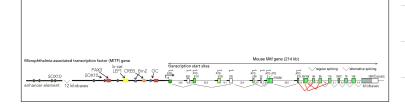


Concept mapping:

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



2. what is a gene (in terms of DNA/chromosome)	
Microphthalmia-associated transcription factor (MITF) gene Mouse Mit gene (214 kb) Local transcription start sites	
PAXS LEFT CREB BITS OC TO THE PASS OF THE	
3. eukaryotic gene expression (features),	
comparison to prokaryotes.	
Microphthalmia-associated transcription factor (MITF) gene Mouse Mild gene (214 kb)	
PAX3 LEFT CREB Brn2 QC Transcription start sites we produce the part of the pa	
4. gene products (functions)	
Microphthalmia-associated transcription factor (MITF) gene University of the Committee of t	
NINODHIS	

probability of negative	suming that mutations occur randomly along the length of a gene (X-axis), draw your to of the relative probability (Y-axis) that a 4 base-pair deletion mutation will lead to a large negative effect on the overall activity of the encoded gene product.	
enhancer promoter Draw Adjust Erase XReset page 1 of 4	Adjust Erase XReset page 1 of 4	
How might your graph change if the mutation were a 3 base-pair insertion, rather than a	the mutation a 3 base-pair rather than a	
deletion?	deletion?	

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

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