

Practice/Sample questions

Many birth defects are classified as a syndrome (e.g. Waardenburg Syndrome and Grieg cephalopolydactyly Syndrome), even when they are cause by a mutation in a single gene.

What is a syndrome?

***A group of phenotypes that consistently occur together
Symptoms characterizing a condition.***

How can a syndrome be caused by a single mutated gene??

One gene often has multiple roles at various sites during development, and its loss will this affect multiple structures in an organism

The DNA sequence of four related species of animals Hs, Cs, Gs and Os is well conserved. In the table below the percentage DIFFERENCE is shown

	H	C	G	O
H		1.7	1.8	3.3
C	1.7		1.8	3.3
G	1.8			3.3
O	3.3	3.3	3.3	

Intensive research of the fossil record has shown that the last common ancestor of Hs and Cs lived 5.1 million years ago. Based on this information give your best estimate when the last common ancestor of Cs and Gs lived, and when the last common ancestor of Hs and Os lived. (10 points for one correct answer, 15 for two)

***1% difference in DNA sequence is accumulated over 3.0 million years.
Last common ancestor of Cs and Gs: 5.4 million years ago
Last common ancestor of Hs and Os: 9.9 million years ago***

If two people with aniridia (because they are Pax6 heterozygotes, a phenotype due to haploinsufficiency) have children, how big is the chance that at birth these children have aniridia?

66%/ or 2/3. The distribution will be 1:2:1 at conception, but the homozygous pax6 mutants will not develop to birth; at birth the distribution will be 1:2:0

Here is another calico cat question. These must be the most interrogated animals in genetics....

Calico cats get their colorful appearance due to X-chromosome inactivation. When you clone a calico cat using nuclear transplantation, there are three potential coat color outcomes.

- 1/ the clone will be calico
- 2/ the clone will be white and orange
- 3/ the clone will be white and grey/black

If the clone is a calico, explain what has happened to the inactivated X-chromosome after the nuclear transplantation process.

The inactivated X-chromosome is reactivated and subsequently randomly inactivated giving rise to a tortoiseshell/calico

If after many attempts the cloned cats are either orange or grey/black, what would be your conclusion regarding X chromosome inactivation?

The same x-chromosome is inactivated in all cells of the clone, so it was never activated (completely) during the cloning process

HIV (the AIDS virus) carries its genetic information in the form of RNA. After a successful infection of a host cell the viral genetic information can be found integrated in the nuclear DNA as a provirus. This means that at some point during this process the RNA carried in the virus is transcribed into DNA. What is the name of the enzyme that catalyses this reaction?

reverse transcriptase or RNA dependent DNA polymerase

Newly synthesized viral particles will contain RNA carrying the viral genome. What enzyme has transcribed the proviral DNA generating the HIV genomic RNA?

RNA polymerase or DNA dependent RNA polymerase (PolIII is OK as well)