4.5 Mistakes Happen Study Guide by Hisrich

4.5.a. Can a person with 45 or 48 chromosomes survive?

Humans normally have 46 chromosomes (23 from mother and 23 from father), though it's possible to survive with 45 IF the missing chromosome is the sex chromosome X and the person is a female (Turner Syndrome). A missing autosomal chromosome OR missing X sex chromosome is ALWAYS deadly.

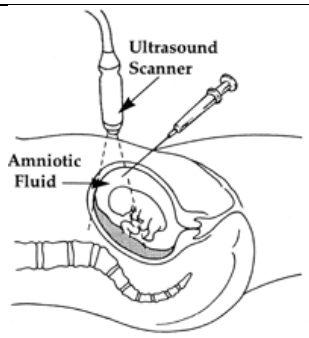
People can also survive with an extra chromosome (depending on the chromosome). It's typically only possible if the extra is a sex chromosome (the exception is Down's Syndrome).

Disorders with extra chromosomes (47)				Disorder with missing chromosome (45)
Trisomy 21 3 of chromosome 21 Down's Syndrome	XXX 3 sex chromosomes Triple X	XXY 3 sex chromosomes Klinefelter's Syndrome	XYY 3 sex chromosomes Superman Syndrome	XO 1 sex chromosome Turner Syndrome

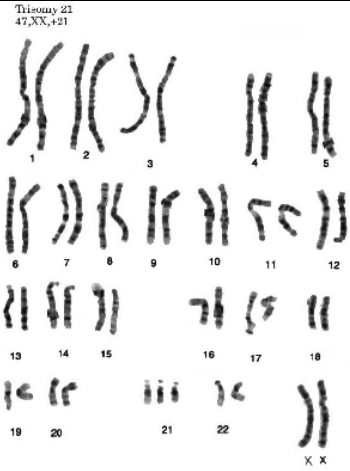
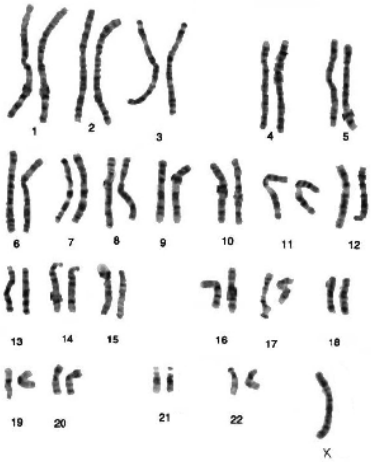
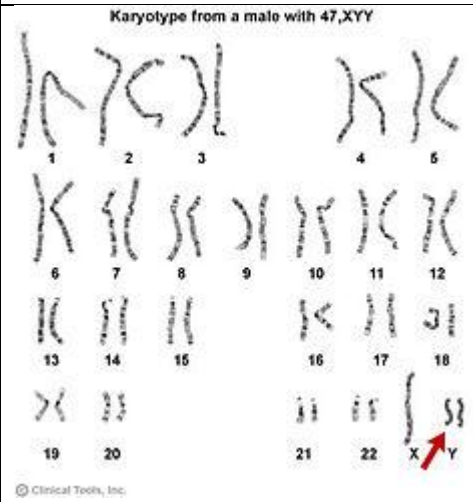
4.5.b. What happens if someone has more or fewer than 46 chromosomes?

Fewer	More
Dies before mother gives birth or within a year after (exception is Turner Syndrome, which results in retardation and infertility).	Dies before mother gives birth (exceptions shown in table above). If the person lives (s)he often experiences mental retardation and physical abnormalities (including infertility).

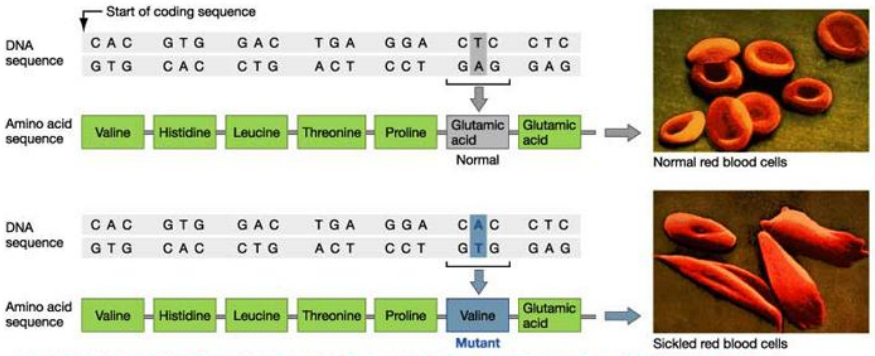
4.5.c. How can doctors detect if a patient has an abnormal number of chromosomes?



Karyotypes show an individual's chromosomes. They are used to diagnose chromosomal abnormalities (see table in 4.5.a.). They can be used on an adult who suspects he has a chromosomal abnormality, but that is unusual. Typically they are done on a fetus at about 20 weeks gestation. The obstetrician must perform amniocentesis to extract genetic material. A **karyotype** of the genetic material is then created. It shows the 22 pairs (hopefully) of autosomal chromosomes and the single pair (hopefully) of sex chromosomes. If there is an abnormal chromosome number, parents must decide whether to carry the child to term or whether to abort the fetus. There is no cure (at least yet) for chromosomal abnormalities.

Down's Syndrome	Turner Syndrome	Superman Syndrome
		

4.5.d. Can changing a single nucleotide in a gene cause a disease?

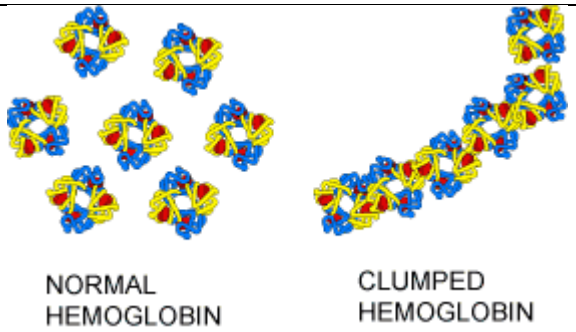


Yes (remember sickle cell?). These **mutations** are called single nucleotide polymorphisms ("many forms") are the most common type of **mutation** that occurs.

Since sickle cell disease is **recessive trait** (as are MOST **mutations**), an individual must receive a mutated allele from EACH parent to be affected. Otherwise, the normal **dominant trait** will be expressed (called sickle cell trait).

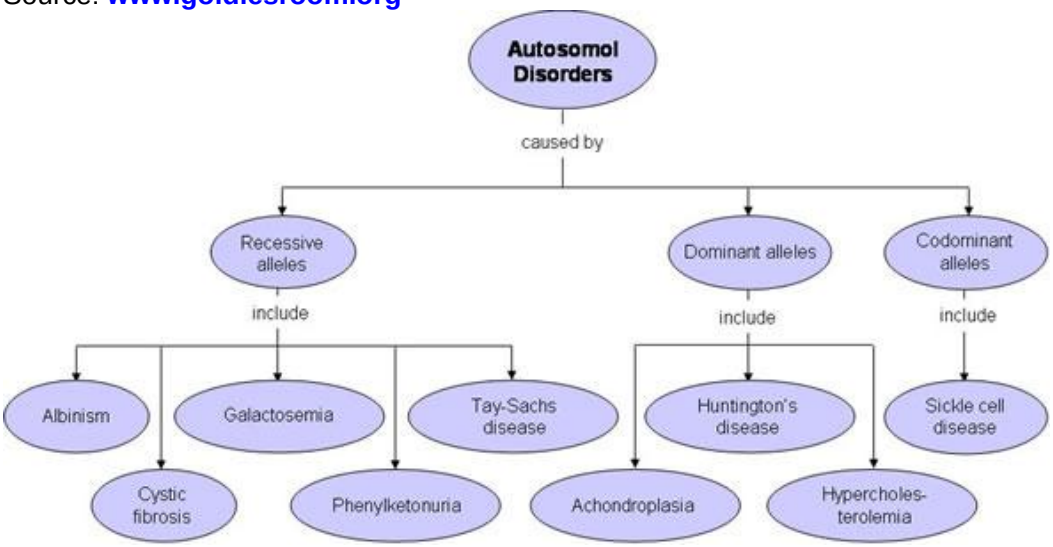
4.5.e. How is sickle cell hemoglobin different from normal hemoglobin?

Sickle cell hemoglobin simply has a single amino acid in the beta globin section that is different from that in normal hemoglobin. Instead of glutamic acid, a person with mutated hemoglobin has valine. Normal hemoglobin molecules don't attract each other, but mutated hemoglobin molecules do, causing them to clump (or **polymerize**) into long chains, pushing the blood cells into an elongated sickle shape. Valine is non-polar (and therefore **hydrophobic**) whereas glutamic acid is negatively charged, making it **hydrophilic**. The substitution of a **hydrophobic** amino acid for a **hydrophilic** one causes the protein to fold differently & behave differently.



4.5.f. What DNA mutations are directly linked to inherited diseases?

Source: **www.goldiesroom.org**



Autosomal mutations (shown in graphic left) are found in chromosome pairs 1-22 (not the sex chromosomes). The most common one are shown in the table left. Recessive diseases require a **mutated** allele to be inherited from each parent, whereas dominant disorders like dwarfism (achondroplasia) require only one. Sickle cell disease is sometimes considered co-dominant because in low oxygen environments, the cells of a person with sickle cell TRAIT may become sickle, though under normal conditions they do not.

Sex-linked **mutations** (carried on X-chromosome) include color-blindness, hemophilia, Duchenne muscular dystrophy, vitamin D resistant rickets, fragile X syndrome, Congenital aqueductal stenosis (hydrocephalus) & more.