IMPORTANT GENETIC DISEASES:

Disorder	Chromosome	Genes/Protein
Brain disorders		
CADASIL	19	NOTCH3
Rett Syndrome		MECP2
Lissencephaly		LIS1 – LISX1 (doublecortin)
Epilepsy		
Febrile, GEF+, Dravet, Doose		SCN1a
ADNFLE		Nicotinic receptor
JME		FHC1 (Myoclonin)
Movement disorders/Ataxia		
DOPA responsive dystonia (Segawa syndrome)	14	DYT5: GTP cyclohydroxelase
PKAN		PANK2
Fridreich's ataxia		Frataxin
Ataxia telangiectasia	11	ATM
Episodic ataxia 1		KCNA: K channel
Episodic ataxia 2		CACNA: Ca channel
AHC		
SMA 1 (werding Hoffman)	5	SMN (survival motor neurone)
SMA 2 (Kugelberg Welander)	5	SMN
Spinobulbar muscular atrophy (Kennedy's)	X	CAG repeat (androgen receptor protein)
Neuromuscular		
CMT 1A	17	PMP22 duplication (peripheral myelin protein)
CMT 1B		MPZ (myelin protein 0)
CMT1E		PMP22 point mutation
CMT2		MFN (mitofusin, mitochondrial protein)
HNPP		PMP22 deletion or point mutation
Duchenne/Becker	X	Dystrophin
Myotonic dystrophy	19	Myotonin
Familial hyperthermia		Ryanodine receptor
Hyperkalemic periodic paralysis		Na channel
Hypokalemic periodic paralysis		Ca channel
Thomsen's myotonia congenita		Cl channel
Hyperkeplexia		Glycine receptor
Tumors		
Retinoblastoma		RB1
Pituitary adenoma		MEN1
Familal meningioma		Merlin
NF1	17	Neurofibromin : shwannoma, astrocytoma, optic nerve glioma, neurofibroma, meningioma
NF2	22	Merlin: bilateral shwannomas, multiple meningiomas, astrocytomas, ependymomas

VHL	3	VHL: hemangioblastomas, renal angioma, RCC, pheochromocytoma

TRINUCLEOTIDE REPEATS EXPANSION:

Disorder	Trineucleotide	Gene/Protein
Huntington's	CAG	Huntigtin
SCA	CAG	Ataxin
Spinobulbar muscle atrophy (Kennedy's)	CAG	Androgen receptor
DRPLA	CAG	Atrophin
Friedreich's ataxia	GAA	Frataxin
Myotonic dystrophy	CTG	Myotonin
Fragile X syndrome (FRAXA)	CGG	FMR1
Fragile X associated tremors/ataxia	CGG	FMR1
Myotonic dystrophy type II	CCTG (tetranucleotide)	
SCA type 10	ATTCT (pentanucleotide)	

ATPASE mutation	Disorder
ATP1	Alternating hemiplegia of childhood
ATP7A	Menkes disease
ATP7B	Wilson disease

CHANNELOPATHIES:

Disorder	Gene	Ion channel
Hyperkalemic periodic paralysis	SCN4A	Sodium
Paramyotonia congenital	SCN4A	Sodium
Potassium aggravated myotonia	SCN4A	Sodium
Myotnonia congnita	CLCN1	Chloride
Hypokalemic periodic paralysis	CACNLA3	Calcium
Familial hemiplegic migraine	CACNA1A	Calcium
Episodic atacia 2	CACNA1A	Calcium
Episodic ataxia 1	KCNA1	Potassium
Anderson Tawil syndrome	KCNJ2	Potassium
Hereditary hyperkeplexia	GLRA1	Glycine

LEUKODYSTROPHIES

Disorder	Enzyme/protein deficiency
Canavan	Aspartocyclase
Metachromatic	Arylsulfatase
Adrenoleukodystrophy	Long chain fatty acids
Krabbe's	Galactocerebrosidase
Pelizeus merzbacher	Proteplipid protein