck in small blood vessels, causing a "crisis" of pain, fever, swelling, and tissue damage that can lead to death.

Capillary with red blood cells: www.tigr.org/tdb/ edb/pfdb/disease.html

Fig 5.21 Sickle-cell disease and hemoglobin structure

- Single amino acid substitution in one protein of hemoglobin
- Causes cell to "sickle" and reduces oxygen carrying capacity

Fig 17.23 Sickle-cell disease is caused by a single mutation

Why does sickle cell persist in human populations?

The distribution of the sickle cell gene coincides with the distribution of malaria

Malaria is caused by a plasmodium parasite
The parasite is carried by the female anopheles mosquito
Each year 400 million people contract malaria
About 2-3 million of these die from the disease
Most malaria fatalities are children

The sickle cell gene confers resistance to malaria!

Humans carry two copies of each gene

SS are susceptible to malaria
Ss are resistant to malaria and slightly anemic
Ss have sickle cell disease
Most survivors are carriers

When carriers have children

All three genotypes are produced

Fig 23.13 The sickle cell gene is maintained by stabilizing selection

ON TO MITOSIS

Chapter 12--Mitosis and the Cell Cycle
Read page 218 to 223 and pages 232-233
Review the Summary of Key Concepts on page 234 for section 12.1 and 12.2
Self-quiz (page 234): 3, 4, 9, 10, 11

From this lecture you should know:

- **What the cell cycle is and what occurs during each of the phases of the cell cycle**
- **The stages of the mitotic phase and the defining features of each stage**
- **The end products of mitosis and cytokinesis**
- **The difference between a chromatid and chromosome**

Two types of nuclear division

- Mitosis
  - Products are genetically identical
  - Same number of chomosomes
  - Same genes
  - Produces somatic cells
- Meiosis
  - Products are genetically different
  - One half the number of chromosomes
  - Different complement of genes
  - Produces gametes

Mitosis and meiosis are usually followed by **cytokinesis**, the division of the cytoplasm

Mitosis + cytokinesis + cell enlargement = growth in multicellular organisms
Mitosis + cytokinesis + cell enlargement = reproduction in unicellular organisms

Fig 12.5 Mitosis is part of the somatic cell cycle

- **Mitosis and cytokinesis = the mitotic phase**
- **The mitotic phase alternates with interphase**
- **Interphase is divided into three subphases**
  - G_1 = Gap 1--cell growth, biosynthesis
  - S = synthesis phase--DNA synthesis, duplication of chromosomes
  - G_2 = Gap 2--cell growth, biosynthesis
- **Differentiation**
  - Cell cycle ceases. Cell becomes specialized for particular function (liver, kidney, etc.)

DNA duplication occurs during S phase

Chromosomes are long strands of DNA

There are usually multiple chromosomes in a eukaryotic cell
Each chromosome exists as a long strand of DNA
Following duplication of a chromosome, it exists as two chromatids closely attached to each other
Chromatids are tightly attached to each other at the centromere

Fig 12.6 G2 of interphase
Cell enters G2 with 2X the amount of DNA
Each chromosome has been copied exactly
Each chromosome consists of two chromatids
Chromosomes not condensed
Nucelar envelope is intact

Fg 12.6 Prophase
DNA begins to condense by coiling
Each chromosome still consists of two chromatids
Mitotic spindle forms from microtubules

Fig 12.4
DNA duplication occurs during S phase
Condensation begins in prophase
Condensation is completed in prometaphase

Fig 12.6 Prometaphase
Nuclear envelope fragments
Chromosomes fully condensed
Centrosomes at poles
Some microtubules extend from poles of the cell toward equator
Some microtubules attach to chromosomes
Mitotic spindle consists of microtubules attached to chromosomes and some not attached

Fig. 12.6 Metaphase
Nuclear envelope is completely gone
Chromosomes at equator = metaphase plate

Fig. 12.7 Metaphase
What it really looks like under a light microscope

Fig. 12.6 Anaphase
Chromatids separate
One chromatid from each pair moves to opposite pole=>Genetic material is equally distributed
Each chromatid is a chromosome as soon as it separates from its sister

Fig 12.6 Telophase and Cytokinesis
Telophase

        Nuclear envelope forms around the chromosomes at each pole
        Chromosomes less condensed
        Two new genetically identical nuclei!
    Cytokinesis
        Division of the cytoplasm
        Differs in plants and animals
        Two new cells!

Fig 12.6 Review

Fig. 12.5 In multicellular organisms most cells will eventually stop dividing and differentiate

Fig. 12.19 Some cancers occur when control of the cell cycle is lost