The disease		The reason	Symptoms
Griscelli syndrome (GS)		Mutations in MYO5A, RAB27A and MLPH genes	Pigmentary dilution of the skin, silver-grey hair, melanin clumps within hair shafts
Lysosomal storage diseases	Glycolipidoses (sphingolipidoses)	defective lysosomal enzymes	lead to severe psychomotor retardation and premature death. 
	Mucopolysaccharidoses	defective lysosomal enzymes For GAGs	
	Oligosaccharidoses	defective lysosomal enzymes	
	Possible diseases	Deficiency of enzyme glucocerebrosidase	
	I-cell disease also called mucolipidosis	A deficiency in tagging enzyme that phosphorylates mannose	
Peroxisomal diseases	Zellweger syndrome	Mutations of PEX genes	Lethal (fatal)
	X-linked adrenoleuko-dystrophy (XALD)	Defective transport of very long-chain fatty acid (VLCFA) across the peroxisomal membrane.	Accumulation of fatty acids in the cell.
	Single peroxisomal enzyme deficiencies	Defective specific peroxisomal enzymes	

	The disease	The reason	Symptoms
	Emery-Dreifuss muscular dystrophy	The emerin gene (X-linked disease) The lamin A gene (autosomal dominant disease).	muscular dystrophy
Defective dystrophin	**Duchenne muscular dystrophy (X-linked)**	Absent protein dystrophin	degenerative muscle diseases (muscular dystrophy)
	Becker muscular dystrophy (X-linked)	Defective protein dystrophin	
Kinesins defective	Alzheimer	**tau protein** aggregates inside the nerve cell (maybe because kinesins defective)leading to the cell's death	Dementia
	amyotrophic lateral sclerosis (ALS) Lou Gehrig's disease	*Mutants in certain kinesin proteins *Lamin A mutations as well *defective neurofilaments	loss of muscle control
	Charcot-Marie-Tooth disease	*Mutants in certain kinesin proteins *Lamin A mutations as well *intermediate filaments are mutated *defective gap junctions and mutated connexins	peripheral neuropathies

	The disease	The reason	Symptoms
	epidermolysis bullosa simplex	keratin gene mutations	defective skin that is susceptible or vulnerable
Collagen-related diseases	Crosslinking Defects	Deficiency in lysyl oxidase, which impairs collagen fiber crosslinking.	Weak tissues, fragile tendons (e.g., Achilles tendon)
	Chondrodysplasias	Mutations in type II collagen affecting cartilage.	bone and joint deformities
	Ehlers-Danlos Syndrome (EDS)	Mutations in type I, III, V collagens or collagen- processing enzymes.	disorders that affect the skin, bones, blood vessels, and other organs skin fragility and hyperextensibility and joint hypermobility.
	Type III EDS	mutations found in the gene of type III collagen.	fragile blood vessels +stretchy and hypermobile joints.
	Osteogenesis Imperfecta (OI)	Mutations in COL1A1 and COL1A2 genes affecting the assembly of type I collagen.	fragile, soft, brittle, and easily broken bones.
	Marfan's syndrome	mutated fibrillin	Rupture of aorta Others: A tall, thin build; Long arms,legs, fingers, and toes and flexible joints; Scoliosis, or curvature of the spine; A chest that sinks in or sticks out; Crowded teeth; Flat feet.
defective gap junctions	Marie-Charcot-Tooth disease Deafness Skin disorders Cataracts	Diseases caused by defective gap junctions and mutated connexins	—

The disease	The reason	Symptoms
Retinoblastoma	Defective in Rb protein	Eye tumor in childrer
Ataxia-Telangiectasia	Mutation in the ATM gene	Ataxia: Uncoordinated movements, difficulty walking /Telangiectasias: Enlarged blood vessels under the skin /Immune Weakness/Cancer Risk.

Mitochondrial diseases

Mitochondrial inheritance	Luft's Disease	Defective oxidative phosphorylation coupling	Hypermetabolism Hyperthermia
	MERRF	Mutation in mitochondrial tRNA genes	
	MELAS	Mutation in mitochondrial DNA	
	LHON	Mutation in mitochondrial DNA, impairing oxidative phosphorylation,	Vision loss/ Degeneration of the optic nerve
	NARP	Mutation in mitochondrial DNA	
Autosomal recessive	Fumarase Deficiency	Defective fumarase enzyme	Excretion of fumarate and succinate in urine
X-linked	Pyruvate Dehydrogenase Deficiency	Defective pyruvate dehydrogenase complex	Metabolic acidosis- Elevated lactate in blood and CSF- Increased pyruvate and alanine levels