Phenylketonuria (PKU)

An example of single gene disorders
Topics to be discussed in this session:

1. What is PKU?
2. What is IEM?
3. What is the basic defect of PKU?
4. What are the biochemical abnormalities?
5. Is there any other cause of PKU?
6. What is the age group at first presentation?
7. What is the main problem(s) in patients with PKU?
8. What are the clinical features of PKU?
9. How to manage such cases?
10. Does therapy exist for this condition?
11. Is there any prenatal diagnostic method?
12. Is prevention possible?
Session objectives

At the end of this SGL session, all students must be able to:

1. Define PKU
2. Describe the basic defect in PKU
3. Classify PKU (if any) by type.
4. Describe in detail the biochemical abnormalities
5. Relate the clinical features to the biochemical defects
6. Describe the steps in management of such patients
7. Apply the understanding of the biochemical defects in diagnosis, screening and treatment of PKU
8. Describe the prenatal diagnostic method (if available)
9. Recognize the preventive measures applicable in PKU
10. Analyze future potential problems related to PKU
What is Phenylketonuria (PKU)?
What is the Inborn Error of Metabolism?

(IEM / IEOM / IBEM)
What is the basic defect in PKU?
Is there any other cause of elevated levels of Phenylalanine apart from PKU?

i.e. non PAH mutations → ↑ Phe
What are the main problem(s) in patients with PKU?
What is the age group at first presentation?
Incidence of PKU
What are the clinical features of PKU?
Why do these clinical features occur in PKU patients?

i.e. what is the biochemical basis of these signs?
How to manage such cases?
Does therapy exist for this condition?
Is there any prenatal diagnostic method?
Is prevention possible?
Maternal phenylketonuria
THANK YOU