Genetic Conditions List

These Genetic conditions were selected for one or more of the following reasons:

- they are commonly encountered in practice
- they illustrate key concepts in human genetics
- they illustrate the knowledge and skills required to collaborate and communicate between laboratory genetic disciplines and / or within the health care community

Category	Disorder
Cancer Genetics	Tuberous sclerosis
Cancer Genetics	Hereditary breast cancer
Cancer Genetics	Chronic myelogenous leukemia (CML)
Cancer Genetics	Familial adenomatous polyposis
Cancer Genetics	Hereditary non-polyposis colon cancer (HNPCC)
Cancer Genetics	Li-Fraumeni syndrome
Cancer Genetics	Retinoblastoma
Clinical Genetics	Duchenne/Becker Muscular Dystrophy
Clinical Genetics	Osteogenesis Imperfecta
Clinical Genetics	Long QT syndrome
Clinical Genetics	Marfan syndrome
Clinical Genetics	Neurofibromatosis typeII
Clinical Genetics	Neurofibromatosis type I
Clinical Genetics	Smith-Lemli-Opitz syndrome
Clinical Genetics	Noonan syndrome
Clinical Genetics	Charge syndrome
Clinical Genetics - Developmental	FGFR-related disorders (Craniosynostosis...)
Clinical Genetics - Hematology	Factor V Leiden thrombophilia
Clinical Genetics - Hematology	G6PD deficiency
Clinical Genetics - Hematology	Hemoglobinopathies (Sickle cell anemia, Alpha / Beta Thalassemia)
Clinical Genetics - Hematology	Hemophilia A / B
Clinical Genetics - Hematology	Hemochromatosis
Clinical Genetics - Sexual development	SRY translocation
Clinical Genetics - Sexual development	Turner syndrome
Clinical Genetics - Sexual development	Androgen insensitivity syndrome
Clinical Genetics - Sexual development	21-Hydroxylase deficiency
Complex Inheritance	Alzheimer disease
Complex Inheritance	Congenital hearing loss
Complex Inheritance	Diabetes mellitus (insulin vs non-insulin dependent)
Cytogenetics	Cri du chat syndrome
Cytogenetics	Pallister-Killian syndrome
Cytogenetics	Triploidy
Cytogenetics	Trisomy 13
Cytogenetics	Trisomy 18

Category	Disorder
Cytogenetics	Trisomy 21
Cytogenetics	Klinefelter syndrome
Cytogenetics - Chromosome instability	Fanconi anemia
Cytogenetics - Chromosome instability	Ataxia-telangiectasia
Cytogenetics - Microdeletion Syndrome	Williams syndrome
Cytogenetics - Microdeletion Syndrome	22q11 deletion syndrome
Metabolics	Acute intermittent porphyria
Metabolics	Alpha-1 antitrypsin deficiency
Metabolics	Canavan disease
Metabolics	Gaucher disease
Metabolics	Homocystinuria (CBS deficiency)
Metabolics	Hurler syndrome
Metabolics	I cell disease
Metabolics	MCAD deficiency
Metabolics	Mitochondrial DNA mutations (MERRF /MELAS / LHON, mtDNA deletion syndromes)
Metabolics	Ornithine transcarbamylase (OTC) deficiency
Metabolics	Peroxisome biogenesis disorder (Zellweger)
Metabolics	Phenylketonuria
Metabolics	Pompe disease
Metabolics	Tay Sachs Disease
Metabolics	Tyrosinemia type I
Metabolics	X-linked Adrenoleukodystrophy
Metabolics	Wilson disease
Metabolics / Cytogenetics	Hyperprolinemia type I
Molecular Genetics	Cystic fibrosis
Molecular Genetics	Achondroplasia
Molecular Genetics - triplet repeat	Huntington disease
Molecular Genetics - triplet repeat	Fragile X
Molecular Genetics - triplet repeat	Friedreich's ataxia
Molecular Genetics - triplet repeat	Myotonic dystrophy type I
Molecular Genetics - Imprinting	Angelman syndrome
Molecular Genetics - Imprinting	Beckwith-Wiedemann syndrome
Molecular Genetics - Imprinting	Prader-Willi syndrome
Molecular Genetics - Imprinting	Russell-Silver syndrome
Molecular Genetics - Methylation	Rett syndrome
Neurogenetics	Charcot-Marie-Tooth disease type IA
Neurogenetics	Hereditary neuropathy with pressure palsies
Neurogenetics	Spinal muscular atrophy

Effective date; September 2017