

## Gene control

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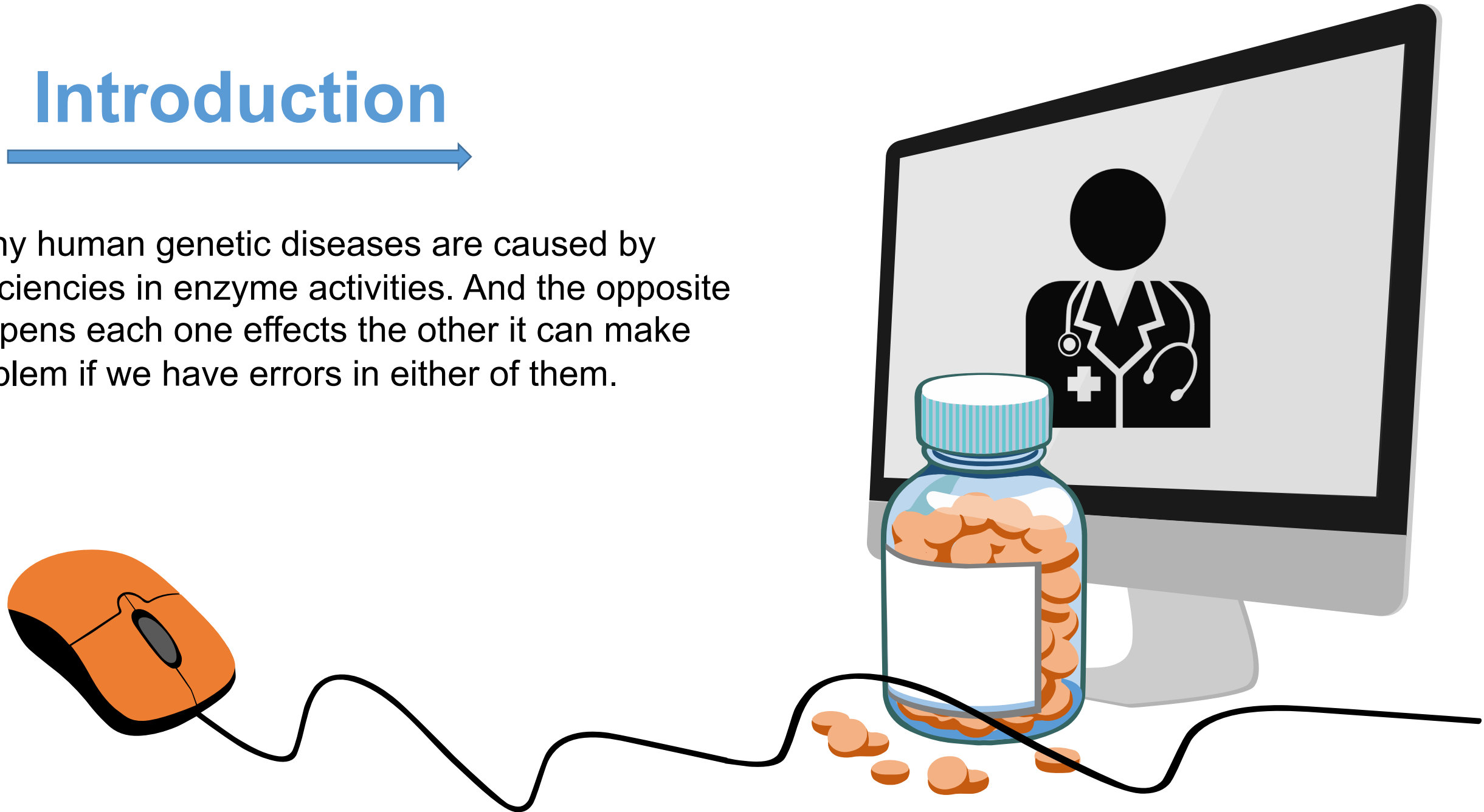
# ILOS:

- 01 Illustrate the history of gene control discovery**
- 02 Define Inborn errors of metabolism**
- 03 Explain diseases and symptoms of inborn errors of metabolism**
- 04 Mention the diagnosis and various test**

# Introduction



Many human genetic diseases are caused by deficiencies in enzyme activities. And the opposite happens each one effects the other it can make problem if we have errors in either of them.



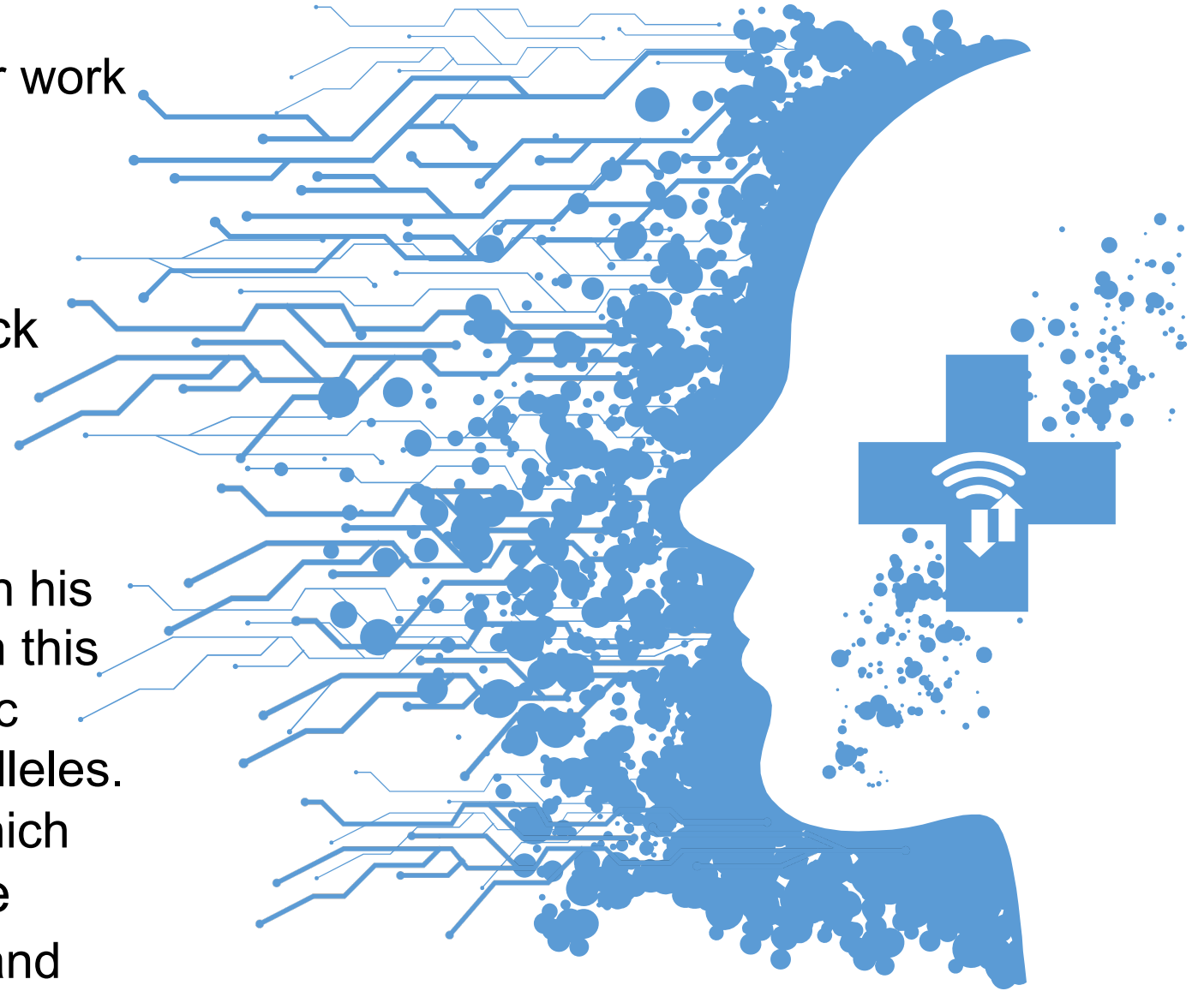
# History of gene control discovery

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In 1902 Archibald Garrod is known for work that prefigured the based on his studies on the nature and inheritance of [alkaptonuria](#) a human disease characterized by urine that turns black upon exposure to the air and by a tendency to develop arthritis later in life.

In 1909 Hi published a book entitled with his main text *Inborn Errors of Metabolism* In this book Garrod documented how metabolic abnormalities can be traced to mutant alleles.

["one gene-one enzyme" hypothesis](#) Which were developed by researchers George Beadle and Edward Tatum in 1942, and upgrade to on gene one polypeptide.

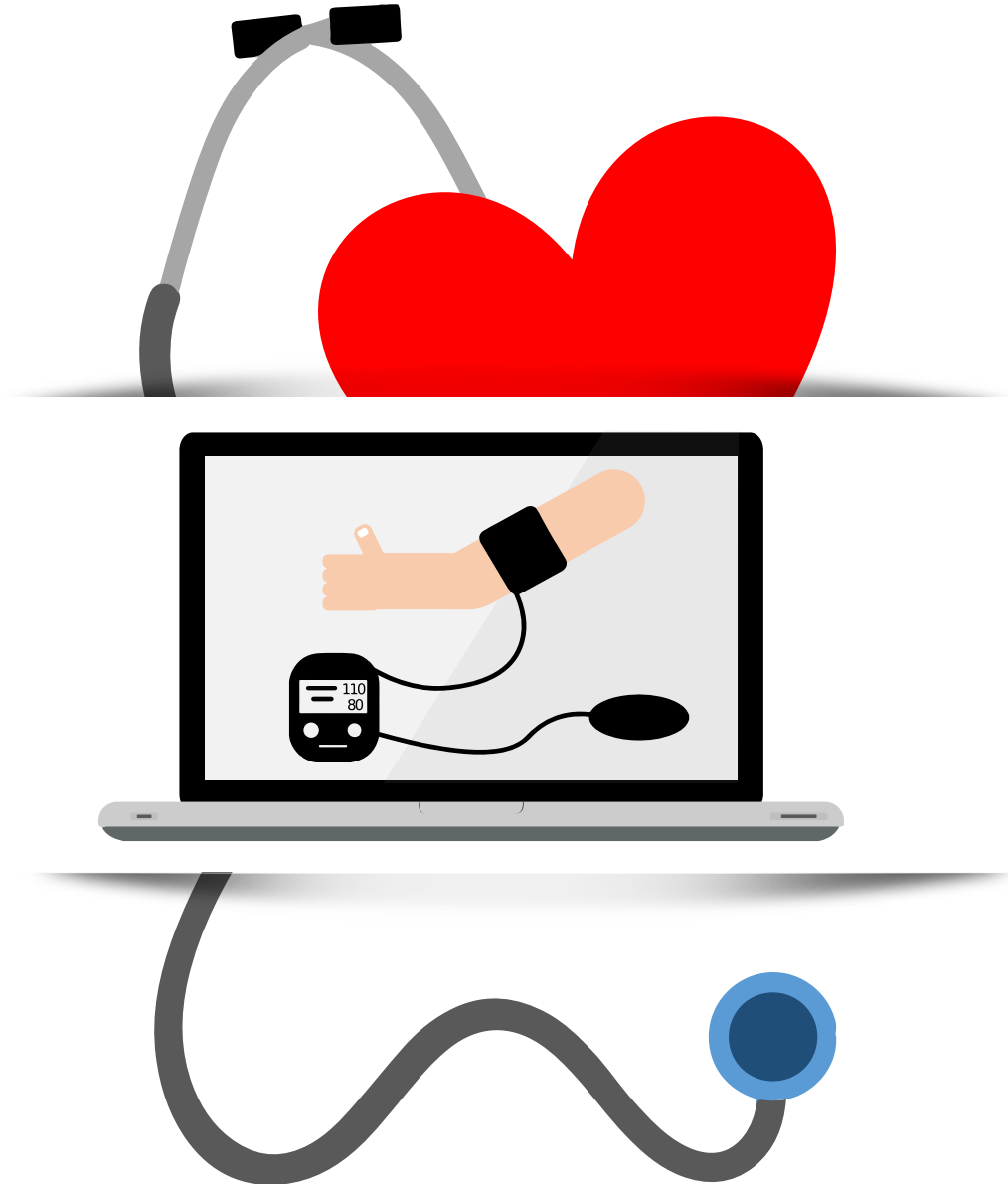




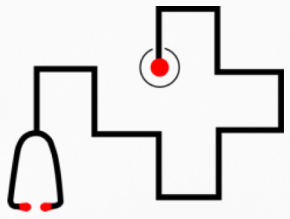
# Inborn errors of metabolism

Inborn errors of metabolism are rare genetic disorders that affect the body's metabolism, which the body cannot properly turn food into energy. The disorders are usually caused by defects in specific proteins (enzymes) that help (metabolize) parts of food.

A food product that is not broken down into energy can build up in the body and cause a wide range of symptoms. Several inborn errors of metabolism cause developmental delays or other medical problems if they are not controlled.



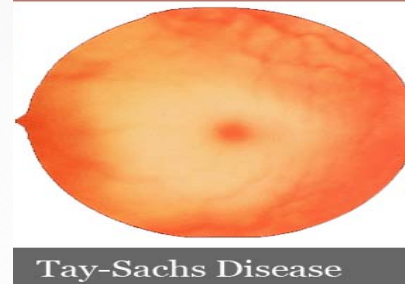
# Explain diseases and symptoms of inborn errors of metabolism



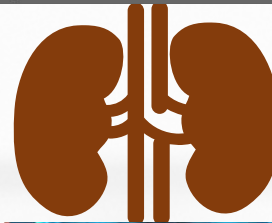
**Niemann-Pick disease NPD**  
Liver enlargement  
Nerve damage  
Liver, spleen and brain damage



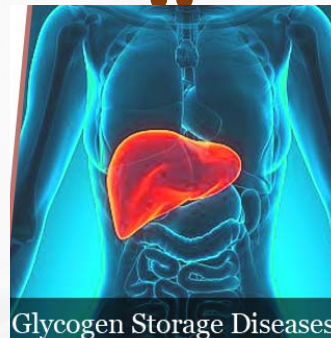
**Tay-sachs disease (TCD)**  
Liver enlargement  
Weakness and sever Nerve damage



**Urea Cycle Disorder**  
Kidney failure and pain in kidney



**Glycogen storage disease Disorder**  
Weakness and muscle pain  
Low or high blood suger



# Diagnosis



Most tests are done to a urine or blood sample to detect amino acids or other substances that might be present in the body, or DNA specific test

● Urine organic acid analysis

● Urine purine and pyrimidine analysis

● Plasma acylcarnitine analysis

● Specific diagnostic tests (or focused screening for a small set of disorders): LIKE  
Tissue biopsy: liver, muscle, brain, bone marrow



# summary



**History discovery of gene control and two hypotheses  
What is Inborn errors of metabolism**

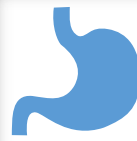


**diseases and symptoms of inborn errors of**

**metabolism**

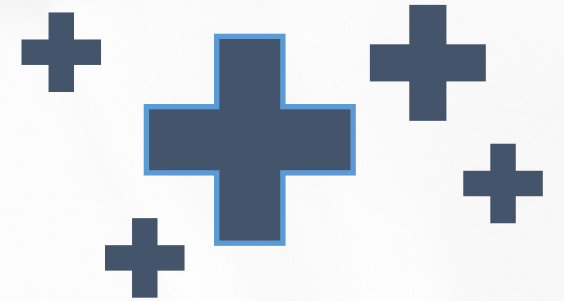


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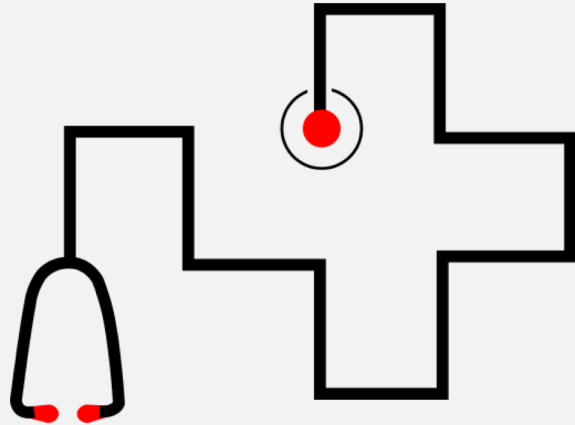




# Reference



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Thank you