

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 /
toxoplasmic encephalitis / 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