Heat-Related Disorders

1. **Heat Cramps** → Painful muscle cramps due to **electrolyte loss** (mainly sodium) from sweating. Treated with **oral rehydration and electrolyte replacement**.
2. **Heat Exhaustion** → **Core temperature <40°C (104°F)** with **profuse sweating, weakness, nausea, and dizziness** due to dehydration. **Mental status remains intact**.
3. **Heat Stroke** → **Core temperature >40°C (104°F)** with **CNS dysfunction** (confusion, seizures, coma) and **anhidrosis (lack of sweating in severe cases)**. **Medical emergency!**
4. **Exertional Heat Stroke** → Occurs in **athletes, military personnel**, and laborers due to excessive exertion. Patients may **still sweat** initially.
5. **Classic (Non-Exertional) Heat Stroke** → Occurs in **elderly, chronically ill, or those with poor heat dissipation** (e.g., no air conditioning). Patients **typically do not sweat**.
6. **Treatment of Heat Stroke** → **Rapid cooling (ice-water immersion is most effective)**, IV fluids, airway protection, and benzodiazepines for shivering. **Avoid antipyretics (e.g., acetaminophen, NSAIDs)**.
7. **Complications of Heat Stroke** → **Rhabdomyolysis, DIC, renal failure, and ARDS** due to excessive heat stress.
8. **Risk Factors for Heat-Related Illness** → **Obesity, dehydration, cardiovascular disease, diuretics, anticholinergics, and beta-blockers**.
9. **Prevention of Heat Illness** → **Adequate hydration, acclimatization, light clothing, and avoiding exertion in high heat**.
10. **Difference Between Heat Exhaustion & Heat Stroke** → **Mental status changes (CNS dysfunction) occur only in heat stroke**.

Hyperthermia & Hypothermia

Hyperthermia (Elevated Body Temperature >38.3°C / 101°F)

1. **Heat Exhaustion** → **Profuse sweating, weakness, tachycardia, and dehydration** but **no CNS dysfunction**. Core temp **<40°C (104°F)**.
2. **Heat Stroke** → **Core temp >40°C (104°F) + CNS dysfunction (confusion, seizures, coma)**. Immediate **cold-water immersion** is the treatment of choice.
3. **Malignant Hyperthermia** → Triggered by **volatile anesthetics (halothane, sevoflurane) or succinylcholine**, due to **defective ryanodine receptors → excessive calcium release**. Treated with **dantrolene**.
4. **Neuroleptic Malignant Syndrome (NMS)** → **Fever, lead-pipe rigidity, altered mental status, autonomic instability** due to **dopamine blockade (antipsychotics)**. Treat with **dantrolene + bromocriptine**.

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5. **Serotonin Syndrome** → **Fever, hyperreflexia, clonus, mydriasis, diarrhea** due to **excess serotonin (SSRIs, MAOIs, MDMA)**. Treat with **cyproheptadine**.
6. **Thyroid Storm** → **Fever, tachycardia, agitation, delirium, hypertension** in a patient with **hyperthyroidism**. Treat with **beta-blockers, PTU, steroids, and iodine**.
7. **Anticholinergic Toxicity** → **Hyperthermia, dry skin, mydriasis, urinary retention, delirium** due to **atropine, antihistamines, TCAs**. Treat with **physostigmine**.
8. **Sympathomimetic Overdose (Cocaine, Amphetamines)** → **Hyperthermia, hypertension, mydriasis, agitation**. Treat with **benzodiazepines and cooling**.
9. **Treatment of Hyperthermia** → Remove from heat, IV fluids, active cooling (**ice packs, cold water immersion**), and treat underlying cause.

Hypothermia (Core Temperature <35°C / 95°F)

1. **Mild Hypothermia (32-35°C / 90-95°F)** → **Shivering, tachycardia, confusion, ataxia**. Passive rewarming (blankets, warm room).
2. **Moderate Hypothermia (28-32°C / 82-90°F)** → **No shivering, bradycardia, slowed reflexes, AMS**. Active **external** warming (warm blankets, heating pads, warm IV fluids).
3. **Severe Hypothermia (<28°C / 82°F)** → **Coma, ventricular arrhythmias (J waves on ECG), asystole**. Requires **active internal rewarming (warm fluids, ECMO if needed).**
4. **ECG Finding in Hypothermia** → **Osborn (J) waves** seen at **<32°C (90°F)**.
5. **Cold-Induced Arrhythmias** → **Bradycardia, atrial fibrillation, and ventricular fibrillation**. Rewarm first before giving antiarrhythmics.
6. **Frostbite** → **Cold-induced tissue necrosis, initially numb and pale, then turns black**. Treat with **rapid rewarming in warm water (37-39°C / 98-102°F), NOT rubbing**.
7. **Rewarming Shock** → **Hypotension due to vasodilation after rapid warming**. Must monitor BP and give fluids.
8. **Hypothermia & Cardiac Arrest** → If a patient is **hypothermic (<30°C)** and in cardiac arrest, **defibrillation & medications are ineffective** until rewarming is done.
9. **Risk Factors for Hypothermia** → **Elderly, alcohol use, malnutrition, hypothyroidism, sepsis, cold exposure**.
10. **Treatment of Hypothermia** → **Passive (blankets, warm room) for mild, active external (warm air, heating pads) for moderate, active internal (warm IV fluids, peritoneal lavage, ECMO) for severe**.

Drowning

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1. **Drowning Definition** → **Respiratory impairment due to submersion in liquid**, leading to **hypoxia**.
2. **Freshwater vs. Saltwater Drowning** → Both lead to **hypoxemia**, but **freshwater causes hemodilution & hemolysis**, while **saltwater causes pulmonary edema**.
3. **Dry vs. Wet Drowning** → **Wet drowning** (most common) involves **aspiration of fluid** into lungs, while **dry drowning** has **laryngospasm preventing aspiration**.
4. **Primary Cause of Death** → **Hypoxia and subsequent cardiac arrest** due to **airway obstruction, pulmonary injury, or neurogenic causes**.
5. **Complications** → **ARDS, pulmonary edema, cerebral edema, metabolic acidosis, hypothermia**.
6. **Resuscitation in Drowning** → **Immediate BLS/ACLS** with **early oxygenation and ventilation**. Intubation if needed.
7. **Hypothermia in Drowning Victims** → **"No one is dead until warm and dead"** → Continue resuscitation until **core temperature >32°C (89.6°F)**.
8. **Surfactant Washout in Drowning** → **Leads to ARDS-like picture** with alveolar collapse & non-cardiogenic pulmonary edema.
9. **Management** → **Oxygen, PEEP, mechanical ventilation if needed, treat hypothermia, correct acidosis**.
10. **Cervical Spine Injury Consideration** → If **diving-related or trauma is suspected**, perform **C-spine immobilization** before airway management.

Electrical Injuries

1. **Types of Electrical Injuries** → **Low-voltage (<1000V, e.g., household current) and high-voltage (>1000V, e.g., power lines, lightning strikes).**
2. **Primary Cause of Death** → **Cardiac arrhythmias** (asystole, ventricular fibrillation).
3. **High-Voltage Injuries** → Cause **deep tissue damage**, compartment syndrome, and **rhabdomyolysis** due to intense heat.
4. **Cardiac Effects** → **AC (Alternating Current) → ventricular fibrillation** (household current), **DC (Direct Current) → asystole** (lightning, batteries).
5. **Neurological Effects** → **Peripheral nerve damage, seizures, spinal cord injury** (especially in lightning injuries).
6. **Cutaneous Findings** → **Entry and exit wounds** (indicating internal damage), but skin appearance **does not correlate** with severity.
7. **Myoglobinuria & Rhabdomyolysis** → Due to muscle breakdown → can cause **acute kidney injury (AKI)**. **Aggressive IV fluids** are required.

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8. **Compartment Syndrome Risk** → **High-voltage burns** → cause **deep tissue edema** → require **fasciotomy if necessary**.
9. **Respiratory Effects** → **Diaphragmatic paralysis** or **laryngeal spasm** from direct electrical injury to the brainstem.
10. **Gastrointestinal Effects** → Risk of **delayed bowel perforation** if abdominal muscles are affected.
11. **Management of Electrical Injury** → ABCs first → **EKG for arrhythmias**, **IV fluids for rhabdomyolysis**, monitor for **compartment syndrome & AKI**.
12. **Lightning Strike Injuries** → Can cause **cardiac arrest, temporary paralysis (keraunoparalysis), ruptured tympanic membranes**, and **autonomic dysfunction**.

High Altitude Sickness

1. **Cause** → **Hypobaric hypoxia** due to **low atmospheric oxygen** at elevations **>2500m (8200 ft)**.
2. **Types of Altitude Sickness** →
    - **Acute Mountain Sickness (AMS)** → Mild, self-limiting.
    - **High-Altitude Cerebral Edema (HACE)** → Life-threatening brain swelling.
    - **High-Altitude Pulmonary Edema (HAPE)** → Severe non-cardiogenic pulmonary edema.
3. **Acute Mountain Sickness (AMS)** → Symptoms include **headache, nausea, dizziness, insomnia**, and **fatigue** within **6-12 hours** of ascent.
4. **HACE Symptoms** → **AMS + altered mental status, ataxia, confusion, seizures, coma**. Requires **immediate descent**.
5. **HAPE Symptoms** → **Dyspnea at rest, cough, pink frothy sputum, cyanosis, tachycardia, tachypnea**. Leading cause of altitude-related death.
6. **HAPE Pathophysiology** → **Hypoxic pulmonary vasoconstriction** → leads to **pulmonary hypertension and capillary leakage** → pulmonary edema.
7. **Prevention of Altitude Sickness** → **Gradual ascent (<500m/day above 3000m)**, adequate hydration, avoid alcohol.
8. **AMS Treatment** → **Acetazolamide** (carbonic anhydrase inhibitor) to promote metabolic acidosis and increase ventilation. **Ibuprofen for headache**.
9. **HACE & HAPE Treatment** → **Immediate descent, supplemental oxygen, hyperbaric chamber** if descent is not possible. **Dexamethasone for HACE, Nifedipine for HAPE**.
10. **Who is at Higher Risk?** → Rapid ascent, prior altitude sickness, obesity, respiratory diseases.

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11. **Golden Rule** → If symptoms **worsen, descend immediately**; **never ascend with symptoms** of altitude sickness.

Decompression Sickness (DCS)

1. **Cause** → Rapid ascent from **high-pressure environments (e.g., diving, high-altitude flights)** leads to **nitrogen bubble formation** in blood and tissues.
2. **Pathophysiology** → At **high pressure, nitrogen dissolves in tissues** → rapid ascent causes **gas to come out of solution, forming bubbles** → tissue damage and embolism.
3. **Types of Decompression Sickness** →
    - **Type I (Mild)** → Skin & musculoskeletal symptoms.
    - **Type II (Severe)** → Neurological, pulmonary, or cardiovascular involvement.
4. **Type I DCS Symptoms** → **"The Bends"** → Joint and muscle pain, skin mottling, pruritus.
5. **Type II DCS Symptoms** → **"The Chokes"** (pulmonary symptoms: dyspnea, chest pain, cough), **"The Staggers"** (neurological symptoms: vertigo, confusion, paralysis), spinal cord involvement.
6. **Neurological DCS** → Can mimic **stroke or spinal cord injury** → presents as **weakness, numbness, confusion, seizures, loss of bladder/bowel control**.
7. **Pulmonary DCS** → **Nitrogen gas emboli in pulmonary vessels** → leads to **severe hypoxia, respiratory distress, and circulatory collapse**.
8. **Management of DCS** →
    - **Immediate 100% oxygen** (helps wash out nitrogen).
    - **Hyperbaric oxygen therapy (HBOT)** in a recompression chamber is the definitive treatment.
    - **IV fluids** to prevent dehydration.
9. **Prevention of DCS** → **Slow, controlled ascent** after diving, using **decompression stops** to allow nitrogen elimination.
10. **Flying After Diving Rule** → Wait at least **12-24 hours before air travel** to prevent altitude-induced DCS.
11. **Differentiation from Arterial Gas Embolism (AGE)** → **AGE occurs immediately after surfacing** (due to pulmonary barotrauma), while **DCS develops over minutes to hours**.

Chromosomal Disorders

Autosomal Trisomies

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1. **Trisomy 21 (Down Syndrome) → Most common chromosomal disorder**, caused by **meiotic nondisjunction**. Features: **intellectual disability, flat facies, epicanthal folds, single palmar crease, congenital heart defects (AVSD), increased risk of leukemia & Alzheimer's**.
2. **Trisomy 18 (Edwards Syndrome) → Clenched fists with overlapping fingers, rocker-bottom feet, congenital heart defects (VSD), prominent occiput, severe intellectual disability**. **Death usually within 1 year**.
3. **Trisomy 13 (Patau Syndrome) → Midline defects → holoprosencephaly, cleft lip/palate, polydactyly, severe intellectual disability, congenital heart defects**.

Sex Chromosome Abnormalities

4. **Turner Syndrome (45, XO) → Short stature, webbed neck, lymphedema, broad chest, coarctation of aorta, primary amenorrhea, streak ovaries, infertility**. **Most common cause of primary ovarian failure**.
5. **Klinefelter Syndrome (47, XXY) → Tall male, gynecomastia, small testes, infertility, mild intellectual disability**. **Increased FSH & estrogen, decreased testosterone**.
6. **XYY Syndrome (47, XYY) → Tall male, normal fertility, aggressive behavior** (controversial), **learning disabilities**.
7. **XXX Syndrome (47, XXX) → Mild intellectual disability**, often asymptomatic, normal fertility.

Microdeletion Syndromes

8. **Cri-du-chat Syndrome (5p deletion) → High-pitched cry (like a cat), microcephaly, intellectual disability, cardiac defects**.
9. **Williams Syndrome (7q deletion) → Elfin facies, extreme friendliness, intellectual disability, supravalvular aortic stenosis, hypercalcemia**.
10. **DiGeorge Syndrome (22q11.2 deletion) → CATCH-22 → Cardiac defects, Abnormal facies, Thymic aplasia (T-cell deficiency), Cleft palate, Hypocalcemia**.

Single Gene Defects

Autosomal Dominant Disorders

1. **Marfan Syndrome → FBN1 gene mutation (Fibrillin-1) → tall stature, lens dislocation (upward), aortic aneurysm/dissection, mitral valve prolapse, hypermobile joints**.
2. **Huntington Disease → CAG trinucleotide repeat in HTT gene (Chromosome 4) → chorea, dementia, psychiatric symptoms, caudate atrophy, anticipation**.
3. **Neurofibromatosis Type 1 (NF1) → NF1 gene mutation (Chromosome 17, neurofibromin) → café-au-lait spots, neurofibromas, Lisch nodules (iris hamartomas), optic glioma**.

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4. **Neurofibromatosis Type 2 (NF2) → NF2 gene mutation (Chromosome 22, merlin protein) → bilateral vestibular schwannomas, cataracts, meningiomas**.
5. **Familial Hypercholesterolemia → LDL receptor mutation → early-onset atherosclerosis, tendon xanthomas, xanthelasmas**.

Autosomal Recessive Disorders

6. **Cystic Fibrosis → CFTR gene mutation (Chromosome 7, ΔF508 deletion) → thick mucus, recurrent lung infections, pancreatic insufficiency, meconium ileus, infertility (absent vas deferens in males)**.
7. **Phenylketonuria (PKU) → Phenylalanine hydroxylase deficiency → musty odor, intellectual disability, fair skin, seizures** (managed by a **low-phenylalanine diet**).
8. **Sickle Cell Disease → HBB gene mutation (Glutamic acid → Valine substitution) → sickled RBCs, vaso-occlusive crises, dactylitis, autosplenectomy, stroke, priapism**.
9. **Tay-Sachs Disease → HEXA gene mutation (Hexosaminidase A deficiency) → cherry-red macula, neurodegeneration, NO hepatosplenomegaly**.
10. **Wilson Disease → ATP7B gene mutation (Chromosome 13) → Kayser-Fleischer rings, hepatolenticular degeneration, psychiatric symptoms, low ceruloplasmin**.

X-Linked Disorders

11. **Duchenne Muscular Dystrophy → DMD gene mutation (Dystrophin) → Gower sign, pseudohypertrophy of calves, early death due to cardiomyopathy**.
12. **Hemophilia A & B → Factor VIII (A) or Factor IX (B) deficiency → hemarthrosis, easy bruising, prolonged PTT**.
13. **Fragile X Syndrome → FMR1 gene (CGG repeat expansion) → intellectual disability, large ears, macroorchidism, hyperactivity**.
14. **G6PD Deficiency → G6PD enzyme defect → hemolysis triggered by fava beans, infections, sulfa drugs**.
15. **Lesch-Nyhan Syndrome → HGPRT deficiency → self-mutilation, gout, dystonia, hyperuricemia**.

Sex-Linked Disorders

X-Linked Recessive Disorders (More common in males, no male-to-male transmission)

1. **Duchenne Muscular Dystrophy (DMD) → Dystrophin gene deletion → Gower's sign, pseudohypertrophy of calves, cardiomyopathy, early wheelchair dependence**.
2. **Becker Muscular Dystrophy → Partially functional dystrophin mutation → similar to DMD but milder and later onset**.

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3. **Hemophilia A & B → Factor VIII (A) or Factor IX (B) deficiency → hemarthrosis, easy bruising, prolonged PTT, normal PT and platelet count**.
4. **Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency → Oxidative stress triggers hemolysis → Heinz bodies, bite cells, triggered by fava beans, sulfa drugs, infections**.
5. **Lesch-Nyhan Syndrome → HGPRT deficiency → Self-mutilation, hyperuricemia, gout, dystonia, intellectual disability**.
6. **Bruton Agammaglobulinemia (X-Linked Agammaglobulinemia - XLA) → Defective BTK gene (B-cell maturation defect) → recurrent bacterial infections, absent B cells, low IgG, absent lymph nodes/tonsils**.
7. **Wiskott-Aldrich Syndrome → WAS gene mutation (T-cell cytoskeleton defect) → "WATER": Wiskott-Aldrich, Thrombocytopenia, Eczema, Recurrent infections**.
8. **Chronic Granulomatous Disease (CGD) → NADPH oxidase deficiency → recurrent catalase-positive infections (S. aureus, Aspergillus), negative nitroblue tetrazolium test**.
9. **Hunter Syndrome → Iduronate-2-sulfatase deficiency → Coarse facies, hepatosplenomegaly, aggressive behavior, NO corneal clouding (unlike Hurler Syndrome)**.
10. **Red-Green Color Blindness → Defective opsin genes → X-linked inheritance, inability to distinguish red and green colors**.

X-Linked Dominant Disorders (More common in females, affected fathers transmit to all daughters)

11. **Fragile X Syndrome → CGG trinucleotide repeat in FMR1 gene → macroorchidism, intellectual disability, autism, long face, large ears**.
12. **Rett Syndrome → MECP2 gene mutation → normal development until 6-18 months, then regression, loss of speech, hand-wringing, seizures, seen only in females**.
13. **Alport Syndrome → Collagen IV mutation → hereditary nephritis, hearing loss, ocular abnormalities ("Can't see, can't pee, can't hear a bee")**.

Polygenic Inheritance Disorders

Polygenic inheritance involves multiple genes contributing to a trait or disorder, often influenced by environmental factors.

1. **Diabetes Mellitus (Type 1 & Type 2)** → Strong genetic component, but **Type 1 has HLA-DR3/DR4 association**, while **Type 2 is linked to obesity and lifestyle**.
2. **Hypertension → Multifactorial inheritance** with genetic predisposition and environmental triggers like salt intake and stress.
3. **Coronary Artery Disease (CAD)** → Polygenic risk with contributions from **hypertension, dyslipidemia, smoking, and diabetes**.

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4. **Schizophrenia** → Strong **polygenic** component with an **increased risk in first-degree relatives**.
5. **Bipolar Disorder** → **Highly heritable**, but no single gene identified; multiple genes interact with environmental factors.
6. **Epilepsy** → Multiple genetic factors involved, with varying inheritance patterns depending on the epilepsy syndrome.
7. **Glaucoma** → **Open-angle glaucoma has a polygenic basis** with family history being a strong risk factor.
8. **Psoriasis** → Polygenic disorder involving immune dysregulation and environmental triggers (stress, infections, trauma).
9. **Multiple Sclerosis (MS)** → **HLA-DR2 association**, polygenic inheritance with environmental triggers like **low vitamin D and infections**.
10. **Rheumatoid Arthritis (RA)** → **HLA-DR4 association**, polygenic with autoimmune components.
11. **Cleft Lip and Palate** → **Polygenic and multifactorial** with environmental influences like maternal smoking and folate deficiency.
12. **Pyloric Stenosis** → **Polygenic inheritance with a male predominance**, leading to projectile vomiting in infants.
13. **Asthma** → Polygenic, with genes affecting **airway hyperresponsiveness and inflammation** combined with environmental factors.

Marfan Syndrome

1. **Marfan syndrome** is an **autosomal dominant** disorder caused by a mutation in the **FBN1 gene (Fibrillin-1)** on **chromosome 15**.
2. **Skeletal features**: **Tall stature, long limbs (arachnodactyly), pectus excavatum/carinatum, hypermobile joints, scoliosis**.
3. **Ocular involvement**: **Lens dislocation (upward and outward - ectopia lentis)**, myopia, retinal detachment risk.
4. **Cardiovascular complications**: **Aortic root dilation → aortic dissection**, mitral valve prolapse (MVP), and increased risk of sudden death.
5. **Aortic complications** are the **leading cause of death** in Marfan syndrome.
6. **Steinberg (thumb) sign** and **Walker-Murdoch (wrist) sign** are clinical tests for **long extremities** in Marfan syndrome.
7. **Differential diagnosis** includes **Loeys-Dietz syndrome (TGF-beta receptor mutation, more severe vascular issues)** and **Homocystinuria (downward lens dislocation, thrombosis, intellectual disability)**.

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8. **Management**: **Beta-blockers (reduce aortic stress)**, lifestyle modifications (avoid contact sports), and elective aortic surgery if dilation progresses.
9. **Pregnancy risk**: Increased aortic dissection risk due to **hemodynamic changes**.
10. **Ghent criteria** are used for the **clinical diagnosis** of Marfan syndrome.

Obesity

1. **Obesity is defined as BMI ≥ 30 kg/m².**
2. **Increased visceral (central) fat is more strongly associated with cardiovascular risk than subcutaneous fat.**
3. **Most common secondary causes of obesity include hypothyroidism, Cushing's syndrome, and insulin resistance.**
4. **Leptin resistance is a key factor in obesity, leading to impaired appetite suppression.**
5. **First-line treatment for obesity is lifestyle modification (diet + exercise).**
6. **Pharmacologic therapy (e.g., orlistat, GLP-1 agonists like semaglutide) is considered for BMI ≥ 30 or BMI ≥ 27 with comorbidities.**
7. **Bariatric surgery is indicated for BMI ≥ 40 or BMI ≥ 35 with severe obesity-related conditions.**
8. **Obesity is a major risk factor for metabolic syndrome, type 2 diabetes, hypertension, and cardiovascular disease.**
9. **Obstructive sleep apnea is strongly associated with obesity and increases cardiovascular mortality.**
10. **Non-alcoholic fatty liver disease (NAFLD) is a common obesity-related complication that can progress to cirrhosis.**
11. **Obesity increases the risk of certain cancers, including breast, colon, and endometrial cancer.**
12. **Weight loss of 5–10% significantly reduces obesity-related complications.**
13. **Central obesity is measured by waist circumference (>102 cm in men, >88 cm in women).**
14. **Hyperinsulinemia and insulin resistance play a key role in obesity-related metabolic dysfunction.**
15. **Long-term weight maintenance requires sustained caloric restriction and increased physical activity.**

By Muhammad Taha(RMC,Final-Yr)

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