Teaching topics：Genetic disorders

Teaching outline：

A. Mutations
B. Mendelian Disorders:
  a. Transmission patterns of single-gene disorders
    1. Autosomal dominant disorders
    2. Autosomal recessive disorders
    3. X-linked disorders
  b. Biochemical and molecular basis of single-gene (Mendelian) disorders
    1.Enzyme defect
    2. Defects in receptors and transport systems
    3. Alterations in structure, function, or quantity of nonenzyme proteins
    4. Genetically determined adverse reactions to drugs
  c. Disorders associated with defects in structural proteins
    1. Marfan syndrome
    2. Ehlers-Danlos syndromes
  d. Disorders associated with defects in receptor proteins
    1. Familial hypercholesterolemia
  e. Disorders associated with defects in enzymes
    1. Lysosomal storage diseases
    2. Glycogen storage diseases
    3. Alkaptonuria
  f. Disorders associated with defects in proteins that regulate cell growth
    1.Neurofibromatosis: type 1 and 2
C. Disorders with Multifactorial Inheritance
D. Normal Karyotype
E. Cytogenetic Disorders
  a. Cytogenetic disorders involving autosomes
    1. Trisomy 21
    2. Other trisomies
    3. Chromosome 22q11.2 deletion syndrome
  b. cytogenetic disorders involving sex chromosomes
    1. Klinefelter syndrome
    2. Turner syndrome
    3. Hermaphroditism and pseudohermaphroditism
F. Single-Gene Disorders with Nonclassic Inheritance
  a. Triplet-repeat mutations – fragile-X syndrome
  b. Mutations in mitochondrial genes – Lever hereditary optic neuropathy
  c. Genomic imprinting
    1. Prader-Willi syndrome and Angelman syndrome
  d. Gonadal mosaicism

G. Molecular Diagnosis

H. Diagnosis of Genetic Diseases

   a. Direct gene diagnosis

   b. Indirect DNA diagnosis: linkage analysis