

DISORDERS AND DISEASES COMMONLY COVERED ON THE MCAT

GENETIC DISORDERS	1
NUTRITIONAL DISORDERS	4
INFECTIOUS DISEASES.....	5
OTHER COMMON DISEASES	6

GENETIC DISORDERS**Single-Gene Disorders**

This category encompasses a wide variety of diseases and disorders, ranging in frequency from very rare to fairly frequent, having a mild to lethal impact on the patient. All are caused by variation at a single locus. They can be classified into six categories, based on what type of chromosome they are found on and their inheritance pattern.

Autosomal dominant

- Only one mutated copy of the gene is needed for a person to be affected by an autosomal dominant disorder.
- Each affected person usually has one affected parent.
- There is a 50% chance that a child will inherit the mutated gene.
- Many disease conditions that are autosomal dominant have low penetrance, which means that, although only one mutated copy is needed, a relatively small proportion of those who inherit that mutation go on to develop the disease, often later in life.
- Examples include: Huntingtons disease, Neurofibromatosis 1, HBOC syndrome, Hereditary nonpolyposis colorectal cancer

Autosomal recessive

- Two copies of the gene must be mutated for a person to be affected by an autosomal recessive disorder.
- An affected person usually has unaffected parents who each carry a single copy of the mutated gene (and are referred to as carriers).
- Two unaffected people who each carry one copy of the mutated gene have a 25% chance with each pregnancy of having a child affected by the disorder.
- Examples include: Cystic fibrosis, Sickle cell anemia, Tay-Sachs disease, Spinal muscular atrophy, Muscular dystrophy

X-linked dominant

- X-linked dominant disorders are caused by mutations in genes on the X chromosome.
- Only a few disorders have this inheritance pattern.

- Females are more frequently affected than males, and the chance of passing on an X-linked dominant disorder differs between men and women. The sons of a man with an X-linked dominant disorder will not be affected, and his daughters will all inherit the condition. A woman with an X-linked dominant disorder has a 50% chance of having an affected daughter or son with each pregnancy.
- Some X-linked dominant conditions, such as Aicardi Syndrome, are fatal to boys, therefore only girls have them (and boys with Klinefelter Syndrome).
- Examples include: Hypophosphatemia, Aicardi Syndrome

X-linked recessive

- X-linked recessive disorders are also caused by mutations in genes on the X chromosome.
- Males are more frequently affected than females, and the chance of passing on the disorder differs between men and women. The sons of a man with an X-linked recessive disorder will not be affected, and his daughters will carry one copy of the mutated gene. With each pregnancy, a woman who carries an X-linked recessive disorder has a 50% chance of having sons who are affected and a 50% chance of having daughters who carry one copy of the mutated gene.
- Examples include: Hemophilia A, Duchenne muscular dystrophy, Color blindness, Turner Syndrome

Y-linked

- Y-linked disorders are caused by mutations on the Y chromosome.
- Only males can get them, and all of the sons of an affected father are affected.
- Since the Y chromosome is very small, Y-linked disorders only cause infertility, and may be circumvented with the help of some fertility treatments.

Mitochondrial

- This type of inheritance, also known as maternal inheritance, applies to genes in mitochondrial DNA.
- Because only egg cells contribute mitochondria to the developing embryo, only females can pass on mitochondrial conditions to their children (both males and females).
- Examples include: Leber's Hereditary Optic Neuropathy (LHON)

Multifactorial and Polygenic Disorders

This category encompasses disorders that are not the result of simple Mendelian inheritance, but rather the result of multifactorial or polygenic causes. They are usually associated with the effects of multiple genes, often in combination with lifestyle and environmental factors.

- Although complex disorders often cluster in families, they do not have a clear-cut pattern of inheritance. This makes it difficult to determine a person's risk of inheriting or passing on these disorders. Complex disorders are also difficult to study and treat because the specific factors that cause most of these disorders have not yet been identified.
- Examples of multifactorial disorders include: heart disease and diabetes
- Examples of polygenic disorders may include autism

Chromosomal Disorders

Changes that affect entire chromosomes or segments of chromosomes can cause problems with growth, development, and function of the body's systems. These changes can affect many genes along the chromosome and alter the proteins made by those genes. Conditions caused by a change in the number or structure of chromosomes are known as chromosomal disorders.

Aneuploidy

- Chromosomal conditions caused by changes in the number of chromosomes.
- These changes are not inherited, but occur as random events during the formation of reproductive cells (ova and sperm cells). An error in cell division called nondisjunction results in reproductive cells with an abnormal number of chromosomes.
- For example, a reproductive cell may accidentally gain or lose one copy of a chromosome. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra (trisomy) or missing chromosome (monosomy) in each of the body's cells.
- Examples of monosomy include: X0 (Turner Syndrome)
- Examples of trisomy include: Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), Trisomy 9, Trisomy 8 (Warkany syndrome 2), and Trisomy 16 (the most common trisomy in humans, occurring in more than 1% of pregnancies; usually results in spontaneous miscarriage in the first trimester)
- Examples of trisomy involving sex chromosomes include: XXX (Triple X syndrome), XXY (Klinefelter's syndrome), XYY (XYY syndrome)

Ring Chromosomes

- A ring chromosome is a chromosome that is formed when the ends have been lost, and the arms fuse together to form a ring. Ring chromosomes may form in cells following genetic damage by mutagens like radiation, they may also arise spontaneously during development.
- Human genetic disorders can be caused by spontaneous ring chromosome formation; although ring chromosomes are very rare, they have been found in for all human chromosomes.

- Examples include: Ring Chromosome 20 syndrome where a ring formed by one copy of chromosome 20 is associated with epilepsy; ring chromosome 14 and ring chromosome 13 syndrome are associated with mental retardation and dysmorphic facial features; ring chromosome 15 is associated with mental retardation, dwarfism and microcephaly

Aberrent Chromosomal Structure

- These changes are caused by the breakage and reunion of chromosome segments when an egg or sperm cell is formed or in early fetal development. Pieces of DNA can be rearranged within one chromosome, or transferred between two or more chromosomes.
- The effects of structural changes depend on their size and location. Many different structural changes are possible; some cause medical problems, while others may have no effect on a person's health.
- Although it is possible to inherit some types of chromosomal abnormalities, most chromosomal disorders are not passed from one generation to the next.

NUTRITIONAL DISORDERS

Avitaminosis

- Any disease caused by chronic or long-term vitamin deficiency or caused by a defect in metabolic conversion, such as tryptophan to niacin.
- Vitamin A deficiency causes xerophthalmia (night blindness), xerosis (replacement of mucus-secreting cells with keratin-producing cells), corneal ulcers/liquefaction (interruption of glycoprotein synthesis in eyes), decreases production of human growth hormone
- Vitamin B1 (thiamine) deficiency causes neurodegeneration, wasting and eventually, death. Well-known syndromes include Wernicke-Korsakoff syndrome and beriberi, diseases also common in chronic abusers of alcohol
- Vitamin B3 (niacin) deficiency contributes to pellagra (along with a tryptophan deficiency)
- Vitamin B12 (cyanocobalamin) deficiency leads to various forms of anemia
- Vitamin C (ascorbic acid) deficiency leads to scurvy, a disease involving the breakdown of collagen-containing structures in the body (resulting in excessive bruising, spongy gums, hair loss, poor healing of wounds, bleeding from mucos-membranes and anemia
- Vitamin D deficiency causes rickets, a softening of the bone (resulting in bow legs, increased rate of fractures, and impaired growth)
- Vitamin K deficiency causes bleeding, cartilage calcification, and deposition of calcium ions in blood vessels, by impairing the formation of the amino acid glutamate (and therefore glutamate-containing proteins)

Hypervitaminosis

- Vitamin poisoning refers to a condition of high storage levels of vitamins, which can lead to toxic symptoms.
- High dosage vitamin A, high dosage, slow release vitamin B₃ and very high dosage vitamin B₆ alone without vitamin B complex are sometimes associated with vitamin side effects that usually rapidly cease with supplement reduction or cessation. Hypervitaminosis A can lead to birth defects, liver abnormalities, reduced bone mineral density (osteoporosis), and loss of muscular coordination.
- Conversely, certain vitamins do not produce toxicity in excess levels. Vitamin C has been used in dosages over 100,000 mg for serious illness – over 1000 times the daily recommended intake – without ill effects (except a pronounced laxative effect).
- High doses of mineral supplements can also lead to side effects and toxicity. Mineral supplement poisoning does occur occasionally due to excessive and unusual intake of iron containing supplements, including some multivitamins

INFECTIOUS DISEASES

Infectious diseases are caused by a biological agent such as a virus, bacterium, fungi or parasite. Infectious diseases require an agent and a mode of transmission (or vector). For example, malaria, which is caused by Plasmodial parasites (the agent), cannot affect humans unless the vector, the Anopheles mosquito, is transmits the agent to the human bloodstream. The vector does not have to be biological. Many infectious diseases are transmitted by droplets which enter the airway (e.g. common cold or tuberculosis). Modes of transmission may include droplet contact, fecal-oral transmission, sexual transmission, oral-sexual transmission, oral transmission, transmission by direct contact, vertical transmission, and latrogenic transmission.

Viral Infectious Diseases

- Diseases caused by viruses; treated with anti-virals; often prevented with vaccines
- Common examples include AIDS; Chickenpox; Common cold; Dengue fever; Ebola; Hepatitis; Herpes simplex and zoster; Influenza; Measles; Mumps; Polio; Rabies; Rubella; Smallpox; West Nile disease; Yellow fever

Bacterial Infectious Diseases

- Diseases caused by bacteria; treated with antibiotics; many disease strains are antibiotic resistant
- Common examples include Anthrax; Cholera; Diphtheria; Gonorrhea; Leprosy; Lyme Disease; Pertussis (Whooping Cough); Plague; Scarlet Fever; Syphilis; Tetanus; Tuberculosis; Typhus

Parasitic Infectious Diseases

- Diseases caused by parasites; treated with anti-infective agents which target protozoa and other parasites
- Common examples include Chagas Disease; Fascioliasis; Giardiasis; Malaria; Pinworm Infection; Scabies; Schistosomiasis; Toxoplasmosis; Trichinosis

Fungal Infectious Diseases

- Diseases caused by a fungus; can be superficial, cutaneous, subcutaneous or systemic
- Treated with anti-fungals
- Common examples include Blastomycosis; Candidiasis (Yeast infections and Thrush); Tinea pedis (Athlete's foot)

Prion Infectious Diseases

- Diseases caused by an abnormally-structured host protein known as a prion (short for **proteinaceous infectious particle**)
- There are no known treatments for prion-related diseases
- Common examples include Bovine spongiform encephalopathy (Mad cow); Creutzfeldt-Jakob disease; Kuru

OTHER COMMON DISEASES

The following is a list of common diseases and conditions that are not infectious and not genetic (although people may have a genetic predisposition, that is, they tend to run in families).

Cancer

- Cancer is a class of diseases characterized by uncontrolled division of cells and the ability of these cells to invade other tissues, either by direct growth into adjacent tissue through *invasion* or by implantation into distant sites by *metastasis*.
- Metastasis is defined as the stage in which cancer cells are transported through the bloodstream or lymphatic system.
- Cancer may affect people at all ages, but risk tends to increase with age, due to the fact that DNA damage becomes more apparent in aging DNA.
- There are many types of cancer. Severity of symptoms depends on the site and character of the malignancy and whether there is metastasis. A definitive diagnosis usually requires the histologic examination of tissue by a pathologist. This tissue is obtained by biopsy or surgery.
- Most cancers can be treated and some cured, depending on the specific type, location, and stage. Once diagnosed, cancer is usually treated with a combination

of surgery, chemotherapy and radiotherapy. As research develops, treatments are becoming more specific for the type of cancer pathology. Drugs that target specific cancers already exist for several cancers.

- Many forms of cancer are associated with exposure to environmental factors such as tobacco smoke, radiation, alcohol and certain viruses. While some of these can be avoided, there is no known way to entirely avoid the disease.

Diabetes

- Diabetes mellitus is a disease characterized by persistent hyperglycemia (high blood sugar levels), resulting either from inadequate secretion of the hormone insulin, an inadequate response of target cells to insulin, or a combination of these factors.
- The three most common patterns of diabetes have been recognized over the last thirty years as *type 1*, *type 2* and *gestational diabetes* (or *type 3*), although these three "types" of diabetes are more accurately considered patterns of pancreatic failure rather than single diseases.

Arthritis

- Arthritis is a group of conditions where there is damage caused to the joints of the body. Arthritis is the leading cause of disability over the age of 65.
- There are many forms of arthritis, each of which has a different cause. Rheumatoid arthritis and psoriatic arthritis are autoimmune diseases in which the body is attacking itself. Septic arthritis is caused by joint infection. Gouty arthritis is caused by deposition of uric acid crystals in the joint and subsequent inflammation. The most common form of arthritis, osteoarthritis is also known as degenerative joint disease and occurs following trauma to the joint, following an infection of the joint or simply as a result of aging.

Alzheimer's Disease

- Alzheimer's disease is a neurodegenerative disease characterized by progressive cognitive deterioration together with declining activities of daily living and neuropsychiatric symptoms or behavioral changes. It is the most common cause of dementia.
- This pathological process consists principally of neuronal loss or atrophy, principally in the temporoparietal cortex, but also in the frontal cortex, together with an inflammatory response to the deposition of amyloid plaques and neurofibrillary tangles.

Parkinson's

- Parkinson's disease is a degenerative disorder of the central nervous system that affects the control of muscles, and so may affect movement, speech and posture.

- It is often characterized by muscle rigidity, tremor, a slowing of physical movement (bradykinesia), and in extreme cases, a loss of physical movement (akinesia).
- The primary symptoms are the results of excessive muscle contraction, normally caused by the insufficient formation and action of dopamine, which is produced in the dopaminergic neurons of the brain. Secondary symptoms may include high level cognitive function and subtle language problems. PD is both chronic, meaning it persists over a long period of time, and is progressive.

Heart and circulatory-related diseases

Heart disease is one of a number of different diseases which afflict the heart. Circulatory diseases refer to diseases that affect the arteries and veins. Together they are known as cardiovascular diseases. Pulmonary diseases are those that affect the lungs and gas exchange system.

Coronary heart disease

- Coronary heart disease, also called coronary artery disease and atherosclerotic heart disease, is the end result of the accumulation of atheromatous plaques within the walls of the arteries that supply the myocardium (the muscle of the heart).
- While the symptoms and signs of coronary heart disease are noted in the advanced state of disease, most individuals with coronary heart disease show no evidence of disease for decades as the disease progresses before the first onset of symptoms, often a "sudden" heart attack, finally arise. After decades of progression, some of these atheromatous plaques may rupture and (along with the activation of the blood clotting system) start limiting blood flow to the heart muscle. The disease is the most common cause of sudden death.

Pulmonary heart disease

- Cor pulmonale is a medical term used to describe a change in structure and function of the right ventricle of the heart as a result of a respiratory disorder. To be classified as cor pulmonale, the cause must originate in the pulmonary circulation. Vascular remodelling of the pulmonary circulation as a result of tissue damage (causes including disease, hypoxic injury, chemical agents, etc.) or chronic hypoxic vasoconstriction are two major causes.

Hypertension

- Hypertension or high blood pressure is a medical condition wherein the blood pressure is chronically elevated.
- Persistent hypertension is one of the risk factors for strokes, heart attacks, heart failure and arterial aneurysm, and is a leading cause of chronic renal failure.

Hypotension

- Hypotension refers to abnormally low blood pressure.
- It is often associated with shock, though not necessarily indicative of it.
- The cardinal symptom of hypotension is lightheadedness or dizziness. If the blood pressure is sufficiently low, seizures can occur

Cardiac Arrest

- A cardiac arrest, or circulatory arrest, is the abrupt cessation of normal circulation of the blood due to failure of the heart to contract effectively during systole.

Aneurysm

- An aneurysm is a localized dilation or ballooning of a blood vessel by more than 50% of the diameter of the vessel and can lead to instant death.
- Aneurysms most commonly occur in arteries at the base of the brain and in the aorta (this is an aortic aneurysm).
- This bulge in a blood vessel, much like a bulge on an over-inflated innertube, can lead to death at anytime. The larger an aneurysm becomes, the more likely it is to burst.

Pulmonary edema

- Pulmonary edema is swelling and/or fluid accumulation in the lungs. It leads to impaired gas exchange and may cause respiratory failure.
- It is due to either failure of the heart to remove fluid from the lung circulation or due to a direct injury to the lungs. Treatment depends on the cause, but focuses on maximizing respiratory function and removing the cause.

Asthma

- Asthma is a disease of the respiratory system in which the airways constrict, become inflamed, and are lined with excessive amounts of mucus. This airway narrowing causes symptoms such as wheezing, shortness of breath, chest tightness, and coughing, which respond to bronchodilators. Between episodes, most patients feel fine.
- Asthma occurs as a response to one or more "triggers," such as exposure to an environmental stimulant (or allergen), cold air, exercise, or emotional stress.
- In children, the most common triggers are viral illnesses such as those that cause the common cold.