PWS 340
Homework 4 – Cytoplasmic Inheritance and Pedigree Analysis

Instructions. Work each of the problems below on paper until you are sure that you have the right solution. Input your answers on Learning Suite by following the appropriate link under the Exams tab (at the top).

Problems 1 – 2. Mutations in the *URF13* gene in maize (corn) mitochondria cause male sterility when the plant is homozygous for a recessive nuclear allele. In recent years, several nuclear genes have been discovered that are essential for restoration of male fertility in plants with a mutant *URF13* allele. Two of these genes are named *rf1* and *rf2*. (Recessive mutant alleles are designated in lower case, *rf1* and *rf2*, and the dominant alleles as upper case *Rf1* and *Rf2*.) These two nuclear genes assort independently of one another. For a plant to be male sterile, it must be homoplasmic for a mutant *URF13* allele and homozygous for a recessive mutant allele in *either* the *rf1* gene *or* the *rf2* gene. Suppose that a male-sterile plant with the genotype *rf1rf1Rf2Rf2* is fertilized by a male-fertile plant with the genotype *Rf1Rf1rf2rf2*. For the purposes of this question, assume that mitochondrial inheritance in maize is purely uniparental-maternal.

1. What proportion of the F_1 progeny are expected to be male sterile?
   A. 0 B. 1/4 C. 7/16 D. 9/16 E. 3/4

2. What proportion of the F_2 progeny are expected to be male sterile?
   A. 0 B. 1/4 C. 7/16 D. 9/16 E. 3/4

Problems 3 – 5. The *bicoid* (*bcd*) gene in *Drosophila melanogaster* has a role in establishing the polarity of the insect larva early in development. When homozygous in the maternal parent, a mutation in *bcd* has no effect on the parent but causes failure of anterior development in all of the offspring. The developing larvae have two abdomens, lack a head and fail to develop fully to adulthood. Suppose a heterozygous (bcd^+/bcd^-) female is testcrossed with a homozygous (bcd^-/bcd^-) male.

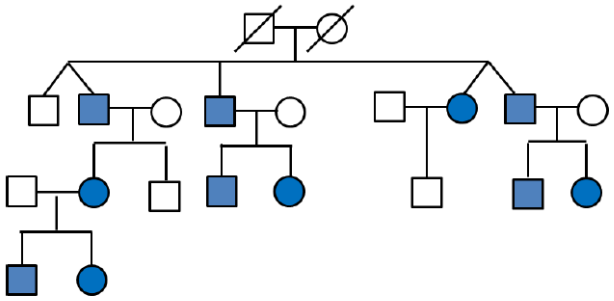
3. What is the expected frequency of bcd^-/bcd^- homozygotes in the adult progeny from the testcross?
   A. 0 B. 1/8 C. 1/4 D. 3/8 E. 1/2

4. If progeny flies from the testcross were allowed to randomly mate with each other, what would be the expected frequency of bcd^+/bcd^+ homozygotes in their adult progeny?
   A. 0 B. 1/8 C. 1/4 D. 3/8 E. 1/2

5. If progeny flies from the testcross were allowed to randomly mate with each other, what would be the expected frequency of bcd^-/bcd^- homozygotes in their adult progeny?
   A. 0 B. 1/8 C. 1/4 D. 3/8 E. 1/2

Problem 6. A dextral female *Limnnaea peregra* snail (P1) is mated with a sinistral male (P2) and all of their progeny are sinistral. Their sinistral progeny are allowed to self-fertilize producing one-half dextral and one-half sinistral snails. What is the genotype of the original P2 sinistral male parent?
   A. *rr* B. *Rr* C. *RR*

Problem 7. The complete absence of one or more teeth (tooth agenesis) is a common trait in humans—indeed, more than 20% of humans lack one or more of their third molars. However, more severe absence of teeth (severe tooth agenesis), defined as missing six or more teeth, is very rare and most likely due to an inherited condition. Lammi *et al.* (2004), *Am J. Hum. Genet.* 74:1043-1050) examined severe tooth agenesis in the Finnish family shown in the pedigree below. The parents in the first generation were deceased at the time the pedigree was constructed and their phenotype is unknown. What is the most likely mode of inheritance for severe tooth agenesis in this family?



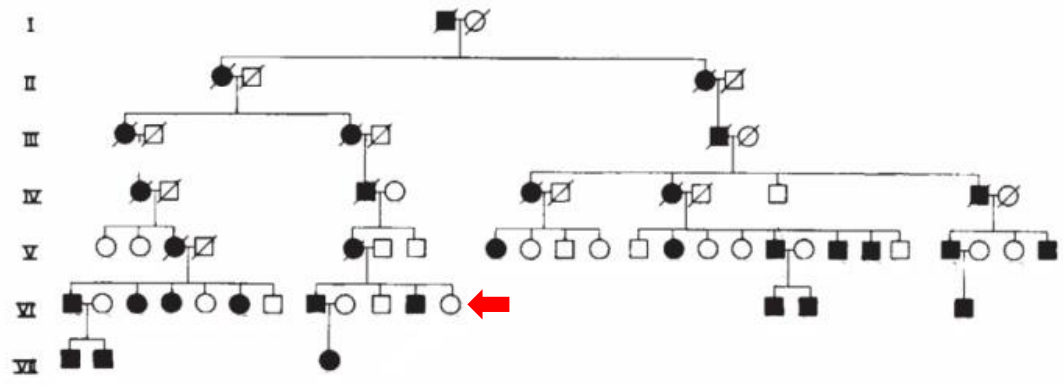
A. Autosomal recessive
B. Autosomal dominant
C. X-linked recessive
D. X-linked dominant
E. Y-linked

Problem 8. *Chlamydomonas reinhardtii* is a unicellular algae that contains a single chloroplast, each with about 50 – 100 genomes and multiple mitochondrial organelles also with multiple genomes. Although *C. reinhardtii* exists mainly as a haploid organism, occasionally two cells will fuse to form a diploid cell which immediately undergoes meiosis to form four haploid progeny. The cells that fuse must be of opposite mating type, mt^+ and mt^-. Inheritance of the plastid genome is uniparental mt^+, whereas inheritance of the mitochondrial genome is uniparental mt^-. Mutant alleles which confer resistance to antibiotics have been identified in *C. reinhardtii* and these are quite often variants of genes found in organellar genomes. For instance, a mutant allele conferring resistance to streptomycin (str^R) maps to the chloroplast genome and a mutant allele conferring resistance to hygromycin (hyg^R) maps to the mitochondrial genome (wild-type sensitive alleles are designated with a superscript *S*). Suppose you mate an mt^+ str^R hyg^S strain of the algae with an mt^- str^S hyg^R strain. What will be the most likely phenotype of a progeny cell?

A. streptomycin resistant, hygromycin resistant
B. streptomycin resistant, hygromycin sensitive
C. streptomycin sensitive, hygromycin resistant
D. streptomycin sensitive, hygromycin sensitive

Problem 9. Huntington's disease is a devastating human genetic disorder that is caused by expansion of a CAG repeat in a gene located on chromosome 4. Normally, the repeat is found about 10 to 28 times but in mutant alleles it is repeated 36 to 120 times. The disease affects neurological function that first manifests as behavioral disturbances but soon develops into extreme personality changes, speech changes, loss of motor function and dementia. It ultimately takes the life of its victims. In 1983, Gusella *et al.* (*Nature* 306:234-238) published the pedigree shown below of a Venezuelan family in which the disease was prevalent. Presence or absence of the disease was confirmed by DNA marker analysis for living members of the family and from medical records for deceased family members. Suppose the woman in generation VI (indicated with a red arrow) were to marry a man who does not have the disease. What is the probability that a child born to the couple would be afflicted with Huntington's disease?

A. 0 B. 1/4 C. 1/2 D. 3/4 E. 1



Problem 10. In the human retina specialized cells called cones provide vision in bright light, including color vision. Three types of cones each contain a photopigment that is most sensitive to wavelengths of light in either the red, green or blue area of the spectrum. The brain combines input from all three types of cones to produce normal color vision. Protanopia and deuteranopia are the most common forms of color blindness in humans, both affecting the ability to distinguish between red and green colors. Protanopia is caused by mutations in the *OPN1LW* gene resulting in the loss of red pigment cones known as L cones, whereas deuteranopia is caused by mutations in the *OPN1MW* gene resulting in loss of green pigment cones known as M cones. Both genes are located on the X chromosome, although they are not genetically linked. In 1957, Franceschetti and Klein (*Acta Genet. Stat. Med.* 7:255-259) published the pedigrees of two families in which both parents were colorblind but each for a different type. In one case, a female with deuteranopia married a male with protanopia and they had three sons and one daughter. How many of the children born to this couple do you expect to be colorblind?

A. 0 B. 1 C. 2 D. 3 E. 4