<u>Tutorial № 4</u> X-linked pattern of inheritance: pedigree criteria, genetic risks, general phenotypic features, examples. Carrier detection: obligate carriers, screening for carrier state

I. Main characteristics of X-linked pattern of inheritance

II. X-recessive pattern of inheritance

1. Pedigree criteria, risks
2. Clinical features
3. Cases of affected females
4. Roll of specific factors
5. Examples
    - Duchenne/Becker muscular dystrophy
    - Haemophilia – A, B
    - Fragile X syndrome

III. X-dominant pattern of inheritance

1. Pedigree criteria, risks
2. Clinical features
3. Roll of specific factors
4. Examples
    - Familial hypophosphatemic /vitamin D-resistant rickets

IV. Definition of a carrier status, *obligate carriers*

1. Detection of *obligate carriers* by pedigree analysis in different type of inheritance.
    a) Autosomal dominant disorders
    b) Autosomal recessive disorders
    a) X-linked recessive disorders

2. Screening for carrier detection – disorders suitable for population carrier screening (Thalassaemias, sickle-cell disease, Tay-Sachs disease, Cystic fibrosis)