

You may use various Internet sources to answer any of the questions, but please cite any sources that you use if they are not ones that I suggest.

Part II: Hemophilia

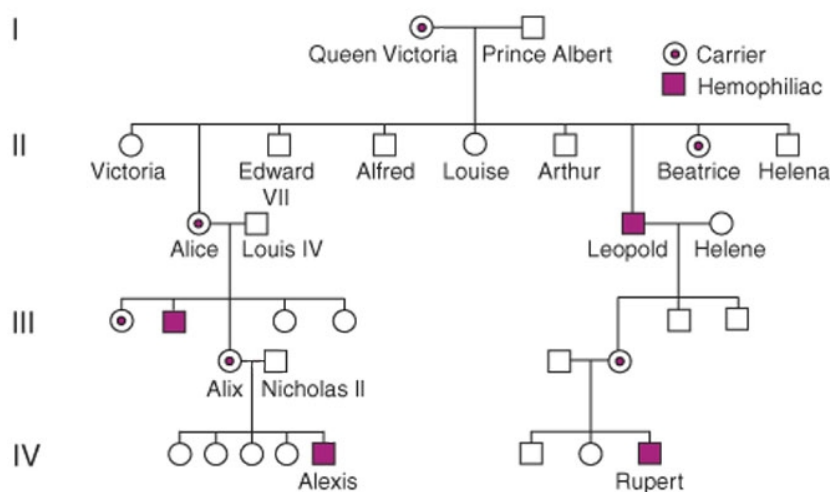
The pedigree chart below comes from the Module powerpoint lecture notes.

Use the following source for the questions 2: <http://www.ncbi.nlm.nih.gov/pubmed/20557352> (You won't be able to access the entire article, but the abstract will give you the information you need to answer the questions.)

- Using your knowledge from Module 4, on what chromosome is the gene that causes hemophilia?
 - Answer: X Chromosome
- Describe the mutation that apparently caused hemophilia in Alix, (and probably all of the European families that had hemophilia).
 - Answer: Hemophilia B is the type of hemophilia in Alix and most likely all the European families had. Hemophilia B is a mutation caused by a substitution in the splice acceptor site of 4 in the F9 gene.
- Using your knowledge from Module 7, describe how the mutation you described in #10 could result in a faulty gene product.
 - Answer: The occurrence of this type of mutation causes a person to lack one of the clotting factors, which makes the body cells unable to clot properly to prevent excessive bleeding out. Some genes if missing they will not combine to their usual structures, which changes their shape, changes their gene and ultimately changes the genes functionality. Due to genes being transformed into other genes caused by mutations this transformation can create diseases such as hemophilia.
- Again, using your knowledge from Module 4, give the genotype for a carrier of hemophilia.
 - Answer: $X^h X^H$
Source: <https://www.vet.cornell.edu/animal-health-diagnostic-center/laboratories/comparative-coagulation/clinical-topics/hemophilia-a>

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(a) X-linked recessive: Hemophilia



Part III: Molecular Analysis of People in a Mass Grave

Use the following source to help you answer the following questions:

<http://journals.plos.org/plosone/article?id=10.1371/journal.pone.0004838> (You should be able to access the entire article.)

5. Mitochondrial DNA testing was also done on both Nicholas II and Alix. Why was information from Alix's, but not Nicholas', mitochondrial DNA used to identify three females as belonging to Alix?
 - a. Answer: Hemophilia is only inherited from a woman, so only the mother could pass it on. Also only the females would be recessive carriers of the gene.
6. HRH Prince Philip, the Duke of Edinburgh, provided mitochondrial DNA used to identify Alix and her three daughters.
 - a. Why was his mitochondrial DNA used?
 - i. Answer: All share the same maternal lineage, so they all will have the same mitochondrial DNA from Queen Victoria.
 - b. Who was the HRH Prince Philip, the Duke of Edinburgh in today's world? Do you ever hear of his grandchildren (in magazines while you are waiting to check out of a store)?
 - i. Answer: Prince Philip was the husband of the late Queen Elizabeth II. Yes, I have seen their grandchildren on magazines and on the news from time to time.
7. Who was missing from the mass grave?
 - a. Answer: Alexei and one of his sisters Maria/Anastasia
8. The Duke of Fife and Princess Xenia provided mitochondrial DNA used to identify Nicholas. One of these is a female and another is a male. Does that matter? What general statement can you make about their genetic relationship to Nicholas.
 - a. Answer: This does not matter because they are all maternal relatives of Nicholas.
9. What was discovered in the mitochondrial DNA of Nicholas that was not identified in either the Duke of Fife or Princess Xenia?
 - a. Answer: In the mtDNA of Nicholas a single point of heteroplasmy was found at position 16169 (C/T=Y).
10. What is the term given to the existence of two (or more) genetically different mitochondria in the cell?
 - a. Answer: Heteroplasmy