

## Sensory Systems Basics and Principles I

### Lecture: Pathology of the Eye - Genetics

1. Define the term incidence. Why is the incidence for a disease mostly lower than the prevalence ?
2. Consider a couple in which both are heterozygous gene carriers for a recessive disease. If this couple has two children, what is the probability that both are affected/unaffected ?
3. Retinitis pigmentosa (RP) is the most common hereditary retinal dystrophy. Which statement(s) are wrong:
  - RP can follow various modes of inheritance
  - RP patients are blind from birth
  - RP affect more males than females
  - RP can be efficiently treated by corneal transplantation
4. Give (clinical) parameters that are used to categorize subtypes of hereditary retinal dystrophies
5. Hereditary Retinal Dystrophies (HRDs): Which of the following disorders belong to the group of HRDs:
  - Retinitis pigmentosa
  - Glaucoma
  - Achromatopsia
  - Usher Syndrome
6. What does the term *disease allele heterogeneity* mean ?
7. What is the advantage of the linkage analysis & positional cloning approach to identify a disease gene
8. List main types of mutations and their consequences
9. List at least two types of markers that can be applied in linkage analysis
10. Give potential modes of action of a dominant mutation.