Genetics

System	Genetic Principles & Processes	Genetic Disorders
General Genetics	- Mendelian inheritance (dominant, recessive, X-linked) - Chromosomal abnormalities (deletions, duplications, translocations) - Population genetics (Hardy-Weinberg equilibrium) - Epigenetics and imprinting - Multifactorial inheritance - Genetic testing and counseling	- Down syndrome (trisomy 21) - Turner syndrome (45,X) - Klinefelter syndrome (47,XXY) - Fragile X syndrome - Prader-Willi and Angelman syndromes (imprinting disorders)
Hematopoietic & Lymphoreticular	- Hemoglobin gene structure and regulation - Genetic basis of blood group antigens - Clonal hematopoiesis and genetic mutations in blood cancers	- Sickle cell disease (HbS mutation) - Thalassemia (alpha and beta) - Hemophilia A and B (factor VIII and IX deficiency) - Hereditary spherocytosis - Von Willebrand disease
Central & Peripheral Nervous	- Genetic regulation of neuronal development - Neurotransmitter receptor gene mutations - Mitochondrial DNA inheritance	- Huntington disease (trinucleotide repeat disorder) - Alzheimer's disease (APOE4 risk allele) - Parkinson's disease (LRRK2, PARK genes) - Charcot-Marie-Tooth disease - Rett syndrome (MECP2 mutation)
Skin & Connective Tissue	- Collagen and elastin gene expression - Keratin gene mutations	- Ehlers-Danlos syndrome (collagen defects) - Marfan syndrome (fibrillin-1 mutation) - Osteogenesis imperfecta (COL1A1, COL1A2) - Albinism (tyrosinase mutation) - Epidermolysis bullosa
Musculoskeletal	- Regulation of bone growth and remodeling genes - Muscle protein gene expression	- Achondroplasia (FGFR3 mutation) - Duchenne and Becker muscular dystrophy (dystrophin gene mutations) - McArdle disease (myophosphorylase deficiency) - Osteopetrosis (carbonic anhydrase II deficiency)
Respiratory	- Genes controlling lung development - Genetic basis of airway diseases	- Cystic fibrosis (CFTR gene mutation) - α1-Antitrypsin deficiency - Primary ciliary dyskinesia (Kartagener syndrome) - Pulmonary arterial hypertension (BMPR2 mutation)
Cardiovascular	- Genetic regulation of cardiac development - Lipid metabolism genes (LDL receptor, APOB)	- Familial hypercholesterolemia - Hypertrophic cardiomyopathy (MYH7, MYBPC3 mutations) - Marfan syndrome (FBN1 mutation) - Long QT syndrome (ion channel mutations)
Gastrointestinal	- Genetic regulation of digestive enzymes and absorption - Gut microbiome interactions with host genetics	- Hereditary hemochromatosis (HFE mutation) - Wilson disease (ATP7B mutation) - Celiac disease (HLA-DQ2, HLA-DQ8) - Lynch syndrome (DNA mismatch repair gene mutations)
Renal/Urinary	- Kidney development and genetic signaling pathways - Water and electrolyte balance gene regulation	- Polycystic kidney disease (PKD1, PKD2 mutations) - Alport syndrome (COL4A5 mutation) - Bartter and Gitelman syndromes (ion transporter defects) - Congenital nephrotic syndrome

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System	Genetic Principles & Processes	Genetic Disorders
Reproductive	- Genetic determination of sex and gonadal differentiation - Hormone receptor gene mutations	- Turner syndrome (45,X) - Klinefelter syndrome (47,XXY) - Androgen insensitivity syndrome - Polycystic ovary syndrome (multifactorial inheritance) - Congenital adrenal hyperplasia (CYP21A2 mutation)
Endocrine	- Hormone synthesis and receptor gene regulation - Genetic basis of metabolic regulation	- Multiple endocrine neoplasia (MEN1, RET mutations) - Diabetes mellitus Type 1 (HLA association) - Congenital hypothyroidism (TSH receptor mutations) - Pseudohypoparathyroidism (GNAS mutation)