

PCSK9 Enzyme and LDL

Researchers interested in genetic factors affecting susceptibility to cardiovascular disease examined the DNA of 15,000 individuals. They found that 3% of the individuals had a mutation that inactivated one copy of the gene for PCSK9 (Proprotein convertase subtilisin/kexin type 9), a liver enzyme. Because mutations that increase the activity of PCSK9 are known to increase levels of LDL cholesterol in the blood, the researchers hypothesized that inactivating mutations in this gene would lower LDL levels.

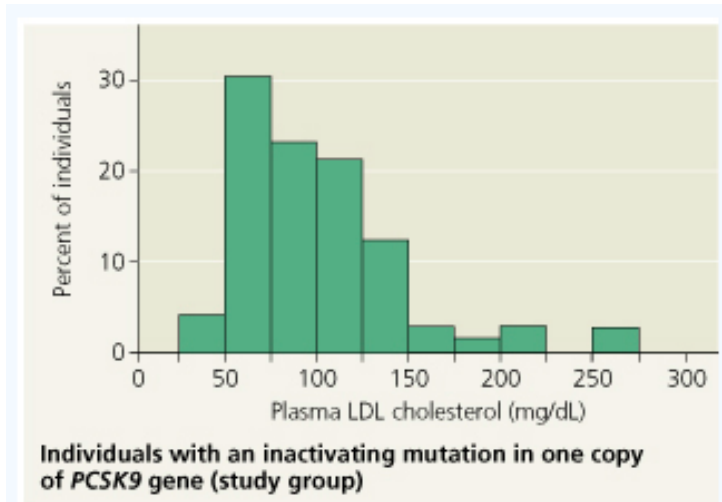
Q1. a) What is a mutation?

b) What is LDL?

c) Why is LDL correlated to heart disease?

Researchers measured LDL cholesterol levels in the blood plasma of 85 individuals with one copy of the PCSK9 gene inactivated and from 3,278 individuals with two functional copies of the gene.

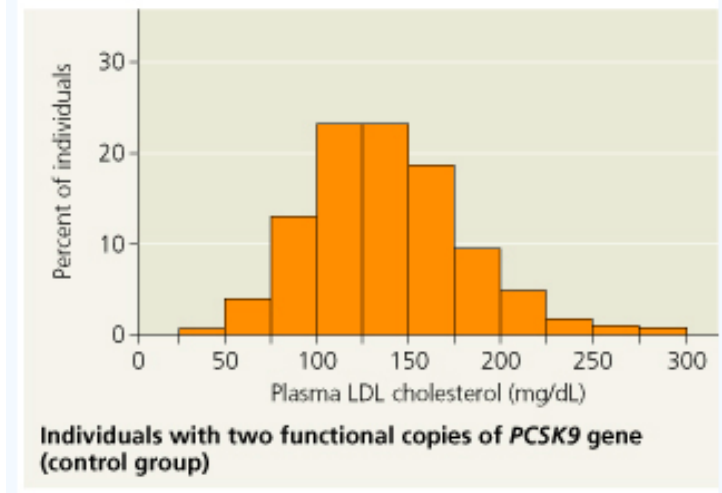
Q2. Identify the study (or experimental) group and the control group in this experiment.



The results are shown in the histograms. In a histogram, the variable on the X-axis is grouped into ranges. The height of each bar in the histograms shows the percentage of individuals that fall into a range.

Q3. a) What percentage of individuals in the study group had an LDL level below 100 mg/dL?

b) What percentage of individuals in the control group had an LDL level below 100 mg/dL?



Q4. Do the data in the two histograms support the researchers' hypothesis? Justify your response.

Q5. Provide an explanation for the fact that the two histograms overlap as much as they do.

Q6. Imagine instead of making histograms, the researchers had simply compared the range of LDL found in the two groups. How would their conclusion have been different?

Q7. How would you compare the risk of cardiovascular disease between the study group and the control group?

Q8. a) Consider two individuals with a plasma LDL level of 160 mg/dL, one from the study group and one from the control group. What do you predict regarding their relative risk for cardiovascular disease?

b) Identify a factor that might affect the LDL level of a person.