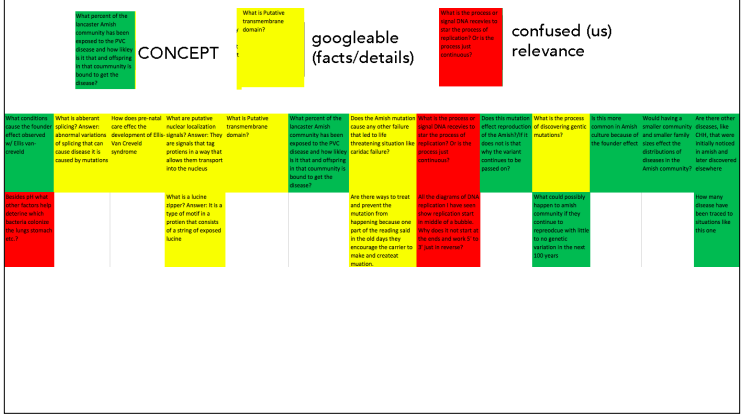
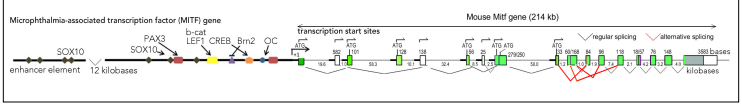
coreBio II - genetics: Monday 21 Jan. 2018

hand in questions

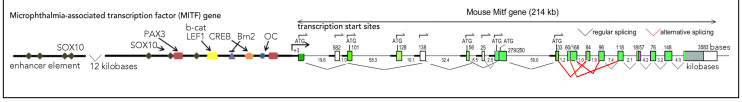


Concept mapping:

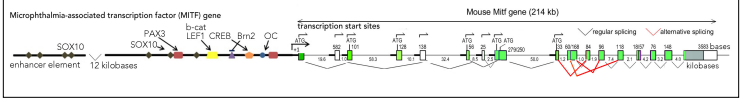
1. DNA (what features relevant to genes, how do the impact organization of genes)



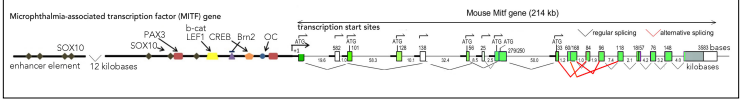
2. what is a gene (in terms of DNA/chromosome)

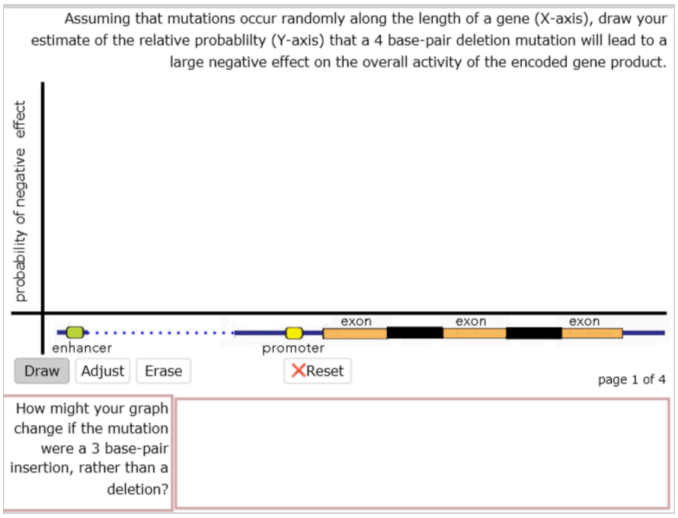


3. eukaryotic gene expression (features), comparison to prokaryotes.



4. gene products (functions)





Questions to answer and ponder:

178. How are transcription and translation similar, how are they different?

179. Within a gene, what signals and signal binding proteins are involved in gene expression? make a diagram.

180. How would having two copies of a gene (in a diploid cell) alter the behavior the cell?

Ellis-van Creveld syndrome and the Amish

Victor A. McKusick

Institute of Genetic Medicine, Johns Hopkins Hospital, Baltimore, Maryland 21287, USA. e-mail: mckusick@peas.welch.jhu.edu

Genetic studies often involve the cooperation of large numbers of affected persons and their families. The discovery of the gene that, when mutated, causes a form of dwarfism (Ellis-van Creveld syndrome) has been accelerated through a collaborative effort between geneticists and the Old Order Amish, of Lancaster County, Pennsylvania.

Aidan leads!

Exac Browser: EVC

Ellis-van Creveld syndrome and the Amish

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Originally described in 1940 (ref. 2) by paediatricians Richard Ellis and Simon Van Creveld, EvC syndrome is an autosomal recessive disorder, involving postaxial polydactyly of the hands (see figure), short stature with shortening especially of the forearms and lower legs and, in at least half of all cases, congenital heart malformation.

The mutation in the Amish of Lancaster County, Pennsylvania, in whom the disorder occurs at unprecedentedly high frequency, is predicted to cause aberrant splicing. It occurs in the fifth nucleotide of intron 13 of a novel gene, *EVC*, that is predicted to encode a protein containing a leucine zipper, three putative nuclear localization signals and a putative transmembrane domain.

Exac Browser: EVC

next:

Wednesday 24 January	4. How do allelic differences / mutations influence gene function?	**Read**: Chapter 11 pp. 229-231