

Regulation of the Lactase Gene



About This Worksheet

This worksheet complements the Click and Learn "Regulation of the Lactase Gene" developed in conjunction with the 2011 HHMI Holiday Lectures on Science "Bones, Stones, and Genes: The Origin of Modern Humans."

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Key Concepts

- Eukaryotic gene expression is regulated at the levels of transcription, RNA processing, translation, and post-translation.
- Proteins called transcription factors bind to DNA and control transcription. Different types of transcription factors can increase or decrease transcription.
- Mutations in DNA regions that control gene expression can alter the way genes are expressed.
- Infants have high lactase levels so that they can digest their mothers' milk, but lactase gene expression is turned off after weaning.
- Lactase persistence is the trait that allows adults to continue to digest milk. It results from a mutation in an enhancer region of the lactase gene.

Instructions

1. Go to BioInteractive.org.
2. In the menu bar under the masthead, mouse over Topics → Evolution → Interactive.
3. Scroll down the page and click on "Regulation of the Lactase Gene."
4. Proceed through the slides, watch the embedded video clips, and answer the following questions in the space provided.

Questions

1. What is the difference between lactose tolerance and lactose intolerance? Be specific.

The main difference between lactose tolerance and lactose intolerance is the ability to digest lactose, a carbohydrate commonly found on dairy products, to its simpler units – glucose and galactose through the enzyme lactase. Basically, a person is considered lactose intolerant if he/she does not have/ have reduced lactase in his/her small intestine.

2. Why is lactose tolerance also called lactase persistence?

Lactase is a normal enzyme found in the human body most especially during its infant to childhood stages. As we age, the ability to produce the enzyme reduces and in some cases it completely stops. This makes a person lactose intolerant. Lactose tolerance is also called lactase persistence because these people did not stop from producing lactase even post childhood stage. They have persistently produced this enzyme which allow the person to easily digest dairy products with ease.

3. In which cells or tissues is lactase produced and what is its role?

Lactase is primarily produced in the cells of the walls of the small intestine. The small intestine functions for the absorption of nutrients from digested food as well as further digestion of food. To be more specific, proteins, fats, and carbohydrates are digested here with the help of many gastric juices. Hence, lactase is one of the carbohydrates that is digested in the small intestine with the help of lactase.

4. What normally happens to the levels of lactase produced throughout a person's lifetime?

LCT gene, the gene responsible for the translation of lactase, is very active during infant to childhood stage. As a person ages, the LCT gene becomes inactive thus greatly affecting the production of lactase. In general, as a person gets older, the more the person becomes lactose intolerant.

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Lactase gene expression Edmond Henri Herman Maria Rings, 1994 *Molecular and Population Genetic Analyses of Variation Within and Surrounding the Human Lactase Gene* Edward John Hollox, 2000 **Molecular and Population Genetic Analysis of Variation Within and Surrounding the Human Lactase Gene**, 2000 **The Origins of Lactase Persistence and Ongoing Convergent Evolution** Beth A. Keller, 2011 As a primary factor in human evolution natural selection is an important component of genetic research Studies of lactase persistence suggest that positive selection has played a powerful role in the adaptation to a lifelong consumption of fresh milk Using multiple research studies of lactase persistence and suspected corresponding single nucleotide genetic polymorphisms this study combines data sources to determine whether evidence exists for natural selection of a specific cytosine to thymine genetic mutation located 13 910 base pairs T 13910 upstream from the lactase gene This polymorphism has potential to be a causal element for lactase persistence and data suggest that natural selection has played a role in the rising frequency and distribution of this allele if only in some regions European and neighboring regions appear to have the highest frequencies with little or no frequency in Asia Africa and Indonesia however the presence of lactase persistence in those areas suggests convergent evolution may be occurring on a phenotypic level To examine this possibility several other identified polymorphisms in the same region as the T 13910 will be included in this study

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Table of Contents Regulation Of The Lactase Gene Answer Key

1. Understanding the eBook Regulation Of The Lactase Gene Answer Key
 - The Rise of Digital Reading Regulation Of The Lactase Gene Answer Key
 - Advantages of eBooks Over Traditional Books
2. Identifying Regulation Of The Lactase Gene Answer Key
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Regulation Of The Lactase Gene Answer Key
 - User-Friendly Interface
4. Exploring eBook Recommendations from Regulation Of The Lactase Gene Answer Key
 - Personalized Recommendations
 - Regulation Of The Lactase Gene Answer Key User Reviews and Ratings
 - Regulation Of The Lactase Gene Answer Key and Bestseller Lists
5. Accessing Regulation Of The Lactase Gene Answer Key Free and Paid eBooks

- Regulation Of The Lactase Gene Answer Key Public Domain eBooks
 - Regulation Of The Lactase Gene Answer Key eBook Subscription Services
 - Regulation Of The Lactase Gene Answer Key Budget-Friendly Options
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 - ePub, PDF, MOBI, and More
 - Regulation Of The Lactase Gene Answer Key Compatibility with Devices
 - Regulation Of The Lactase Gene Answer Key Enhanced eBook Features
 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Regulation Of The Lactase Gene Answer Key
 - Highlighting and Note-Taking Regulation Of The Lactase Gene Answer Key
 - Interactive Elements Regulation Of The Lactase Gene Answer Key
 8. Staying Engaged with Regulation Of The Lactase Gene Answer Key
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Regulation Of The Lactase Gene Answer Key
 9. Balancing eBooks and Physical Books Regulation Of The Lactase Gene Answer Key
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Regulation Of The Lactase Gene Answer Key
 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
 11. Cultivating a Reading Routine Regulation Of The Lactase Gene Answer Key
 - Setting Reading Goals Regulation Of The Lactase Gene Answer Key
 - Carving Out Dedicated Reading Time
 12. Sourcing Reliable Information of Regulation Of The Lactase Gene Answer Key
 - Fact-Checking eBook Content of Regulation Of The Lactase Gene Answer Key
 - Distinguishing Credible Sources
 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development

- Exploring Educational eBooks

14. Embracing eBook Trends

- Integration of Multimedia Elements
- Interactive and Gamified eBooks

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