**LECTURE SYLLABUS**

**(General medicine)**

**Intrinsic etiological factors**

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**Etiological factors of diseases**

Age

Gender

Immunity

Rhythmicity of physiological processes

Genetic factors

Genetic diseases

Constitution (asthenic, picnic, athletic)

Race differences (e.g. lactose-, alcohol intolerance)

**Age**

(see a special lecture)

Childhood

Senium

**Gender**

role of **estrogenes and progesterone**

cardiovascular system

neural system

metabolism & distribution of lipids

skeletal system

malignancies

patophysiology of atherosclerosis and heart coronary disease

incidence of the Parkinson’s and Alzheimer’s dis., cognitive decline

increases incidence of DM type II.

**Rhythms in biology**

(see a special seminary)

Subsecond

Seconds

Minutes

Hours

Days

Month(s)

Year

**Circadian (diurnal) rhythmicity**

Melatonin – production, kinetics, other functions

Intrinsic

Extrinsic

**Genetics**

inherited

congenital

acquired

gene therapy

Genetic intrinsic factors

Carried independently of DNA – epigenetic mechanisms

Driven by DNA – pure genetic disorders

**Epigenetics**

Överkalix Study

Sensitive periods for epigen. inheritance

Concept - Lamarck x Darwin

Molecular Basis of Epigenetics

Primary mechanisms

Diseases connected with epigenetics

Therapies Targeting Epigenetic Errors

**Genetic factors driven by DNA**

genotype x phenotype

Mutations

Mechanism – **substitution**

**- deletion**

**- insertion**…..of a nucleotide into DNA sequence

**in-frame** mut.

**frame-shift** mut.

Result - **neutral mutation**

**- amorphic or hypomorphic m.**

**- hypermorphic m.**

**- antimorphic m.**

**- neomorphic m.**

**Mechanisms of dominance/recessivity**

haplosuficiency

haploinsuficiency

gain of function

Shift from heterozygosity to homozygosity

**Inheritance**

Mendelian

Non-mendelian

Polygenic

Epistasis

Gene imprinting (epigenetics)

Extranuclear

Gonosomal – X, Y (Holandric)

**Mitochondrial diseases**

Prenat. diagnostics

Pedigree definition

Interpreting a Pedigree

What can you tell from a pedigree?

**Autosomal dominant disorders**

Familial hypercholesterolemia

Pathobiochemistry, types.

Huntington’s disease

Triplet character, transgeneration progression

Achondroplasia

Marfan syndrome

Malignant hyperthermia

**Autosomal incomplete dominant disorders**

Sickle Cell Anaemia

Mechanisms, epidemiology, relation to malaria

**Autosomal recessive diseases**

Cystic fibrosis

mechanisms

Phenylketonuria (PKU)

Pathobiochemistry of Phe, pathways to melanin/catecholamines/thyroxin

Mechanisms of neural damage

Patterns of risks between mother and fetus

Galactosemia

Glycogenoses

Lysosomal storage diseases

Mucopolysacharidoses

**Sex-linked inherited diseases**

pseudoautosomal regions on X and Y

pathology Y

**SRY - Sex-determining region Y protein** *(TDF -testis-determining factor)*

XX male sy.

Sex phenotype x genotype

SRY testing

Sex-determining region Y protein *(TDF -testis-determining factor)*

**X-linked diseases**

Haemophilia

Colour blindness

Muscle dystrophy (Duchenne disease)

**Chromosomal aberrations**

**Chromosomal structural abnormalities**

Fragile chromosome sy., Chromosome instability sy.

**Numerical abnormalities**

Trisomy 21 (Down’s syndrome)

Trisomy 18 (Edwards syndrome)

Trisomy 13 (Patau syndrome)

**Sex chromosomal disorders**

Klinefelter syndrome (47, XXY)

Turner syndrome (45, X0)