

**TERM 1 DISEASE CHEAT SHEET**

This is a cheat sheet consisting of the main diseases covered in Term 1 across metabolism, tissues of the body and molecules, genes and diseases. The information on this sheet is not extensive but provides an overall summary. Enjoy!

1. Metabolic syndrome
2. Hashimoto’s thyroiditis
3. Gestational diabetes
4. Hypocalcaemia
5. Lactose intolerance
6. Essential fructose
7. Fructose intolerance
8. Hyperlactaemia
9. Lactic acidosis
10. Glucose-6-phosphate dehydrogenase deficiency
11. Von Geirke
12. McArdle disease
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14. Phenylketonuria
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29. Hyperthyroidism
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31. Anencephaly
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34. Ehler’s danlos
35. Scurvy
36. Osteogenesis imperfecta
37. Marfan’s syndrome
38. William’s disease
39. Actinic keratosis
40. Squamous cell carcinoma
41. Basal cell carcinoma
42. Psoriasis
43. Sun burn
44. Vitiligo
45. Malignant melanoma
46. Alopecia
47. Acne
48. Epidermolysis bullosa
49. Solar elastosis
50. Stretch marks
51. Erythrodermic psoriasis
52. Osteoarthritis
53. Acromegaly
54. Neonatal hypothyroidism
55. Osteoporosis
56. Achondroplasia
57. Rickets
58. Osteomalacia
59. Disuse atrophy
60. Denervation atrophy
61. Myasthenia gravis
62. Botulism
63. Organophosphate poisoning
64. Duchene muscular dystrophy
65. Multiple sclerosis
66. Gullian Barre
67. Parkinson’s
68. Vasovagal syncope
69. Horner’s syndrome
70. Hirschprung’s
71. Down’s syndrome
72. Edward’s syndrome
73. Patau’s syndrome
74. Klinefelters
75. Turners
76. Double Y
77. Triple X
78. Philadelphia chromosomes
79. Malignant hyperthermia
80. Thyrotoxidosis
81. Rhabdomyolitis
82. Polymyolitis
83. Alzheimer’s
84. Pyruvate dehydrogenase deficiency
85. Swyer’s syndrome
86. Leri-Weill dyschondroplasia
87. Rhizomelic chondroplasia punctate
88. Emphysema
89. Classic haemophilia
90. Sickle cell disease
91. Thalassemia
* **Metabolic syndrome**
	+ Insulin resistance
	+ Glucose intolerance
	+ Hypertension
	+ Abdominal obesity
* **Hashimoto’s thyroiditis**
	+ Autoantibodies to the TSH receptor and to thyroid peroxidases
	+ Extensive infiltration of the thyroid parenchyma by lymphocytes and plasma cells
	+ Most common cause of hypothyroidism and goitre in non-iodine deficient areas
* **Gestational diabetes**
	+ Normally during pregnancy, oestrogen and progesterone hormones increase the sensitivity of beta cells in the pancreas to glucose
	+ Increase in glucose intake during pregnancy
	+ Increases sensitivity = increased insulin secretion and production
	+ Beta cell dysfunction = gestational diabetes
	+ Complications
		- Miscarriage
		- Macrosomia

* **Hypocalcaemia**
	+ Causes
		- Hypoparathyroidism
		- Hypovitaminosis D
			* Osteomalacia
			* Rickets
		- GI malabsorption
* **Lactose intolerance**
	+ Reduced/inability of lactase to break down lactose
	+ Symptoms
		- Abdominal pain
		- Bloating
		- Diarrhoea
			* Increases osmotic pressure and draws in water
		- Gas
	+ Fermented by colonic bacteria to form hydrogen gas, organic acids, carbon dioxide and methane
* **Essential fructose**
	+ Caused by lack of fructokinase
	+ Accumulation of fructose in blood but just passed out in urine
	+ No clinical signs or symptoms
* **Fructose intolerance**
	+ Lack of aldolase
	+ Accumulation of fructose-1-phosphate – toxic to liver – liver damage
	+ Treatment
		- Remove fructose from diet
* **Hyperlactaemia**
	+ Elevated plasma lactate levels
	+ But not high enough to cause change in pH due to blood’s buffering capacity
	+ Below renal threshold
* **Lactic acidosis**
	+ Elevated plasma lactate levels
	+ Causes decrease in blood pH
	+ Above renal threshold
* **Glucose-6-phosphate dehydrogenase deficiency**
	+ Lack of glucose-6-phosphate dehydrogenase
	+ Decreased NADPH synthesis and nucleic acid synthesis
	+ Decrease in NADPH causes decrease in glutathione which makes cells more susceptible to oxidative stress
	+ RBC only get NADPH through pentose phosphate pathway
	+ RBC do not have NADPH, so haemoglobin aggregates forming Heinz bodies
	+ Heinz bodies rupture leading to haemolytic anaemia
* **Von Geirke**
	+ Excess glycogen stores
	+ Liver glucose-6-phosphotase deficiency
	+ Enlarged liver
	+ Hypoglycaemia
* **McArdle disease**
	+ Insufficient glycogen stores
	+ Glycogen phosphorylase deficiency in muscle
	+ Exercise induced muscle pain and cramps
* **Hypothyroidism**
	+ Symptoms
	+ Positive nitrogen balance
	+ Weight gain
	+ Reduced amino acid catabolism
	+ Under-stimulation of metabolism
	+ Depression
* **Phenylketonuria**
	+ Autosomal recessive on chromosome 12
	+ Accumulation of phenylalanine in tissues and plasma due to lack of phenylalanine hydroxylase
	+ Phenyl ketones in urine
	+ Treatment
		- Low phenylalanine diet
		- Tyrosine supplements
	+ Symptoms
		- Severe learning difficulties
		- Seizures
		- Microcephaly
* **Homocystinurias**
	+ Problem breaking down methionine
	+ Excess homocysteine in urine
	+ Autosomal recessive
	+ Defect in cystathionine beta synthase
	+ Affects CNS, connective tissue
	+ Treatment
		- Low methionine diet
		- Folate and vitamin B supplements
* **Defects in urea cycle**
	+ Increase of ammonia
	+ Symptoms
		- Vomiting
		- Seizures
		- Coma
		- Mental retardation
* **Hyperlipidaemia**
	+ Hypercholesterolaemia
		- Symptoms
			* Corneal arcus
			* Xanthalesma
			* Tendon Xanthoma
	+ Treatments of hyperlipidaemias
		- Diet
			* Reduced cholesterol and increased fibre
		- Lifestyle
			* Reduce smoking
		- Statins
			* Reduces cholesterol formation
		- Bile salt sequestrants
			* Forces liver to produce more bile lipids using up more cholesterol
* **Diabetes mellitus**
	+ Chronic hyperglycaemia
		- Decreases insulin secretion
		- Increased lipolysis
		- Increase glycogenolysis
		- Decreased incretins
	+ Presentations
		- Glucouria
			* Opportunistic infections such as thrush or UTIs
		- Polyuria
		- Polydipsia
	+ Type 1
		- Insulin dependent
		- Autoimmune destruction of beta cells
	+ Type 2
		- Not insulin dependent
		- Associated with insulin resistance
		- Insulin secretion increased
		- Overworking of beta cells
		- Insulin resistance
			* Causes hyperinsulinaemia
			* Beta cell failure
			* Impaired glucose tolerance
			* Causing type 2 diabetes
		- Aims of diabetes treatment
			* Alleviate symptoms
			* Normalise long term glucose levels
			* Reduce risk of long term cardiovascular problems
	+ Type 3
		- Type 2 diabetes in type 1 diabetics
	+ Gestational diabetes
		- Beta cell dysfunction
		- Third trimester of pregnancy
	+ Insulin resistance occurs from
		- Genetic factors
		- Environmental factors
			* Diet
			* Lifestyle
* **Long term effects of hyperglycaemia**
	+ Glycation of proteins
		- Microvascular
			* Sticky blood
			* Retinopathy
			* Peripheral neuropathy
			* Nephropathy
		- Formation of ROS
* **Hypercalcaemia**
	+ Forms stones, depression and abdominal pain
	+ Treatment
		- Increased fluids
	+ Signs and symptoms
		- Altered behaviour, Depression
		- Anorexia
		- Increased bone resorption
		- Increased risk of fractures
		- Risk for kidney stones
	+ Causes
		- Primary hyperparathyroidism
		- Hypervitaminosis D
* **Hypocalcaemia**
	+ Signs and symptoms
		- Involuntary muscle contractions
		- Seizures
		- Personality changes
		- Respiratory arrest
		- Death
	+ Causes
		- Hypoparathyroidism
		- Hypovitaminosis D
		- GI malabsorption
* **Primary hyperparathyroidism**
	+ PTH above normal range
	+ Increased risk of fractures
	+ Increased risk of kidney stones
* **Addison’s disease**
	+ Autoimmune destruction of the adrenal cortex
	+ Female predominance
	+ Deficiency of all hormones
	+ Clinical features
		- Weight loss
		- Anorexia
		- Depression
		- Abdominal pain
	+ Pigmentation – ACTH
	+ Biomedical features
		- Low sodium
		- High potassium
		- High urea
		- Low glucose
* **Hypoadrenal crisis**
	+ Hyponatremia
	+ Hypoglycaemia
	+ Hyperkalaemia
	+ Hypotension
	+ Severe dehydration
* Treatment
	+ Hydrocortisone
	+ Fludrocortisone
* Confirmation tests
	+ 0900 cortisol test
	+ ACTH
	+ Short synacthen test
	+ Plasma renin
	+ Adrenal antibodies
* **Secondary adrenal failure**
	+ Inadequate ACTH secretion – pituitary tumour
* **Exogenous steroids**
	+ Lower ACTH and CRH
* **Additional symptoms**
	+ Visual field problems
* **Hypoadrenalism**
	+ Primary
		- High ATCH
		- Pigmented
		- GC and MC
	+ Secondary
		- ACTH low
		- Pale
		- GM
* **Hypothyroidism**
	+ Symptoms and signs
		- Bradycardia
		- Weight gain
		- Anorexia
		- Goitre
		- Cold intolerance
		- Cretenism in infants
	+ Causes
		- Autoimmune disease
		- Pituitary tumour – secondary
		- Iodine deficiency
* **Hyperthyroidism**
	+ Signs and symptoms
		- Tachycardia
		- Heat intolerance
		- Goitre
		- Weight loss
	+ Causes
		- Autoimmune
		- Toxic multinodular goitre
		- Solitary toxic adenoma
* **Tay Sach’s disease**
	+ HEX8 gene affected
	+ Autosomal recessive
	+ Defect of GM2-gangliosidase causes accumulation of gangliosides
	+ Leads to nerve cell impairment
	+ Deterioration of mental and physical abilities leading to early death
* **Anencephaly**
	+ Failure of the neural tube forming at the cranial region thus no brain is formed
* **Spina bifida**
	+ Failure of the neural tube forming at the caudal region
	+ Folic acid administration reduces neural tube defects up to 70%
* **Cystic fibrosis**
	+ 3 base pair deletion of CFTR-1 gene
	+ Defect in chloride channel
	+ Chlorine unable to leave the cell thus sodium and water reabsorbed back into the lumen
	+ Causes sticky mucus = recurrent chest infections as cilia unable to move thick mucus into the oropharynx
	+ Infertility issues in men = missing vas deferens
	+ Salty sweat
	+ Blockage of pancreatic ducts = sterrohoerea = fatty stools
	+ Can also result in pancreatisis = digestion of pancreas by pancreatic enzymes
* **Ehler’s Danlos**
	+ Various failures of collagen biosynthesis
	+ In type IV – failure to produce type III collagen leads to aortic rupture
* **Scurvy**
	+ Lack of vitamin C reduces prolyl hydroxylase activity leads to gum ulceration and haemorrhage
* **Osteogenesis imperfecta**
	+ Type 1 – mutation in COL1A1 gene coding for collagen
		- Spontaneous bone fractures
		- Bones are thin and curved
		- Sclera appear blue
		- Decreases in severity with growth
		- Slightly shorter in stature compared to siblings
	+ Type 2 – mutation in COL1A1 gene or genes coding for collagen
		- Incompatible with life
		- Infant dies due to contractions of the uterine wall or breaks all bones in body during child birth
* **Marfan’s syndrome**
	+ Autosomal dominant disorder
	+ Fibrillin-1 gene
	+ Abnormally tall
	+ Arachnodactyly
	+ Frequent joint dislocation
* **William’s disease**
	+ Spontaneous deletion of chromosome 7
	+ Associated with learning difficulties and cardiovascular problems
* **Actinic keratosis**
	+ Chronic sun exposure = epidermal dysplasia
* **Squamous cell carcinoma**
	+ Basal keratinocytes carcinoma
	+ Higher chance of metastasis
* **Basal cell carcinoma**
	+ Follicular keratinocytes
	+ Lower chance of metastasis
* **Psoriasis**
	+ Extreme proliferation of basal epidermal layer in response to inflammatory cytokines
	+ Epidermal turnover reduced from 28 to 3-7 days
	+ Treatment – vitamin D analogues and topical steroids
	+ Can compromise thermoregulation
* **Sun burn**
	+ Keratinocyte apoptosis
	+ Redding
	+ Oedema
* **Vitiligo**
	+ Autoimmune destruction of melanocytes
	+ Distinct depigmentation
* **Malignant melanoma**
	+ Retention above basement membrane has good prognosis, if penetrated not good
* **Alopecia**
	+ Miniaturisation of hair follicle due to exposure of dihydrotestosterone
* **Acne**
	+ Increased sebum production
	+ Infection of normally harmless bacteria
* **Epidermolysis bullosa**
	+ Mechanically fragile skin
	+ Mutation in range of structural proteins leading to blistering
* **Solar elastosis**
	+ Excessive exposure to UV
	+ Overproduction of elastic fibres
* **Stretch marks**
	+ Collagen fibre deposition along the areas of most stress caused by scar tissue
* **Erythrodermic psoriasis**
	+ Inability to vasoconstrict vessels during cold temperatures = heat loss
* **Osteoarthritis**
	+ Focal and progressive loss of hyaline cartilage with changes to underlying bone
	+ Formation of osteophytes
	+ Causes
		- Severe bone injury
		- Fractures
		- Tears in menisci
* **Acromegaly**
	+ Broadened hands and feet
	+ Increased appositional growth as epiphyseal growth plates have fused
* **Neonatal hypothyroidism**
	+ Deficiency of thyroid hormones in neonates results in permanent neurological damage in infants and cretenism and decreased bone growth
* **Osteoporosis**
	+ Primary
		- Type 1
			* Only occurs in most menopausal women
				+ Due to decrease in oestrogen as oestrogen acts to decrease osteoclast activity
		- Type 2
			* Occurs in both men and women due to decrease in osteoblast activity
	+ Non-modifiable risks
		- Age
		- Gender
		- Previous fractures
	+ Modifiable factors
		- Vitamin D intake
		- Calcium intake
		- Exercise
		- Smoking
* **Achondroplasia**
	+ Mutation in FGFR-3 gene causes epiphyseal plates to close early
	+ No effect on intellect
	+ Autosomal dominant
* **Rickets**
	+ Lack of calcium in bone in children
	+ Insufficient calcium deposition
	+ Bones become soft or malformed
	+ Bowed legs
* **Osteomalacia**
	+ Lack of vitamin D and calcium
	+ Insufficient calcium deposition
	+ Surfaces of trabeculae become thicker due to deposition of non-mineralised osteoid leading to weakening
* **Disuse atrophy**
	+ Seen in bed rest, limb immobilisation and sedentary lifestyle
	+ Loss of contractile proteins leads to reduced fibre diameter and loss of power
* **Denervation atrophy**
	+ Lower motor neuron lesions between spinal cord and muscle – associated with loss of muscle and tone and muscle atrophy
* **Myasthenia gravis**
	+ Autoimmune disease
	+ End plate destruction of ACh receptors
	+ Loss of junctional folds at the end of the plate and widening of the synaptic cleft
	+ Symptoms
		- Drooping eyelids
		- Fatigability
	+ Treatment
		- Acetylcholinesterase inhibitors
* **Botulism**
	+ Clostridium botulinum released botulism toxin which blocks ACh release leading to paralysis
* **Organophosphate poisoning**
	+ Organophosphates inhibit acetylcholinesterase irreversibly
	+ Death from respiratory failure or CVS problems
* **Duchene muscular dystrophy**
	+ X-linked recessive
	+ Completely loss of dystrophin
	+ Dystrophin links the ECM to the cytoskeleton and stabilises the sarcolemma
	+ Respiratory failure as disease progresses to the head, chest and cardiac muscles
	+ Fragile sarcolemma tears during contraction causing inflammation and necrosis
	+ Treatment
		- Steroid therapy
		- Gene therapy
* **Duchene muscular dystrophy**
	+ X linked recessive
	+ Deficiency of dystrophin
	+ Which links the ECM and cytoskeleton and stabilises the sarcolemma
	+ Tears in the sarcolemma causes inflammation and necrosis
	+ Treatment – steroid therapy and gene therapy
* **Multiple sclerosis**
	+ Autoimmune destruction of myelin in CNS
	+ Beta-interferons
* **Guillian Barre**
	+ Autoimmune destruction of myelin in PNS
* **Parkinson’s**
	+ Loss of dopamine-secreting neurons in substantia nigra
* **Vasovagal syncope**
	+ Increased sympathetic followed by decreased sympathetic
	+ Parasympathetic unopposed – causing vasodilation, bradycardia, and syncope
	+ Causes
		- Severe pain
		- Blood/needle phobia
* **Horner’s syndrome**
	+ Disruption of sympathetic nerves supplying the eye
	+ Symptoms
		- Upper eyelid drooping
		- Pupillary constriction
		- Difference in pupil sizes
* **Hirschprung’s**
	+ Congenital disease
	+ Parasympathetic innervation does not develop properly
	+ Ineffective peristalsis
	+ Faeces accumulates in colon
	+ Causes serious infections = fatal
* **Down’s syndrome +21**
	+ Risk increases with maternal age above 35
	+ Clinical features
		- Mild to moderate intellectual disability
		- Hypothyroidism
		- Infertility
		- Eye disorders
* **Edward’s syndrome +18**
	+ Lifespan 1-5 days
	+ Severe learning difficulties
	+ Heart and kidney problems
	+ Growth retardation
* **Patau’s syndrome – +13**
	+ Severe learning disabilities
	+ Polydactyly
	+ Multiple heart and brain abnormalities
* **Turner’s syndrome, X**
	+ Heart defects
	+ Infertility
	+ No mental retardation
* **Triple X syndrome**
	+ Tall stature
	+ Microcephaly
	+ Auditory processing defects
* **Klinefelter syndrome - XXY**
	+ Reduced portion of testosterone
	+ Gynecomastia
	+ Infertility
	+ Language learning and reading impairment
* **Double Y syndrome**
	+ Normal testosterone and fertility
	+ Slightly lower IQ levels
* **Philadelphia chromosome**
	+ A reciprocal translocation between chromosome 9 and 22
	+ Translocation brings ABL1 gene on chromosome 9 to BCR gene on chromosome 22
	+ This results in fusion proteins which results in fusion protein which is oncogenic
* **Malignant hyperthermia**
	+ Exposure to anaesthetics stimulates the release of stored Ca2+, leading to muscle contraction and generation of excessive heat
	+ Treated with muscle relaxant dantrolene and cooling
* **Thyrotoxicosis**
	+ Excess thyroid hormones cause protein catabolism and loss of muscle mass
* **Rhabdomyolysis**
	+ Factors released by damaged muscle causes kidney damage
* **Polymyositis**
	+ Myalgia from over use or from influenza infection
* **Alzheimer’s**
	+ Associated with accumulation of extracellular plaques and neurofibrillary tangles in neurons
* **Pyruvate dehydrogenase deficiency**
	+ Mutation in codon 10 of the N-MTS of PDH E1a subunit resulting in arginine to proline substitution
	+ Reduced reuptake in the mitochondria
	+ X-linked dominant disorder
		- Results in build-up of lactic acid and neurological problems
* **Swyers syndrome**
	+ Loss or mutation in the NLS in sex determining region
	+ XY genotype but outwardly female
	+ Mutation of nuclear localisation signals
* **Leri-Weill dyschondroplasia**
	+ Mutation in the SHOX gene
	+ Short stature
* **Rhizomelic Chondroplasia puntata**
	+ Mutation in PEX7
* **Emphysema**
	+ Deficiency of alpha-1-antitrypsin
	+ Destruction of alveolar wall by elastase
* **Classic haemophilia**
	+ Loss of factor VIII causes prolonged bleeding as unable to form clot
	+ Treated with recombinant factor VIII
* **Sickle cell disease**
	+ Single base substitution causing replacement of glutamic acid to valine
	+ Causes sticky pockets due to polymerization of haemoglobin
	+ Sickle cell more prone to lyse = anaemia
	+ More rigid = blocked vasculature
	+ Treatment
		- Folic acid
		- Stem cell transplant
* **Thalassaemia**
	+ Genetic blood disorder
		- Inadequate form and amount of haemoglobin
		- Increased destruction of red blood cells = anaemia