7.013 Recitation 5 – Spring 2018

*(**Note:** The recitation summary should NOT be regarded as the substitute for lectures)*

Summary of Lecture 6 (2/23):

Classic experiment by Morgan that laid the foundation of human genetics: Morgan identified a gene located on the X chromosome that controls eye color in fruit flies. Since females have XX as their sex chromosomes, they have two alleles of each gene located on the X chromosome. In comparison, males have XY as their sex chromosomes and will only have one allele for all the genes located on the X chromosome (i.e. they are hemizygous). Morgan designed a mating experiment between a homozygous recessive female fly with white eyes (genotype X^wX^w) with a normal male fly with red eyes (genotype X^+Y) and obtained red-eyed females (genotype X^wX^+) and white-eyed male flies (genotype X^wY). This proved that white-eye color is a recessive trait and the allele regulating the eye color is located on the X- chromosome (i.e. eye color in flies shows an X- linked recessive mode of inheritance). You can read through the original article (link below) if interested!

http://www.nature.com/scitable/topicpage/thomas-hunt-morgan-and-sex-linkage-452

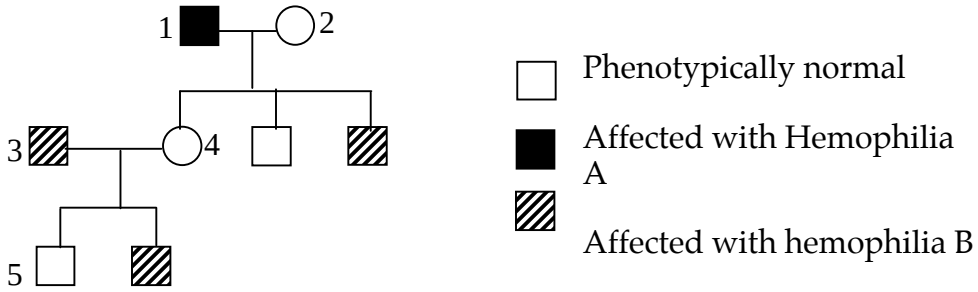
Pedigrees: A pedigree shows how a trait runs through a family. A person displaying the trait is indicated by a filled in circle (female) or square (male). Simple human traits that are determined by a single gene display one of six different modes of inheritance: autosomal dominant, autosomal recessive, X-linked dominant or X-linked recessive, Y- linked recessive or dominant and mitochondrial inheritance (which is always passed on from the mother to all her children). Autosomal traits are due to genes that lie on chromosomes #1 - #22 in humans. X-linked traits are due to genes that lie on the X chromosome. Since ~~the~~ females are diploid for X-linked genes but males are haploid, a male only needs to inherit the X-linked allele from his mother to show an X-linked recessive trait or phenotype. Thus, X-linked recessive traits appear more commonly in males. Females are usually carriers, meaning they have one X-linked recessive allele and appear phenotypically normal. X-linked dominant inheritance is extremely rare.

The table below summarizes the genotypes of affected, carrier, and healthy individuals for each mode of inheritance for a trait caused by a mutation in Gene A:

Mode of inheritance	**Genotype of…**		
	Affected Individuals	**Carriers**	**Normal healthy individuals**
Autosomal dominant	Aa or AA irrespective of their gender	No carriers	aa irrespective of their gender
Autosomal recessive	aa irrespective of the gender	Aa irrespective of the gender (phenotypically normal and healthy)	AA or Aa irrespective of the gender
X- linked dominant	Males: X^AY Females: $X^A X^A$ or $X^A X^a$	No carriers	Males: X^aY Females: $X^a X^a$
X- linked recessive	Males: X^aY Females: $X^a X^a$	Males are not carriers Females carriers: $X^A X^a$ (phenotypically normal)	Males: X^AY Females: $X^A X^A$ or $X^A X^a$
Mitochondrial	All children of an affected mother (irrespective of their gender) will have the disease.		
Y linked	All sons of an affected father will have the disease but no daughter of an affected father will have the disease.		

Questions

1. Consider the pedigree below showing the inheritance of two X-linked diseases, hemophilia A and hemophilia B. Hemophilia A is due to a lack of one clotting factor, and hemophilia B is due to a lack of a different clotting factor. Each clotting factor is a protein that is encoded by a specific gene located on the X chromosome. **Note that no individual shown in this pedigree is affected with both hemophilia A and hemophilia B.**



Write the genotypes for individuals 1-4 at both the hemophilia A and hemophilia B disease loci. How do you account for individual 5 not being affected with either hemophilia A or hemophilia B?

Individual	Genotype
1	
2	
3	
4	
5	

7.013 Introductory Biology

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