

GEN3051 Exam Potential Questions (Practice)

Part B - Short Answer

1. What Are Some Complications In Inheritance Patterns? (Week 1)

1. Expressivity
 - phenotype is variable in its degree of expression
2. Penetrance
 - some individuals with affected genotype do not show the phenotype
 - phenotype is variable in whether it is expressed or not.
3. New Mutations Arising
 - sporadic occurrence of phenotype due to new mutation
4. Locus heterogeneity
 - Mutations in different genes may show same phenotype

2. What Are Some Variations On Positional Cloning (Week 2)

1. assisted by patients with chromosomal breaks that cause the disease
Eg: Duchenne Muscular Dystrophy (DMD)
2. use position-independent approaches – relies on knowledge of protein product
Eg: Hemophilia A
3. identify gene first in a model organism with similar mutant phenotype
Eg: Waardenburg syndrome type 2

3. What are the factors to consider when creating and using a model genetic organism? (Week 3)

10 Factors to consider in a model genetic organism

1. ease and cost of breeding
2. number of progeny
3. generation time
4. genetic techniques
5. genetic background variation
6. cell/organ culture techniques
7. genome sequence
8. similarity to human
9. ethical concerns – is the question worth asking / worth using the animal for?
10. Many possible models – select the most suitable for the question asked

4. What Are Mechanisms Of A Gain Of Function Mutation? (Week 3)

- requires a much more specific change \therefore less common.
- generally dominant
- same condition should not be produced by a deletion mutation.
- Mechanisms of gain of function mutations:
 1. **Exon Shuffling** - chromosomal rearrangement joins functional exons of two different genes to give novel gene
→ Eg: Pittsburgh variant of α_1 -antitrypsin

2. **Overexpression** - gene duplication or transposition to different chromatin environment.
→ Eg: Charcot-Marie-Tooth syndrome:
3. **Constitutively active signalling proteins** - G protein coupled receptors.
Receptor signals whether ligand present or not.
→ Eg:: Achondroplasia