

CYTOGENETICS

Lesson 7: GENETIC MUTATION | BSMT - 2nd Year

GENETIC MUTATION

HOW TO EASILY REMEMBER START CODON AND STOP CODON?

- **AUG** k let us **START**
- **UAA**/ umu-**UGA**, **UAG** po **STOP** po

THE NATURE OF GENE VARIANTS

MUTATION

- Alter proteins resulting sequence alteration
- Mutations may occur spontaneously or by exposure to a chemical or radiation
- **Mutagen** - agent that causes mutation
- Rare in a population and typically affects the phenotype
- Are important to evolution
- Our evolutionary relatedness to other species allows us to study many mutations in nonhuman species

EXPOSURE TO MUTAGENS

- Some mutagen exposure is unintentional
 - Workplace
 - Industrial accidents
 - Chernobyl
- Medical treatments
- Weapons
- Natural sources
 - Cosmic rays, sunlight, earth's crust

POLYMORPHISM

- Simply means **many forms**
- Change in a gene that is less rare than a mutation and more prevalent in a population

THE EFFECT OF MUTATIONS VARY

"LOSS OF FUNCTION" MUTATIONS

- Recessive trait
- A mutation that results in reduced or abolished or inactive or less active protein function

"GAIN OF FUNCTION" MUTATIONS

- Dominant trait
- Are less common
- Confer an abnormal activity or more protein activity or acquisition of a different function

MUTANT

- Refers to an individual having a phenotype that differs from the normal phenotype
- Usually connotes an abnormal or unusual, or even uncommon variant that is **nevertheless "normal"**

TYPES OF MUTATIONS

MUTATIONS CAN BE CLASSIFIED IN SEVERAL WAYS

- By whether they remove, alter or add a function
- By exactly how they structurally alter DNA

SPECIFICALLY:

1. Germline Mutations
2. Somatic Mutations
3. Chromosomal Alterations
4. Point Mutations
5. Frameshift Mutations

1. GERMALINE MUTATIONS

- Occur in gametes
- Change occurs during the DNA replication that precedes meiosis
- Transmitted to the next generation of individuals

Mutation And Genetic Unit 7 Lesson 2

Peipei Pang



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Health: Teacher ed Linda Brower Meeks,1990 Advanced Genetic Analysis Philip Meneely,Matthew R. Willmann,2009-01-15 Advanced Genetic Analysis explores the question How can the principles of genetics be used as analytical tools to solve biological problems Drawing on the latest experimental tools including microarrays RNAi and bioinformatics approaches it provides a state of the art review of the field but in a truly student friendly manner

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