GENE SUITE

Lesson B: What Can Pseudogenes Tell Us About Common Ancestry?

BACKGROUND

Introduction

The human GULO gene is an example of a **pseudogene**, a DNA sequence that is similar to that of a known gene, but that does not yield the expected gene product. Pseudogenes can occur by a gene becoming permanently inactivated, as in the GULO example, or by gene duplication followed by inactivation. (Still another type of pseudogene is a DNA sequence that resembles just the mRNA for a particular protein. These **processed pseudogenes** may occur by reverse transcription of mRNA and insertion of the cDNA at some other site in the DNA, usually distant from the original gene.)

Pseudogenes and Common Ancestry

Suppose a mutation that inactivates a gene becomes common over generations so that eventually all the individuals (descendants of the original mutation carrier) carry only the inactive version of the gene. Since other mutations are possible over time, two distant descendants would not necessarily receive identical DNA sequences for that gene, but their sequences might still match for the original mutation that caused the gene to become inactive in the first place, or for other mutations carried in their "common ancestor".

Since deletions are not likely to occur independently at the same site and are highly unlikely to be "undone" by later mutations, **finding the same deletion in two different individuals or two different species is highly suggestive of common ancestry**. (This is in agreement with what is observed in "tracing" certain deletion mutations in human pedigrees.)

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Name_____ Date_____

WORKSHEET B: What Can Pseudogenes Tell Us About Common Ancestry?

B-1: The GULO Gene as an Example of Shared Deletions:

Given below is the alignment for the same part of the GULO gene that we examined in lesson A on Vitamin C, along with the corresponding sequences from 3 primate species that are incapable of synthesizing Vitamin C, the chimpanzee, the orangutan, and the crab-eating macaque.

Deletion

Human	**TACCTGGTGGGGGTACGCTTCACCTGGAG-GATGACATCCTACTGAGCCCC**
Chimpanzee	**TACCTGGTGGGGCTACGCTTCACCTGGAG-GATGACATCCTACTGAGCCCC**
Orangutan	**TACCCGGTGGGGGTGCGCTTCACCCAGAG-GATGACGTCCTACTGAGCCCC**
Macaque	**TAACCGGTGGGGGTGCGCTTCACCCAAGG-GATGACATCATACTGAGCCCC**
Rat	**TACCCCGTAGAGGTGCGCTTCACCCGAGGCGATGACATTCTGCTGAGCCCC**

1. Use a green colored pencil or highlighter to mark the positions in the sequence at which all five species are identical. Use a yellow colored pencil or highlighter to mark shared differences among the pseudogenes.

2. What do you **observe** about the similarities between the pseudogenes and the rat gene?

3. What do you **observe** about the pattern of similarities among the pseudogenes?

4. What do you **observe** about the differences among the pseudogenes?

5. How would an evolutionary biologist **explain** the **similarities between** the pseudogenes and the rat gene?

6. How would an evolutionary biologist **explain** the pattern of **similarities** among the pseudogenes?

7. How would an evolutionary biologist **explain** the **differences** among the pseudogenes?

B-2: The Hemoglobin Beta Pseudogene as an Example of Shared Deletions

Adult hemoglobin consists of two alpha chains and two beta chains per molecule. In addition to the beta-chain gene on chromosome 11, humans have a nearby similar but inactive sequence, the hemoglobin beta pseudogene. (In naming genes, the Greek letter **psi** is used to designate pseudogenes, so the hemoglobin beta pseudogene is also referred to as the **psi beta gene**.

Given below are the sequences in the human, gorilla, and chimpanzee pseudogenes that correspond to the first 25 codons of the functional human gene. (Since all 25 could not be listed on one line, the first 13 are listed together in the first set of four lines, and then the next 12 are listed together in the second set.)

Codon numbers	1	2	3	4	5	6	7	8	9	10	11	12	13
Human Psi Beta	**GTA**	**GTG**	**CAT**	**TTC**	**ACT**	**GCT**	**GAC**	**AAG**	**AAG**	**GCT**	**GCT**	**GCC**	**ACC**
Gorilla Psi Beta	**GTA**	**GTG**	**CAT**	**TTC**	**ACT**	**GCT**	**GAC**	**AAG**	**AAG**	**GCT**	**GCT**	**GCC**	**ACC**
Chimp Psi Beta	**GTA**	**GTG**	**CAT**	**TTC**	**ACT**	**GCT**	**GAC**	**AAG**	**AAG**	**GCT**	**GCT**	**GCC**	**ACC**
Human Beta	**ATG**	**GTG**	**CAC**	**CTG**	**ACT**	**CCT**	**GAG**	**GAG**	**AAG**	**TCT**	**GCC**	**GTT**	**ACT**

Codon numbers	14	15	16	17	18	19	20	21	22	23	24	25
Human Psi Beta	**AGC**	**CTG**	**TGA**	**AGC**	**AAG**	**GTT**	**AAG**	**GT-**	**GAG**	**AAG**	**GCT**	**GGA**
Gorilla Psi Beta	**AGC**	**CTG**	**TGA**	**AGC**	**AAG**	**GTT**	**AAG**	**GT-**	**GAG**	**AAG**	**GCT**	**GGA**
Chimp Psi Beta	**AGC**	**CTG**	**TGA**	**AGC**	**AAG**	**GTT**	**AAG**	**GT-**	**GAG**	**AAG**	**GCT**	**GGA**
Human Beta	**GCC**	**CTG**	**TGG**	**GGC**	**AAG**	**GTG**	**AAC**	**GTG**	**GAT**	**GAA**	**GTT**	**GGT**

Note that the pseudogenes all have the same sequence for the first codon and that this sequence is **not** the typical **start codon**, ATG. This change alone could prevent normal gene expression.

8. Use a yellow highlighter or colored pencil to mark the **shared** differences between the pseudogenes and the functional human gene. How many of the functional human beta nucleotides are different in the psi beta sequences? What percentage of the total is that?

____ differences out of _____ total bases = _____% different.

9. There are two other shared differences (other than the lack of a start codon) that would each be expected to prevent normal expression. Identify them! (Hint: Check each altered codon against the Genetic Code Chart.)

10. What percentage of the nucleotides are the **same** in all four sequences? How would an evolutionary biologist explain the similarities between the human pseudogene and the functional human gene?

11. How would an evolutionary biologist explain the pattern of similarities among the pseudogenes?

12. How would an evolutionary biologist explain the differences among the pseudogenes?

13. Why is the shared deletion an especially strong indication of common ancestry?