Disease	Type	Cause	Function (normally)	Genetic
Acid Maltase Deficiency	autosomal recessive metabolic disorder	acid alpha-glucosidase (acid maltase) deficiency	necessary for the break down of glycogen	long arm of chromosome **17q25.2-q25.3**
Carnitine deficiency inborn error of fatty acid transport		Defect in the transporter responsible for moving carnitine across the plasma membrane	Carnitine is involved in the oxidation of fatty acids	gene responsible for the OCTN2 carnitine transporter is **SLC22A5** at **5q31.1-32.**
Carnitine palmitoyl transferase deficiency		enzymatic defect that prevents long-chain fatty acids from being transported into the mitochondria	CPT1 + CPT2 → oxidizes long chain fatty acids in the mitochondria	---
Myoadenylate deaminase deficiency	---	genetic defect in the myoadenylate deaminase enzyme	processing adenosine triphosphate (ATP) in muscle cells	---
Debrancher enzyme deficiency	interferes with the processing of food (carbohydrates) for energy production.	defect in the debrancher enzyme gene	breakdown of glycogen in the muscles and liver	---
Lactate dehydrogenase deficiency		genetic defect in the lactate dehydrogenase enzyme	recycles byproducts of carbohydrate metabolism	---
Phosphofructokinase deficiency (Tarui disease)		genetic defect in the phosphofructokinase enzyme	breakdown glucose	---
Phosphoglycerate kinase deficiency		genetic defect in the phosphoglycerate kinase enzyme	breakdown glucose	---

Metabolic muscle diseases

Metabolic diseases of bones

Disease	Characteristics	Cause
Osteoporosis	Bone microarchitecture deteriorates // Bone mineral density (BMD) is reduced // Amount and variety of proteins in bone are altered Primary type 1: postmenopausal Primary type 2 (senile): after age 75, ratio of 2:1 (F, M)	-calcium deficiency -Vit D3 deficiency -drop in estrogen / testosterone
Paget's Disease	bones to become thickened and enlarged but also brittle	Viral or genetic → disorder of osteoblasts and osteoclasts
Osteogenesis Imperfecta	brittle bones that break or fracture easily	gene defect in the production of collagen (type 1) substitution of glycine to bulkier amino acids
Rickets	softening of bones in **children**	Deficiency or impaired metabolism of vitamin D phosphorus or calcium
Osteomalacia	softening of bones in **adults** have a normal amount of collagen	-insufficient calcium absorption -resistance to the action of vitamin D -phosphate deficiency caused by increased renal losses -hyperparathyroidism
Acromegaly	Overgrown bones	Benign tumor on the pituitary gland in the brain → excess of growth hormone production by the body
Fibrous Dysplasia	Normal bone is replaced with fibrous bone tissue abnormal growth or swelling of bone	Gene mutation that affects the cells that produce bone
Osteomyelitis	**---**	Bacterial infection of bone fungi or other germs
Hypocalcaemia	Presence of low serum calcium levels in the blood (unbound / ionized)	-Parathyroid hormone [PTH] deficiency / malfunction -Vitamin D deficiency.
Hypophosphatasia	Affect bones and teeth	Mutations in the ALPL (inherited disorder) → defect in alkaline phosphatase

Bone cancer

Cancer	Characteristics	Occurs in	Age group
Osteosarcoma	most common primary malignant bone cancer risk of spread to the lungs.	long bones of the arms & legs knees and shoulders	10 - 25 years
Ewing's sarcoma	most aggressive bone tumor	middle of the long bones of the arms and legs	4 - 15 years of age
Chondrosarcoma	second most common bone tumor 25% of all malignant bone tumors spread to the lungs and lymph nodes.	affects the bones of the pelvis and hips	people over 40 years of age

Collagen linked diseases

Disease	Characteristics	Genetics
Collagenopathy	Defect in cartilage found in joints and the spinal column, the inner ear, and the jelly-like substance that fills the eyeball	Mutations in **COL2A1** gene (codes for type 2) and **COL11A1, COL11A2** (with **COL2A1** codes for type 11)
Alport syndrome	Defect basement membranes in the kidney, and inner ear (most often affects males)	Mutation in **COL4A3, COL4A4, COL4A5**
Ullrich congenital muscular dystrophy	muscle weakness soon after birth not able to walk unassisted	Mutations in the **COL6A1, COL6A2**, and **COL6A3** genes on chromosomes 21 and 2

Ehlers-Danlos syndrome (EDS)

Causes:

-Defect in the synthesis of collagen (Type I or III)
→ increased elasticity within these structures
-Abnormalities in tenascin protein
regulating the normal distribution of collagen in the connective tissues of the body.

-Mutations in:
Fibrous proteins: COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, and TNXB
Enzymes: ADAMTS2, PLOD1, B4GALT7

Type	Genetics	Characteristics
Classical type	autosomal dominant genetic trait	Joint hypermobility // skin hyperextensibility (laxity) // fragility
Hypermobility type		Joint hypermobility // dislocations are frequent
Kyphoscoliosis type	autosomal recessive genetic trait	Fragile globe of the eyes // significant skin and joint laxity // severe curvature of the spine (scoliosis)
Tenascin-X deficient type		Joint hypermobility // hyperelastic skin // fragile tissue
Dermatosparaxis type	---	fragile skin that is soft and doughy with sagging and folding // diagnosed with a skin biopsy
Arthrochalasia type (arthrochalasis multiplex congenita)	autosomal dominant and recessive inheritance	short in height // joint laxity and dislocations // skin involvement is variable
Vascular type (the arterial form)		spontaneous rupture of arteries and bowel can lead to death

Elastin linked diseases

Disease	Type	Characteristics	Genetics
Cutis laxa	autosomal dominant, autosomal recessive (more severe)	loose, sagging skin // increased risk of abnormal bulging (an aneurysm) in aorta // lung disease called emphysema	mutations in **ATP6V0A2, ATP7A (makes protein important for regulating copper levels), EFEMP2, ELN,** or **FBLN5** genes
occipital horn syndrome	X-linked form of cutis laxa	mild type of Menkes syndrome (condition that affects copper levels in the body)	---
Supravalvular aortic stenosis (SVAS)	autosomal dominant	narrowing of aorta	mutations in the **ELN** gene
Williams syndrome		reduces the production of elastin by half // connective tissue abnormalities // cardiovascular disease	CLIP2, ELN, GTF2I, GTF2IRD1, and LIMK1 genes is deleted from chromosome 7
Marfan syndrome		unusually tall // long limbs // long, thin fingers	Mutation in FBN1 gene, encodes fibrillin-1 (essential for extracellular matrix, biogenesis and maintenance of elastic fibers)

Melanin disorders

Disease	Cause	Characteristics	Genetics
Albinism	defect in tyrosine metabolism	white hair, skin & iris color // vision defects and photophobia.	autosomal recessive, autosomal dominant
Oculocutaneous albinism	deficiency of tyrosinase activity	total absence of pigment from the hair, eyes & skin	**---**
hyperpigmentation	caused by sun damage, inflammation or acne vulgaris.	patches of skin become darker	**---**