Hereditary disorders and developmental anomalies

congenital anomaly: a generic term for functional and morphological anomalies based on genetic and developmental abnormality
hereditary disease / gametopathy / embryopathy / fetopathy
malformation: grossly distinguishable morphological anomalies among congenital ones

- external malformation, organ malformation,
- single malformation, multiple malformation
- major malformation, minor malformation (clinical classification: fatal or life-threatening malformation labeled as major)
- anomalad or malformation syndrome
- simple(isolated) malformation, single monster
- complete symmetrical double malformations: considered malformed when conjoined
- craniopagus / cephalothoracopagus
- thracoomphalopagus / pygopagus
- incomplete symmetrical double malformations: partially duplicated on one side:
  - Janus malformation (monocephalus diprosopus), dicephalus, dipygus
- asymmetrical double malformations—acardius, parasite

malformation causing factors: hereditary factors, environmental factors
conundrum of craniofacial anomaly:
- environmental factors: radiation-induced microcephaly / rubella / thalidomide / organic mercury
- late childbearing

Chromosome aberration
  - Down's syndrome
  - Turner's syndrome

Genetics of diseases
  - fundamental knowledge of genetics
  - quantitative mutation
    - recessive homozygous
  - haploid insufficiency
loss of heterozygosity
imprinting
gene duplicating type
qualitative mutation
gain of function
dominant negative mutation
combined type: increase in triplet repeats
abnormality of regulatory region

Cause of diseases
hereditary factors and environmental factors

Environmental factors and diseases
A) Infectious agents
   concept of chronic infection
   relations with immunologic mechanism
   opportunistic infectious diseases: carinii pneumonia / candidiasis / cryptococcosis /
toxoplastic encephalosis / cytomegalovirus / atypical mycobacteriosis /
cryptosporidiosis / Entamoeba histolytica / PML

B) Chemicals
   1) smoke
   chronic bronchitis, pulmonary emphysema / laryngeal cancer, lung cancer /
   ischemic heart disease
   2) industry
   asbestos: pleural callosity, lung fibrosis, pleural mesothelioma
   aluminum: Alzheimer(s) disease
   3) alcohol
   chronic alcoholism / liver cirrhosis / Wernicke's encephalopathy

C) Physical influence
   1) radiation damage
   radiation syndrome: prodromal symptom and exposed dose / radiation burn / organ
damage
   genetic consequences / carcinogenesis
Modification of disease process due to therapy

A) Drug-induced organ damage
    lung damage: methotrexate / Iressa / Shousaikotou
B) Effect of radiotherapy
    histology of radiation effect/ side effect
C) Organ replacement
    prosthetic valves / synthetic blood vessel
    coronary-artery bypass
D) Organ transplantation
    blood stem cell transplantation, graft versus host disease, thrombotic microangiopathy