Autosomal recessive human diseases:

-Phenylketonuria (phenylalanine hydroxylase ch 12) missense mutation (transversion from a G to a C at codon 413)
-Albinism (tyrosinase)
-Tay-sachs disease (hexosaminidase(HEX)A ch 15)
Sandhoff disease(hexosaminidase B)
-Cystic fibrosis (chloride transporter ch7) pleiotropy
Deletion of three base pairs
-Lesch-Nyah's syndrome (hypoxanthine guanine phosphoribosal transferase)
-Thalassemia (ch 16&11)
-Sickle-cell anemia (ch 11)

Overdominace :
-Sickle cell anemia prevents malaria
-Cystic fibrosis prevents cholera
-Ray-Sachs prevents TB

Incomplete penetrance &expressivity : polydactyly,neurofibromatosis , retinoblastoma

Reduced penetrance : ectrodactyly (SHFM)

Autosomal dominant:
-Familial hypercholesterolemia
-Myotonic dystrophy
-Huntington disease
-Neurofibromatosis, Variable expressivity
-Polycystic kidney disease, dominant or recessive
-Achondroplasia, dominant (Aa)

X-linked recessive inheritance:
-Hemophilia A
-Duchenne muscular dystrophy
-Color blindness

Dominant-Negative Variant: Marfan syndrome due to mutations in the FBN1(fibrillin-1 gene)

Haplo-insufficiency:polydactyly , GLI3 gene

hypermorphic(gain of function): achondroplasia due to mutation in FGFR3 gene.

Myeloproliferative neoplasms(MPNs) due to mutation in JAK2 gene
Neomorphic (BCR-ABL In chronic myeloid leukemia,IDH1,2 in gliomas)

Inter locus heterogeneity(one disease-several genes): deafness, hypertrophic cardiomyopathy,retinitis pigmentosa

-Allelic heterogeneity: cystic fibrosis
Class 1: defect in the synthesis of CFTR (nosense,framshift mutations)
Class2:defect in CFTR protein processing and trafficking (missence,amino acid deletion)
Class3: defect in CFTR channel gating and regulation
(missence,amino acid deletion)
Class4: defect in CFTR channel conductance
(missence,amino acid deletion)
Class5: reduced synthesis of CFTR proteins
(Splicing defect,missence)
Class6: reduction in the stability of CFTR proteins
(missence,amino acid deletion)

Mitochondrial diseases:
-Type II Diabetes Mellitus,
-Parkinson Disease,
-Atherosclerotic Heart Disease,
Stroke,
-Alzheimer Dementia, and
-Cancer.

Abbreviation	MIM No.	Designation
LHON	535000	Leber's hereditary optical neuropathy (Missence M)
MELAS	540000	Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes
	540050	Lactic acidosis with stroke-like signs (Single base M)
MERRF	545030	Myoclonic epilepsy and ragged red fibers (Single base M)
MMC	590050	Maternally inherited myopathy and cardiomyopathy
NARP	551500	Neurogenic muscular weakness with ataxia and retinitis pigmentosa
CEOP	258470	Progressive external ophthalmoplegia
KSS	530000	Kearns-Sayre syndrome (ophthalmoplegia, pigmental degeneration of the retina, and cardiomyopathy)
PEAR	557000	Pearson syndrome (bone marrow and pancreatic failure)
ADMIMY	157640	Autosomal dominant inherited mitochondrial myopathy with mitochondrial deletion in the D loop (type Zeviani)

-Diseases associated with trinucleotide repeat expansions:
FragileXMentalRetardationsyndrome.(CGG,5'UTR,X-linked(Xq27.3),female)
Huntington disease.(CAG, coding sequence,autosomal dominant, male)
myotonic dystrophy.(CTG, 3'UTR, autosomal dominant,female)

spinocerebellar ataxia.
Kennedy disease.
Joseph-disease.
Friedreich ataxia.

Diseases associated with genomic imprinting:

Paternally-expressed genes (IGF2) enhance growth and proliferation. Hypomethylation of these genes can cause Beckwith-Wiedemann Syndrome (BWS) and cancer.

Maternally-expressed genes (CDKN1C) act as tumor suppressors, Hypermethylation can silence these genes leading to BWS
Prader-Willi Syndrome (PWS): Caused by the absence of paternal alleles on chromosome 15(15q11-13), leading to symptoms like obesity and mental retardation.
Angelman Syndrome (AS): Caused by the absence of maternal alleles on 15q11-13 , leading to symptoms like uncontrollable laughter and motor issues.
Rett syndrome.