

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

Stephanie Zinn

A red circular graphic with a gradient, appearing as a semi-circle or a partial circle, located to the right of the author's name.

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Health: Teacher ed Linda Brower Meeks,1990 *Science in Your World: Teacher edition* Jay K. Hackett,1991

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 Genomes of Herbaceous Land Plants ,2013-12-18 Advances in Botanical Research publishes in depth and up to date reviews on a wide range of topics in plant sciences Currently in its 69th volume the series features several reviews by recognized experts on all aspects of plant genetics biochemistry cell biology molecular biology physiology and ecology This thematic volume features reviews on genomes of herbaceous land plants Publishes in depth and up to date reviews on a wide range of topics in plant sciences Features a wide range of reviews by recognized experts on all aspects of plant genetics biochemistry cell biology molecular biology physiology and ecology Volume features reviews on genomes of herbaceous land plants **NJN ... Secondary Teacher Manual, Grades 7-12** ,1988 *Discover Science: Teacher's annotated edition* ,1991 Science content helps develop the skills needed to understand how science works learn new concepts solve problems and make decisions in today s technological society *College Biology I* James Hall Zimmerman,Sophie E. Merritt,1963 *Paller and Mancini - Hurwitz Clinical Pediatric Dermatology E-Book* Amy S Paller,Anthony J. Mancini,2020-12-08 Written by two leaders in the field of pediatric dermatology this classic text provides both detailed content for the specialist and easily accessible information for the non dermatologist and less experienced clinician Paller and Mancini Hurwitz Clinical Pediatric Dermatology 6th Edition comprehensively covers the full range of skin disorders in children offering authoritative practical guidance on diagnosis and treatment in a single volume This award winning evidence based text has been fully revised and updated and is an essential resource for anyone who sees children with skin disorders Features new content outline boxes for faster navigation hundreds more clinical images and authors tips for the diagnosis and treatment of pediatric skin diseases Contains updated evidence based guidance and the latest drug developments and disease classifications Provides a careful balance of narrative text useful tables and 1 300 high quality clinical photographs helping you recognize virtually any skin condition you re likely to see Includes a greatly expanded discussion of atopic dermatitis and psoriasis and new therapeutic approaches for treating genetic disorders and systemic diseases such as ichthyoses and rheumatologic disorders Discusses new tests for subclassifying disease such as the myositis specific antibodies of juvenile dermatomyositis genotyping and immunophenotypes of inflammatory skin disorders Contains new and updated tables on psoriasis co morbidities genetic syndrome classifications acne therapies pediatric histiocytoses PHACE syndrome criteria HSV therapies and juvenile dermatomyositis Features updated sections on infections exanthems vascular disorders dermatoses and genodermatoses Discusses hot topics such as the use of stem cell and cell therapy as well

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