

2. (20) Garrod:

Give the name of the disease that Garrod studied, explain the symptoms, and describe the metabolic basis of the disease.

What conclusion did Garrod reach about the nature of genes based on his research?

Describe the evidence and Garrod's reasoning for drawing this conclusion.

Name two other genetic diseases that are **related** to Garrod's disease and state why they are related. Describe their symptoms and underlying metabolic problem.

Name one other genetic disease that is **unrelated** to Garrod's disease but has a similar, underlying mechanism. Describe its symptoms and underlying metabolic problem and state why the disease is similar to Garrod's/

4. (20) Fill in the blanks
- a What is the term for de-condensed chromosomes? _____
 - b What is the term for DNA even in interphase never decondenses? _____
 - c What is the metabolic status of the DNA in "b" _____
 - d What is the central constricted region of a chromosome called? _____
 - e What term describes a chromosome whose central constriction is skewed towards one end, but is not at the end? _____
 - f Name one stain that is used for banding chromosomes _____
 - g What is the name of the procedure in which metaphase chromosomes are isolated and examined for aberrations or other defects? _____
 - h What is the role of the drug colchicine in the procedure described in g? _____
 - i What does the designation: 5p23.2 mean? _____
 - j What does the designation: 21q22ter+ mean? _____

