Module 2: Chronic Disease (Battle text Chapters: 3, 12, 13, 33, 34, 35, 36, 38)

Genetics, Cardiovascular Disease, Diabetes, Respiratory Disease, Biology of Addiction and Mental Illness

**Chapter 3 - Genetics And Public Health**

1. **Alleles** -- Alternate versions of the same gene. One allele is inherited from each parent. A gene might have multiple alleles, but each individual can have only two different alleles for any given gene.
2. **Autosomal Dominant Trait** -- A trait that is carried on an autosome (a non-sex chromosome) and requires only one copy of the associated allele for expression of the phenotype.
3. **Autosomal Recessive Trait** -- A trait that is carried on an autosome (a non-sex chromosome) and required two copies of the associated allele for expression of the phenotype.
4. **Chromosomes** -- Thread-like structures in the nucleus of a cell that are made of DNA and structural proteins. Human cells other than egg and sperm normally have 46 chromosomes (23 pairs). Egg and sperm cells have 23 chromosomes.
5. **DNA (deoxyribonucleic acid)** -- The universal genetic material; the information molecule that carries hereditary information from one generation of cells to the next and from one generation of individuals to the next. Genes are made of DNA, which is the molecular basis of heredity.
6. **ELSI** -- The acronym for “ethical, legal, and social implications” of human genome research. The ELSI program began as a central component of the Human Genome Project (q.v.), with the goal to support research and education about the applications of knowledge derived from research in genetics and genomics. The term ELSI now has come to signify any such category of issues, even if they are not directly related to the Human Genome Project.
7. **Evolution** -- The change in gene frequencies in populations of organisms over time, potentially leading to the production of new species. Evolution helps to determine the genetic structure of human populations, thereby helping to determine the nature and extent of disease in those populations.
8. **First-degree Relatives** -- Parents, siblings, children.
9. **Gene** -- A segment of DNA that contains instructions for making a specific protein or proteins required by the body. Genes are found in succession along the length of chromosomes. Human beings have about 20,000 genes.
10. **Genetic Counseling** -- “The process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources and research, counseling to promote informed choices and adaptation to the risk or condition.” (From the National Society of Genetic Counselors).
11. **Genetics** -- The branch of science concerned with the means and consequences of inherited biological variation.
12. **Genome** -- All of the DNA of a given organism. One speaks, for example, of the human genome or the mouse genome.
13. **Genomics** -- The study of whole genomes, usually focusing on extensive DNA sequences.
14. **Genotype** -- The genetic constitution of an organism or cell; the set of alleles inherited at a locus.
15. **Human Genome Project** -- The effort, completed in 2003, to determine the sequence of all 3.2 billions base pairs of human DNA. The results of the project are deposited in public databases that are accessible to all interested parties.
16. **Independent Assortment** -- The principle that different genes (and versions of genes, i.e., alleles) are distributed independently into egg and sperm. Independent assortment explains why it is possible to look like a certain family member, but not to have the same medical conditions or traits, and vice versa.
17. **Mendelian Inheritance** -- Relating the principles of heredity first described by Gregor Mendel, in 1865. Mendelian (single-gene) traits follow well-defined patterns of inheritance.
18. **Pedigree** -- A diagram showing the genetic relationships between members of a family that is annotated with relevant medical information. Pedigrees are used to visualize inheritance patterns and to aid in diagnosis and risk assessment.
19. **Pharmacogenetics** -- The study of genetic factors that directly influence a person’s reaction to medications.
20. **Pharmacogenomics** -- The application of genomic information (all human genes and their interactions relevant to drug response) and technology in drug development and therapy.
21. **Phenotype** -- The physical expression of a trait or disease.
22. **Public Health Genetics** -- The application of knowledge from genetics and genomics, in the context of the principles of public health, to improve the health of populations.
23. **Second-degree Relatives** -- Grandparents, grandchildren, nieces, nephews, aunts, uncles, half siblings.
24. **Segregation** -- The distribution of chromosomes during the formation of an egg or sperm. Each person has two versions of each chromosome, but can only contribute one of each pair to an egg or sperm cell.
25. **Third-degree Relatives** -- First cousins, great-grandchildren, great-grandparents.
26. **X-linked Trait** -- A trait that is carried on the X chromosome; that is, not on an autosome.

**Chapter 12 - Fetal Alcohol Syndrome: A Lifespan Perspective**

1. **Adaptive Skills** -- Skills of daily living including knowledge of basic self care, maintenance of personal hygiene, dressing, social, social judgment, and communication skills.
2. **Alcohol Related Birth Defects** -- Fetal or infant body system disorders of structure or function associated with alcohol exposure during gestation.
3. **Anomaly** -- Abnormal development of a body system.
4. **Biological Plausibility** -- Epidemiological findings of association between the proposed causal agent and the biological disorder supported by existing knowledge of biological systems; also termed coherence. If biological knowledge is absent, replication studies in other populations may be necessary to prove biological plausibility.
5. **Body System** -- Group of organs that function together, e.g. cardiovascular system, gastrointestinal system, neurological system, respiratory system, etc.
6. **Dose Response** -- As the exposure to the proposed causal agent increases the response (risk) of the biological disorder increases.
7. **Fetal Alcohol Effects (FAE)** -- (sometimes called partial FAS or, less commonly, possible Fetal Alcohol Effects) Includes individuals with intrauterine alcohol exposure who meet some of the facial features and at least one of the other criteria of Fetal Alcohol Syndrome, including growth retardation, neurodevelopmental abnormalities, or behavioral or cognitive disorders.
8. **Fetal Alcohol Syndrome (FAS)** -- Includes individuals with a history intrauterine alcohol exposure who have the identifiable facial features, growth retardation, and neurobehavioral dysfunction.
9. **Indicated Prevention** -- Also called tertiary prevention; indicated prevention strategies are applied to populations who demonstrate the disease (disorder or behavior) to prevent further morbidity or illness.
10. **Mental Retardation** -- Intellectual deficit measured by IQ and adaptive tests. An individual must have both IQ and adaptive skills test scores less than two standard deviations below the mean (< 70) to meet diagnostic criteria for mental retardation. Mental retardation is divided into mild (70-55), moderate (54-40), severe (39-25) and profound (<25). Approximately 2.5% of the population has mental retardation; 85% of individuals with mental retardation are in the mild category.
11. **Targeted Prevention** -- Also called selective or secondary prevention; targeted prevention focuses on individuals who are at risk for a disease or disorder.
12. **Temporality** -- Timing of exposure to the proposed causal agent must occur before the disease process.
13. **Teratogen** -- Agent that causes birth defects, death, growth deficiency, and/or functional impairment.
14. **Universal Prevention** -- Also called primary prevention; universal prevention is a prevention strategy that targets the entire population (no-risk to high-risk individuals).

**Chapter 13 - Smoking, Nicotine And Addiction: Tobacco Or Health?**

1. **Addiction (or Dependence)** -- Compulsive use of a drug, even in the face of significant adverse effects and consequences.
2. **Environmental Tobacco Smoke (or Second Hand Smoke)** -- Cigarette smoke blown into the environment by active smokers. Proven to cause cancer, death and respiratory illnesses in non-smokers.
3. **Midbrain Reward Pathway** -- A dopaminergic pathway in the brain that is activated by certain beneficial behaviors such as eating and sex, as well as by addictive drugs, to produce a sensation of pleasure.
4. **Non-Smoking as the Norm** -- Movement that seeks to isolate smokers, discourage initiation and eliminate the proven hazards of environmental smoke by emphasizing everyone’s right to breathe air free of toxic tobacco smoke.
5. **Smoking Cessation** -- An attempt to quit smoking, either unaided (“Cold Turkey”) or assisted by behavioral and/or pharmacological treatments.
6. **Tolerance** -- A state resulting from changes in the brain that occur in response to chronic exposure to an addictive drug. An individual exhibiting tolerance to a drug requires repeated and possibly increasing amounts of that drug to feel “normal”.
7. **Withdrawal** -- Unpleasant adverse effects caused by abrupt cessation of a drug in an individual addicted to that drug.

**Chapter 33 - Hypertension And The Kidney**

1. **Aldosterone** -- A steroid hormone released from the adrenal cortex. It acts in the distal tubule and the collecting duct.
2. **Diastolic Blood Pressure** -- The bottom number is the diastolic blood pressure reading. It represents the pressure in the arteries when the heart is at rest.
3. **Diuretic** -- A drug that increases urine flow, usually by inhibiting sodium reabsorption.
4. **GFR** -- Glomerular filtration rate – The volume of filtrate formed in both kidneys and passing into the proximal tubule. In adults it averages about 120 ml/min.
5. **Nephron** -- The functional unit of the kidney. Each human kidney has about one million of these tiny tubular structures.
6. **Prehypertension** -- Systolic if the blood pressure is between 120-139mmHg and/or diastolic if the blood pressure is between 80-89mmHg in adults.
7. **Reabsorption** -- In the kidney reabsorbed solute are transported out of the fluid in the lumen of the nephron and returned to the blood.
8. **Systolic Blood Pressure** -- The top number is the systolic blood pressure reading. It represents the maximum pressure exerted when the heart contracts.

**Chapter 34 - Epidemiology Of Atherosclerosis**

1. **Atherothrombosis** -- Atherosclerotic plaque disruption with superimposed thrombosis.
2. **Automated External Defibrillator (AED)** -- A small, portable device that is attached to a person's chest with wires. The device checks the person's heart rhythm, decides if that rhythm is abnormal and gives the heart an electric shock (called a defibrillating shock) if needed to restores the hearts’ rhythm to normal.
3. **Food and Drug Administration** -- Agency of the U.S. Department of Health and Human Services responsible for protecting public health by assuring safety, efficacy, and security of human and veterinary drugs, biological products, and medical devices. The FDA is also responsible for advancing the public health.
4. **Plaque** -- Built up material on the inner lining of an artery made up of cholesterol and fatty substances. Plaque build up results in narrowing of the blood vessel.
5. **Sudden cardiac arrest** -- Unexpected death occurring less than an hour from the onset of a cardiac event. Sudden cardiac arrest usually is usually caused by an abnormal heart rhythm that develops during a heart attack. Sudden cardiac arrest is often referred to as sudden death.
6. **Thrombosis** -- Formation or presence of a blood clot inside a blood vessel.
7. **Women’s Health Initiative** -- A 15-year national health study that involved over 161,000 women aged 50-79 that focused on preventing heart disease, breast and colorectal cancer and fractures.

**Chapter 35 - Stroke: A Guide For Prevention For Patients And Families**

1. **Athersclerosis** -- Narrowing of arteries characterized by deposition of lipid, inflammation, and calcification.
2. **Hyperlipidemia** -- Elevations of lipids (cholesterol, triglycerides) in the plasma.
3. **Modifiable Risk Factor** -- Condition present within an individual which increases the risk of disease occurrence that can be changed to alter subsequent risk of disease occurrence.
4. **Non-modifiable Risk Factor** -- Condition present within an individual which increases the risk of disease occurrence but cannot be changed.
5. **Stroke** -- Sudden onset of persistent neurologic deficits resulting from vascular causes.
6. **Transient Ischemic Attack (TIA)** -- Sudden onset neurologic deficits resulting from cessation of blood flow which recover in less than 24 hours.

**Chapter 36 - Diabetes: A Public Health Pandemic**

1. **Alpha Cell** -- A type of cell in the pancreas (in areas called the islets of Langerhans). Alpha cells make and release a hormone called glucagon, which raises the level of glucose (sugar) in the blood.
2. **Beta Cell** -- A type of cell in the pancreas in areas called the Islets of Langerhans. Beta cells make and release insulin, a hormone that controls the level of glucose (sugar) in the blood.
3. **Delta Cell** -- A type of cell in the pancreas in areas called the islets of Langerhans. Delta cells make somatostatin, a hormone that is believed to control how the beta cells make and release insulin and how the alpha cells make and release glucagon.
4. **Diabetic Ketoacidosis (DKA)** -- Severe, out-of-control diabetes (high blood sugar) that needs emergency treatment. DKA is caused by a profound lack of circulating insulin. This may happen because of illness, taking too little insulin, or getting too little exercise. The body starts using stored fat for energy, and ketone bodies (acids) build up in the blood.
5. **Epidemiology** -- The study of the distribution and determinates of health-related states or events in specified populations, and the application of this study to control the health problems.
6. **Hypoglycemia** -- Too low a level of glucose (sugar) in the blood. This occurs when a person with diabetes has injected too much insulin, eaten too little food, or has exercised without extra food. A person with hypoglycemia may feel nervous, shaky, weak, or sweaty, and have a headache, blurred vision, and hunger. Taking small amounts of sugar, sweet juice, or food with sugar will usually help the person feel better within 10-15 minutes.
7. **Islets of Langerhans** -- (Pronunciation: EYE-let cells). The clumps of cells within the pancreas that include those cells that make insulin and other hormones. The cells include several subvarieties, including: alpha cells, which make glucagon; beta cells, which make insulin; delta cells, which make somatostatin; and PP cells and D1 cells, about which little is known. The islet cells appear under low-power magnification to be islands (islets) within the pancreas. First described by Dr. Paul Langerhans in 1869, whose name is now associated with these islands.
8. **Morbidity** -- Any departure, subjective or objective, form of state of physiological or psychological well-being.
9. **Mortality** -- The number of deaths in a given time or place.
10. **Relative Risk** -- The ratio of the risk of disease or death among the exposed to the risk among the unexposed.

**Chapter 38 - Achieving Asthma Control**

1. **Alternative Asthma Medications** -- The cornerstone of asthma therapy are ICS’s. ICS dose can be increased or LABA’s can be added for more severe asthma. In certain situations, LTRA’s are also recommended. There are older medications which do not work as well and/or have side effects, and these include chromolyn and nedocromil (which stabilize mast cells) as well as theophylline (a bronchodilator). Omalizumab is a new, genetically engineered weekly injection reserved for only for the very severe patient.
2. **Asthma** -- Asthma is defined as an obstructive airway disease characterized by episodic airway narrowing that resolves either spontaneously or with treatment, caused by hyperresponsiveness to otherwise normal stimuli and chronic inflammation.
3. **Asthma Control** -- A more recent concept and focus of care that recognizes some of the limitations of severity classification. The parameters of impairment are similar to asthma severity, except now patients are divided into well controlled, not well controlled, and very poorly controlled. The concept is that with appropriate therapy, most patients regardless of classification should be able to achieve control. Though treatment is initiated based on severity level, treatment is adjusted based on control. Providers can initiate higher doses and/or second medications if asthma is not controlled (step up), or decrease treatment if asthma is well controlled (step down).
4. **Asthma Severity** -- Asthma severity has been used to classify patients with asthma since the early 1990’s. Classification is based on daytime and night time symptoms, pulmonary function as measured by spirometry, use of rescue medication, and most recently by impact of asthma on one’s daily activities such as interference with work, school, and play. There are two components of severity, impairment (symptoms, lung function) and risk, specifically for exacerbations. Treatment for asthma should be initiated based on the current severity classification. A limitation of this classification system is that asthma is a variable disease, and even patients with mild persistent asthma can have poor asthma control.
5. **Community Based Asthma Programs** -- Though individual patient and provider interactions can be improved, it is clear that in order to reduce the asthma burden, community programs are needed. Both school and pharmacy based programs have shown some initial success. Mobile asthma vans have also shown promise, especially in minority and impoverished areas.
6. **Controller and reliever therapy** -- Patients with persistent asthma (asthma which is symptomatic more than twice a week) require two kinds of medications: controller and reliever. Controller medications, preferably inhaled corticosteroids, are taken everyday, regardless of symptoms, to prevent exacerbations. Reliever medications are usually short acting bronchodilators (albuterol) to be used only when patients are symptomatic. This concept is important to patient education, since it is not uncommon for patients to discontinue their preventative, controller medications when they are feeling well. In addition, overuse of reliever medications is a sign of poorly controlled asthma.
7. **Environmental Triggers** -- Various factors in the environment can contribute to asthma. Several outdoor triggers include ozone, sulfur dioxide, nitrogen dioxide, acid aerosols. Extremes in temperature or humidity may also play a role. Indoor pollutants such as tobacco smoke also worsen asthma.
8. **Healthcare Disparities** -- A difference in health outcomes between majority and minority populations that can not be accounted for by socioeconomic factors alone. Asthma prevalence and morbidity is particular worse in African Americans. Though some genetic factors have been hypothesized, and socioeconomic factors do play a role; other factors such as discrimination and culture insensitivity may an important part in asthma disparities.
9. **Inhaled Corticosteroids (ICS)** -- Inhaled steroids are the cornerstone of asthma therapy and recommend for all patients with persistent asthma. Inhaled steroids have been shown to be superior than all other agents in the ability to improve lung function, reduce symptoms, prevent exacerbations, and even reduce death. One study showed that ICS given earlier in the course of the disease seemed to halt progression, but other studies have been unable to replicate this.
10. **Leukotriene Modifiers** -- (also know as Leukotriene Receptor Antagonists or LTRA). The leukotriene modifiers work by blocking leukotrienes, which is an important mediator in the inflammatory cascasde. They have a low risk of side effects and can be taken in pill form. However, they are not as effective as ICS.
11. **Long Acting Bronchodilators (LABA)** -- In contrast to short acting bronchodilators (SABA) that are used for rescue/reliever medication, LABA’s are used for asthma control. When combined with and ICS, they have been shown to improve both objective lung function measurements as well as patient factors such as improvement in peak flow readings and days without asthma symptoms. They should not be used without ICS’s, as one study showed an increase in rates of asthma death when used alone.
12. **Peak flow meters** -- Hand held devices which asthmatic patients use to track their daily airflow. Since patients’ airflow is generally reduced prior to an asthma attack, and patients may not perceive this decrease, home measurement of peak flow may prevent serious exacerbations by allowing treatment before an asthma attack happens. Providers will help patients determine their “personal best” peak flow number, and have them track this over time. Peak flow reading can be used in conjunction with asthma action plans. They are recommended for patients with moderate to severe asthma.
13. **Spirometry** -- Pulmonary function testing (PFT’s) are done using a machine called a spirometer. Spirometers can measure how much air one can breathe in or out (lung volumes) as well as how fast one is able to breathe air out (lung flow). Asthma is an obstructive disease, with relatively normal volumes, but reduced flow. In order to properly diagnose asthma and rule out any underlying condition, it is recommended that patients have spirometric assessment. Spirometry is also recommended to objectively measure response to treatment, as well as recommended every 1-2 years to assess progressive decline in lung function. Spirometry can be performed in the primary care setting, but many primary care physicians do not have these devices. Specialists such as allergist and pulmonologists used spirometry as standard of care practice.
14. **Written action plans** -- Written action plans are directions from a health care provider to asthmatic patient or caregiver that instruct exactly what to do when patients are feeling well, when they are symptomatic, and when they having asthma attacks. The instructions usually involve which medications to take and when, and can be also used in conjunction with peak flow meters (see below). Other instructions can include trigger avoidance and contact information. Despite lack of evidence that written action plans improve asthma outcomes, they severe as an excellent communication tool between patients and providers, and continue to be recommended by national guidelines.