

Bio Final Review

Daniel Filipski

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1 General Information

For this, see the google doc.

2 Genetics

2.1 Sex-linked genes

These are genes located on the sex chromosomes. They will show different phenotype frequencies based on gender.

- (1) Gene A is on the X chromosome. A is the wild type, and α is the diseased type.
 $X^A X^\alpha \times X^A Y$:

$$\begin{array}{c|c} X^A Y & X^\alpha Y \\ \hline X^A X^A & X^\alpha X^A \end{array}$$

2.2 Pedigrees

- = Unaffected Female
● = Affected Female
□ = Unaffected Male
■ = Affected Male

Connecting lines on pedigrees work just as they do on family trees. Relatively simple logic can be used to determine the genotypes of each member of the pedigree; however, some can be more difficult than others. My general method is to use the “method of staring” in the words of Mr. Letarte.

2.3 Genetic Disorders – Sickle Cell, Cystic Fibrosis, Huntington’s

Sickle Cell

- Red blood cells contain hemoglobin, which bind O_2
- Hemoglobin is made up of two α -globin and two β -globin polypeptides
- Mutation in β -globin makes it slightly less soluble
- When O_2 is low, hemoglobin without O_2 will start to clump and form long fibers that will change the shape of the red blood cell, which will then get stuck in capillaries
- If one is a heterozygote of this disease, they have an advantage against malarial
- Sickle Cell disease is recessive, because its effects are not great enough with only some of the β -globin broken.

Cystic Fibrosis

- In frame three base pair deletion in gene for CFTR
- CFTR is missing one amino acid (phenylalanene), which causes it to misfold and be destroyed
- CFTR is a channel in the epithelial cell membranes for Cl^-

- Without CFTR, there is too much extracellular Cl^- , which makes the fluid outside the cell thicker
- Mucus clogs lungs and serves as a growth substance for *Pseudomonas aeruginosa*
- The allele for Cystic Fibrosis is recessive, as with one of the two CFTR, cells still have enough paths for Cl^-

Huntington's Disease

- Mutation is dominant, but the disease does not present itself until late 30's or early 40's
- Huntingtin gene expressed in nerve cells. Its developmental role in adults is unclear
- CAG (codes for glutamine)
- Wild type 6-35 repeats
- Diseased 36+ repeats
- Diseased protein forms aggregates in neurons, which lead to cell death.

2.4 Nondisjunction

Nondisjunction Event – Failure to separate chromosomes

This is more common in meiosis I. Trisomy 21 causes Down syndrome.

2.5 Recombinant DNA – Restriction Enzymes, Ligase, Electrophoresis, GFP, PCR, Selectable Markers, Screens, Plasmids, Transformations

Recombinant DNA – Combination of two or more pieces of DNA to create an artificial construct.

Building Pieces of DNA:

1. Synthesize from scratch
2. Cut and Paste
 - Ligase is not specific and will join any two pieces of DNA.
 - Restriction enzymes originate from bacteria, where they serve as a type of immune system.
 - Restriction System: methylase adds CH_3
 - Restriction Enzyme cuts DNA if not regulated.

Eco RI:

- Sticky ends of *GAATTC* (a DNA reverse palindrome)
- Eco RI cuts between the G and the A

Plasmid:

- Mini chromosome in bacteria
- Must have an ori, a selectable marker
- Plasmids can be shared between cells. They also can be picked up from the environment, when they are there for whatever reason.
- An example of plasmids which can be shared between cells is antibiotic resistance.

DNA Sequencing:

- Denature DNA into single strands
- Add primer for only one strand
- Provide DNA polymerase and the four nucleotides, with a small fraction of the nucleotides modified so that DNA polymerase cannot extend from them (remove 3' hydroxyl)

2.6 Selective Breeding – Hybridization vs Inbreeding

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