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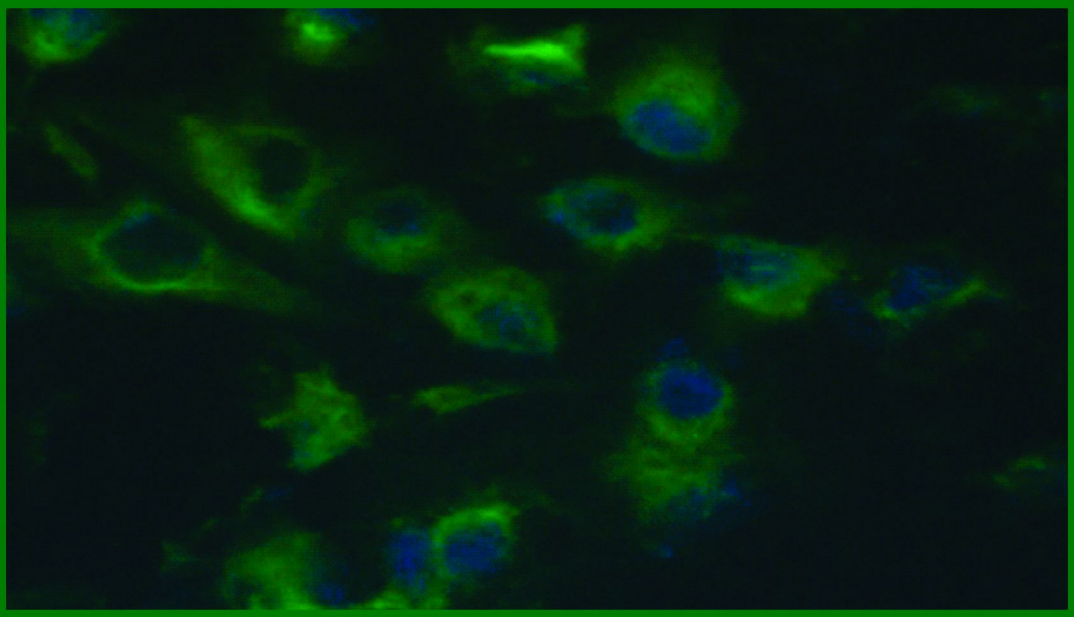
BASICS OF HEALTH DIAGNOISTIC VOL · I



JV'n Dr. Ritu Singh Rajput

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Basics of Health Diagnostic VOL 1

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Chapter 1: Introduction of Health diagnostic and its applications

In studying the human body, it is important to consider natural body systems and functions and how the body is changed by physical or physiological changes. The branch of science that studies structural and functional changes in tissues and organs that lead to disease is pathophysiology. Disease induces changes to normal anatomy and physiology that are either visible or concealed. Pathophysiology also involves anatomy, which is the study of disease-associated modifications to cells and tissues.

Disease is a concept that defines some departure from the usual state of wellbeing or fitness. Physical, emotional, and social disorders are included. The illness triggers homeostasis-sis to disrupt the body. Generally, small modifications are reversed by the body, which gradually returns to its natural state. With measures such as pulse or blood pressure, a 'nor-mal' body status is measured, using particular statistics that reflect an average range signifying normal health. Age, ethnicity, family background, climate, and activity levels all affect the natural state of a person.

Diagnosis of Disease

The several currently available ways of diagnosing illness are categorized as either invasive or non-invasive treatments. Both diagnostic tools available must be familiar with the PTA. Invasive treatments involve a "invasion" of the patient's body, including the use of needles, catheters, and other orders. There is either no risk or just slight risk to the patient for noninvasive procedures. Radiographic tests and urine monitoring require noninvasive procedures.

Invasive operations in general have a greater chance of possible injury than non-invasive procedures. Before continuing, the health care provider must assess the possible benefits against the potential costs of any treatment. Patients need to be completely aware of these advantages and risks.

Biotechnology Applications in Medicine

You may have already known about the technology of Recombinant DNA (rDNA). In healthcare, this biotechnology application is very important because it facilitates the mass manufacturing of safe and more efficient drugs. It also eliminates undesirable immune reactions from non-human origins that are popular in medicinal goods.

Approximately 30 recombinant therapeutics have currently been licensed worldwide for human use and 12 of these are currently being sold in India. Let's have a look at some of the apps.

1) Genetically Engineered Insulin

Using insulin from the pancreas of slaughtered pigs and cattle, diabetes was treated earlier. Do you think any side effects in humans are caused by this insulin? Yes! Yes! Allergies and other unwanted immune responses in humans are caused by insulin from animal products. This is why there was a need for human insulin to be isolated. Is there a way for this to be done? What if we were able to use bacteria for human insulin production? Not only can we cultivate bacteria in massive numbers, but human insulin can also be mass-produced!

Two small, polypeptide chains, chain A and B, connected by disulfide bridges, consist of insulin. Insulin is developed in mammals as a 'prohormone' (including humans). An extra peptide, the C peptide, has this prohormone, which needs to be removed to give rise to mature insulin.

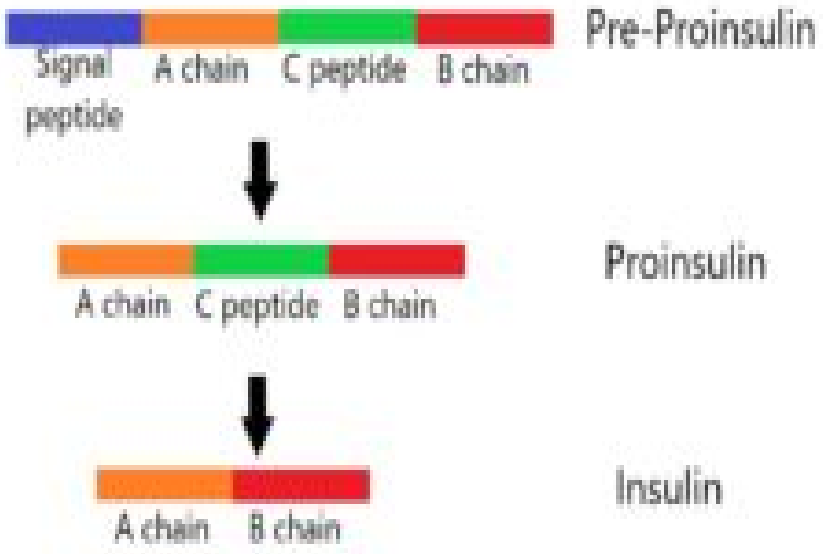

Figure: Maturation of Insulin.

Assembling insulin into its mature form is the main obstacle when producing human insulin. This barrier was solved by an American firm named 'Eli Lilly' in 1983. They prepared two sequences of DNA that correspond to the human insulin chains A and B. They then integrated these sequences into E's plasmids. To produce insulin chains, Coli. In addition, by forming

disulfide bonds to give rise to human insulin, they separately created the chains, removed and fused them.

2) Gene Therapy

Gene therapy is a biotechnology application that requires a series of therapies in an infant or an embryo that can correct a gene defect. To substitute for the non-functional gene, it involves injecting a regular gene into the cells or tissues of the human. How this functions.

The first human gene therapy was used in 1990 to treat a 4-year-old child with a mutation of the adenosine deaminase enzyme (ADA). The loss of the gene for ADA, which is an enzyme essential for the operation of the immune system, is responsible for this condition. In some circumstances, bone marrow transplantation helps treat this condition. In certain cases, enzyme replacement therapy, which includes injecting functional ADA into the patient, is also successful. All these methods, though, are not entirely curative.

In gene therapy, the patient's blood lymphocytes are grown in a culture outside of the body. A functional ADA cDNA is eventually inserted into these lymphocytes and reintroduced into the patient. This soothes the effects of the disease. However, repeated infusions of these genetically modified lymphocytes are expected by the patient, since these cells are not immortal. The insertion of the gene generating ADA from marrow cells into cells at the early embryonic stages of life may be a permanent solution for this.

3) Molecular Diagnosis

We also know that early diagnosis of a disease is necessary for the disease to be handled successfully. Using traditional approaches including serum and urine analysis, early detection is not feasible. Let's look at some applications of biotechnology that aid in the early detection of diseases.

i)Polymerase Chain Reaction (PCR)

Normally, only after the signs of the illness begin to manifest will we detect a pathogen (bacteria, virus, etc.). However, the pathogen prevalence in the body is very high at this point. Using a so called PCR technique. PCR requires amplification of the pathogen's nucleic acid, helping one to recognize the pathogen at a relatively low concentration. Currently, we regularly use PCR to detect HIV in suspected patients with AIDS and to detect gene mutations in suspected patients with cancer.

ii) Enzyme-Linked Immunosorbent Assay (ELISA)

Antigen-antibody reactions are the fundamental concept of ELISA. By detecting the presence of antigens (pathogen proteins) in the patient's serum, or by detecting antibodies produced against the pathogen, ELISA may diagnose infections.

iii) In Situ Hybridization

This approach involves labeling with a radioactive molecule a single-stranded DNA or RNA (probe). This then hybridizes inside a clone of cells with its complementary DNA. The clone with the mutated gene would not appear on the photographic film upon identification using autoradiography because the probe is not complementary to the mutated gene.

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Competitive Questions:

1. What is the name of the disease in man arising out of Vitamin B1 deficiency?

A. Scurvy

B. Beriberi

C. Pellagra

D. Gingivitis

2. What does niacin deficiency cause?

A. Acne

B. Scurvy

C. Boils

D. Pellagra

Chapter 2 Bacterial Diseases

Introduction

Every form of illness caused by bacteria includes bacterial diseases. Bacteria, which are microscopic types of life that can only be seen through a microscope, are a type of microorganism. Viruses, some fungi, and some parasites include other types of microorganisms.

There are millions of bacteria that usually reside in the skin, intestines, and vagina. No disease is caused by the vast majority of bacteria, and many bacteria are often beneficial and even required for good health. Often, these bacteria are referred to as' nice bacteria' or' healthy bacteria.'

Pathogenic bacteria are considered dangerous bacteria that cause bacterial infections and illness. When pathogenic bacteria invade the body and tend to replicate and crowd out healthy bacteria, or to develop in tissues that are typically sterile, bacterial diseases occur. Toxins that affect the body can also be released by toxic bacteria.

- ***Escherichia coli and Salmonella*** cause food poisoning.
- ***Helicobacter pylori*** cause gastritis and ulcers.
- ***Neisseria gonorrhoeae*** causes the sexually transmitted disease gonorrhea.
- ***Neisseria meningitidis*** causes meningitis.
- ***Staphylococcus aureus*** causes a variety of infections in the body, including boils, cellulitis, abscesses, wound infections, toxic shock syndrome, pneumonia, and food poisoning.
- ***Streptococcal bacteria*** cause a variety of infections in the body, including pneumonia, meningitis, ear infections, and strep throat.

Bacterial infections, such as blood poisoning (bacteremia), renal failure and traumatic shock syndrome, are infectious and can result in multiple severe or life-threatening complications. When you believe that you have a bacterial illness, seek immediate medical attention. If you, or anyone you are with, have vital signs of a bacterial illness such as high fever, lethargy or unresponsiveness, seek urgent medical attention.

Symptoms

Bacterial disease signs differ based on the type of bacterial infection, the location of the infected body, and other conditions, such as the age and health history of the patient. Bacterial disease signs can also be close to those of other illnesses, such as colitis, pneumonia, and viral infections. A fever is the classic symptom of a bacterial infection, but not every person with a bacterial infection would have a fever.

Bacterial disease symptoms can include:

- Bloody urine and painful, frequent urination
- Diarrhea
- Flu-like symptoms (fatigue, fever, sore throat, headache, cough, aches and pains)
- Irritability
- Nausea and vomiting
- Pain such as joint, ear or abdominal pain
- Rashes, lesions and abscesses
- Stiff neck
- Weakness

In infants, signs of a bacterial disease can also include:

- Bulging of the soft spot on the top of the head
- Difficulty with feeding
- Excessive crying or fussiness
- Excessive sleepiness

Serious symptoms that might indicate a life-threatening condition

- Confusion or delirium
- Deep, wet chest cough that produces yellow, green or brownish phlegm
- Difficulty breathing, wheezing or shortness of breath
- High fever (higher than 101 degrees)

- Inappropriate change in alertness or level of consciousness
- Infants: sunken fontanel (soft spot) on the top of the head, lethargy, no tears with crying, and few or no wet diapers
- Lethargy or unresponsiveness
- Not urinating or urinating small amounts of tea-colored urine
- Seizure

Causes bacterial diseases

Harmful bacteria cause infectious diseases (pathogenic bacteria). No disease is caused by the vast majority of bacteria, and many bacteria are often beneficial and even required for good health. Bacterial infections arise when pathogenic bacteria invade a part of the body that is usually sterile, such as the bladder, or when areas such as the intestines, vagina, or mouth crowd out the beneficial bacteria. Bacterial infections can occur less frequently when healthy bacteria multiply uncontrollably.

Various ways pathogenic bacteria can enter the body

Pathogenic bacteria can enter the body through a variety of means including:

- Contamination of bites, cuts, rashes, abrasions and other breaks in the skin, gums and tissues
- Eating contaminated food
- Getting bitten by an infected insect
- Having sexual contact with an infected person
- Inhaling contaminated air-borne droplets into the nose and lungs
- Kissing an infected person
- Sharing needles for tattooing or drug use
- Through the eyes, ears or urethra
- Touching infected feces or body fluids, and not washing your hands before eating or touching your mouth, eyes or nose

When bacteria enter the body, the bacteria would be recognized as foreign invaders by a balanced immune system and seek to destroy or avoid the bacteria from reproducing. However, the body is not always able to avoid the bacteria from multiplying and spreading, even in a healthy person. They will crowd out healthy bacteria and microorganisms and release toxins that kill the body's cells as the harmful bacteria replicate.

Risk factors for bacterial diseases

In any age group or population, bacterial diseases may occur, but a variety of factors raise the likelihood of the occurrence of bacterial diseases. Not all persons with risk factors are going to get bacterial diseases. Bacterial disease risk factors include:

- Being an infant, child or older adult
- Eating eggs or meats that are raw or undercooked
- Eating expired foods, or eating leftovers that have been stored for more than two to three days
- Having a genetic predisposition to bacterial infection
- Having a compromised immune system due to an immunodeficiency disorder, HIV/AIDS, diabetes, cancer or cancer treatment, kidney disease, or from taking steroid medications
- Having a chronic disease
- Malnutrition
- Not washing your hands frequently, especially after using the bathroom, touching pet feces, handling reptiles, or touching raw foods or foods contaminated with bacteria
- Significant exposure to a person with a bacterial disease

Reducing your risk of bacterial diseases

You can lower your risk of developing or transmitting bacterial diseases by **:**

- Avoiding contact with a person who has a bacterial disease or its symptoms, such as fever, vomiting or diarrhea
- Covering your mouth and nose with a tissue when sneezing or coughing, then washing your hands

- Defrosting foods in the refrigerator or microwave, not on the counter
- Refrigerating leftovers right away and eating them within two to three days unless they have been frozen
- Eating a healthy diet that is high in whole grains, fruits and vegetables and contains adequate low-fat protein and low-fat dairy products or other calcium sources
- Getting enough rest and minimizing stress
- Getting recommended vaccines for bacterial diseases, such as meningitis, pneumonia, tetanus and rabies
- Seeking regular medical care and following your treatment plan for a chronic disease
- Throwing out expired food or perishable food that has been sitting at room temperature for two hours or longer
- Using antibacterial products to clean surfaces, such as computer keyboards, telephones and sinks
- Washing your hands after using the bathroom and after contact with pet feces, reptiles, dirty diapers, raw foods, and people who are ill
- Washing plates, utensils, and cutting boards that have been exposed to raw meats or poultry in hot soapy water
- Wearing long pants and sleeves, and using insect repellant when in tall grass or wooded areas

Treatments

With antibiotics, bacterial infections are treated. By destroying the unhealthy bacteria or preventing them from reproducing and spreading, antibiotics work. For the treatment of particular species of bacteria, different types of antibiotics are effective. Depending on the nature and severity of the bacterial disease and other conditions, antibiotics can be administered orally, intravenously, or by intramuscular injection.

General types of antibiotics include:

- Aminoglycosides
- Cephalosporins
- Macrolides
- Penicillins
- Quinolones
- Tetracyclines

Treatment of bacterial infections also includes:

- Good nutrition
- Hospitalization and intensive care in some cases, especially if complications occur
- Increased fluids
- Rest

It may also be important to treat and track individuals who have had direct contact with a person with a severe bacterial illness, such as bacterial meningitis, for the disease, even in the absence of symptoms.

An antibiotic that used to be useful in curing a bacterial disease often ceases being effective. This is considered susceptibility to antibiotics. Which makes it harder to treat an infectious illness which can lead to extreme complications, such as sepsis, paralysis and death?

Complications of bacterial diseases

Bacterial diseases can lead to serious, even life-threatening complications in some individuals. Therefore, when you develop signs of a bacterial infection, it is necessary to contact your health care provider. Once the underlying infection has been determined, it will help mitigate any further risks, including: adopting the recovery protocol outlined by the doctor.

- Coma

- Kidney failure
- Septicemia, which is a life-threatening blood infection that can lead to a body-wide response called sepsis
- Severe dehydration and electrolyte imbalance
- Shock
- Toxic shock syndrome

Along with typhoid and paratyphoid fever, diarrheal infections are among the leading causes of death among children worldwide. Although the implementation of clean water and sanitation systems in the United States and other developing countries has significantly reduced the risk of many enteric diseases, most of the world still suffers from a heavy disease burden. In addition, such viruses begin to spread and, closer to home, cause outbreaks. Research on enteric diseases in the Department of Epidemiology of Microbial Diseases includes:

- Describing the transmission dynamics and impact of vaccination against rotavirus in the United States and developing countries
- Developing mathematical and statistical models to estimate the burden of typhoid fever and to predict the impact and cost-effectiveness of typhoid conjugate vaccine strategies across low- and middle-income countries
- Understanding the predominant routes of transmission and control options for cholera epidemics, past and present

References:

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DeLong, D., 2012. Bacterial diseases. In *The Laboratory Rabbit, Guinea Pig, Hamster, and Other Rodents* (pp. 301-363). Academic Press.

Competitive Questions:

1. Using purgatives on a regular basis is harmful to health. Which deficiency does it cause?

A. Iron

B. Potassium

C. Iodine

D. Chromium

2. what is the condition known as, in which the body does not get its fair share of nutrients, either from starvation, or as a result of poor absorption:

A. Marasmus

B. Malnutrition

C. Kwashiorkor

D. Malnutrition & Marasmus

Chapter 3 Infectious diseases

Infectious diseases

Infectious diseases, including bacteria, viruses, fungi or parasites, are illnesses caused by animals. Several animals exist in and on our bodies. Normally, they're harmless or sometimes beneficial. But certain species can cause disease under some conditions.

It is possible to transmit certain infectious diseases from person to person. Any of them are passed on by insects or other species. And by eating polluted food or water or being exposed to organisms in the atmosphere, you can get others.

Depending on the individual causing the infection, signs and symptoms differ, but also include fever and exhaustion. Mild infections may lead to rest and home remedies, while hospitalization may be required for certain life-threatening infections. Vaccines can eliminate certain contagious diseases, such as measles and chickenpox.

Symptoms

Each infectious disease has its own specific signs and symptoms. General signs and symptoms common to a number of infectious diseases include:

- Fever
- Diarrhea
- Fatigue
- Muscle aches
- Coughing

Causes

Infectious diseases can be caused by:

Bacteria: For diseases such as strep throat, urinary tract infections and tuberculosis, these one-cell organisms are responsible.

Viruses: Viruses, much smaller than viruses, cause a variety of ailments ranging from the common cold to AIDS.

Fungi: Some skin diseases are caused by fungi, such as ringworm and athlete's foot. You can infect the lungs or nervous system with other kinds of fungi.

Parasites:A microscopic parasite that is spread by a mosquito bite causes malaria. There may be other parasites spread from animal manure to humans.

Direct contact

An easy way to catch most infectious diseases is by coming in contact with a person or an animal with the infection. Infectious diseases can be spread through direct contact such as:

- **Person to person.** Via the direct transmission of bacteria, viruses or other germs from one individual to another, infectious diseases are widely transmitted. This will happen when someone who is not sick is touched, kissed, or coughed or sneezed by a person with the bacteria or virus.

  The sharing of body fluids from sexual intercourse will also disperse these germs. There may be no signs of the disease in the man who passes the germ, but he may just be a vector.

- **Animal to person.** Getting bitten or scratched by an infectious animal, including a cat, can make you ill and can be dangerous in serious cases. It can be risky to treat animal waste, too. For eg, by scooping your cats litter box, you can get a toxoplasmosis infection.
- **Mother to unborn child.** A pregnant woman can pass on germs to her unborn baby that causes infectious diseases. Via the placenta or through breast milk, certain germs may get through. During birth, germs in the vagina may also be spread to the infant.

Indirect contact

Indirect touch may also be passed on to disease-causing species. On an inanimate object, like a tabletop, doorknob or faucet handle, several germs may remain. For eg, if you pass a door handle treated by someone afflicted with a flu or a cold, you can pick up the germs that he or she left behind. You can get sick if you then cross your eyes, mouth or nose before washing your hands.

Insect bites

To travel from host to host, certain germs rely on insect carriers, such as mosquitoes, fleas, lice or ticks. As vectors, these carriers are identified. The malaria parasite or West Nile virus may be borne by mosquitoes. The bacterium which causes Lyme disease may be carried by deer ticks.

Food contamination

With infected food and drink, disease-causing germs will also kill you. This propagation mechanism causes germs to be transmitted via a single source to several individuals. For starters, Escherichia coli (E. coli) is a bacterium found in or on some products, such as undercooked hamburgers or unpasteurized fruit juices.

References:

Centers for Disease Control and Prevention (CDC, 1999. Control of infectious diseases. *MMWR. Morbidity and mortality weekly report*, *48*(29), p.621.

Competitive Questions:

1. A metastatic cancerous tissue is termed ‘sarcoma’ if the disorder is in

A Fibroblasts
B Epithelial cells
C Immune system
D Circulatory system

Chapter 4 Diagnosis of infectious diseases

The primary cause of death and morbidity in young children is respiratory disorders and, especially, pneumonia. In primary care, the diagnosis of both pneumonia and asthma is primarily based on clinical symptoms, history, and bronchodilator reactivity. This research sought to explain clinical procedures related to the differential diagnosis of respiratory disease in young children, in particular between pneumonia and asthma, in diverse global primary care settings.

Asthma is a disease that narrows and swells the airways and can create excess mucus. This can find it hard to breathe and cause coughing, a whistling sound (wheezing) and shortness of breath as you breathe out.

Symptoms

Symptoms of asthma differ from individual to individual. You can have infrequent asthma attacks, have symptoms only at some occasions, or have symptoms all the time, such as while exercising.

Asthma signs and symptoms include:

- Shortness of breath
- Chest tightness or pain
- Wheezing when exhaling, which is a common sign of asthma in children
- Trouble sleeping caused by shortness of breath, coughing or wheezing
- Coughing or wheezing attacks that are worsened by a respiratory virus, such as a cold or the flu

Signs that your asthma is probably worsening include:

- Asthma signs and symptoms that are more frequent and bothersome
- Increasing difficulty breathing, as measured with a device used to check how well your lungs are working (peak flow meter)
- The need to use a quick-relief inhaler more often

For some people, asthma signs and symptoms flare up in certain situations:

- **Exercise-induced asthma,** which may be worse when the air is cold and dry

- **Occupational asthma,** triggered by workplace irritants such as chemical fumes, gases or dust
- **Allergy-induced asthma,** triggered by airborne substances, such as pollen, mold spores, cockroach waste, or particles of skin and dried saliva shed by pets (pet dander)

Seek emergency treatment

Severe asthma attacks can be life-threatening. Work with your doctor to determine what to do when your signs and symptoms worsen — and when you need emergency treatment. Signs of an asthma emergency include:

- Rapid worsening of shortness of breath or wheezing
- No improvement even after using a quick-relief inhaler
- Shortness of breath when you are doing minimal physical activity

Pneumonia

Pneumonia in one or both lungs is an illness. It is caused by bacteria, viruses, and fungi.

The bacteria allow the air sacs in your lungs, which are called alveoli, to become inflamed. Alveoli are filled with fluid or pus, making it impossible to breathe.

Symptoms of pneumonia

Pneumonia symptoms can be mild to life-threatening. They can include:

- coughing that may produce phlegm (mucus)
- fever
- sweating or chills
- shortness of breath that happens while doing normal activities or even while resting
- chest pain that’s worse when you breathe or cough
- feelings of tiredness or fatigue
- loss of appetite
- nausea or vomiting
- headaches

Other symptoms can vary according to your age and general health:

- Children under 5 years old may have fast breathing or wheezing.
- Infants may appear to have no symptoms, but sometimes they may vomit, lack energy, or have trouble drinking or eating.
- Older people may have milder symptoms. They can also exhibit confusion or a lower than normal body temperature.

Bacterial pneumonia

The most common cause of bacterial pneumonia is *Streptococcus pneumoniae*. Other causes include:

- Mycoplasma pneumoniae
- *Haemophilus influenzae*
- Legionella pneumophila

Viral pneumonia

Respiratory viruses are often the cause of pneumonia. Some examples include:

- influenza (flu)
- respiratory syncytial virus (RSV)
- rhinoviruses (common cold)

Viral pneumonia is usually milder and can improve in one to three weeks without treatment.

Fungal pneumonia

Fungi from soil or bird droppings can cause pneumonia. They most often cause pneumonia in people with weakened immune systems. Examples of fungi that can cause pneumonia include:

- *Pneumocystis jirovecii*
- *Cryptococcus* species
- Histoplasmosis species

Types of pneumonia

Pneumonia can also be classified according to where or how it was acquired.

Hospital-acquired pneumonia (HAP)

During a hospital stay, this type of bacterial pneumonia is acquired. It may be more extreme than other kinds, since antibiotics can be more resistant to the bacteria involved.

Community-acquired pneumonia (CAP) refers to pneumonia that's acquired outside of a medical or institutional setting.

Aspiration pneumonia

Aspiration pneumonia occurs when you inhale bacteria from food, water, or saliva into your lungs. If you have a swallowing problem or if you are too sedated by the use of medications, alcohol, or other substances, this sort is more likely to occur.

Prescription medications

Many forms of bacterial pneumonia should be treated with oral antibiotics. Still take the whole antibiotic course, particularly though you are beginning to feel better. Not doing so will keep the infection from drying up, and in the future, it could be more difficult to handle.

Drugs with antibiotics should not operate on viruses. In certain cases, an antiviral could be recommended by the doctor. However, with at-home treatment, several cases of viral pneumonia obvious on their own.

In order to combat fungal pneumonia, antifungal drugs are used. To get rid of the infection, you will need to take this drug for several weeks.

References:

Fauci, A.S., 2001. Infectious diseases: considerations for the 21st century. *Clinical Infectious Diseases*, *32*(5), pp.675-685.

Competitive Questions:

1. A health disorder that results from the deficiency of thyroxine in a adults and characterized by (i) a low metabolic rate, (ii) increase in body weight and (iii) tendency to retain water in tissues is

A Cretinism

B Hypothyroidism

C Simple goitre

D Myxoedema

2. Common cold is not cured by antibiotics because it is

A Caused by a Gram-negative bacterium

B Not an infectious disease

C Caused by a virus

D Caused by a Gram-positive bacterium

Chapter 5: Antigen-Antibody Reactions

Introduction to Antigen-Antibody Reactions

Antigen-antibody reactions are known as the interactions between antigens and antibodies. The reactions are highly selective and an antigen only interacts with antibodies formed by it or with antigens closely linked to it. Antibodies on antigens identify molecular forms (epitopes). In general, the higher the epitope's match (in terms of geometry and chemical character) to the combining location of the antibody, the more desirable the associations that would be produced between the antibody and antigen, and the greater the antigen affinity of the antibody. One of the most significant considerations in evaluating antibody effectiveness in vivo is the affinity of the antibody to the antigen.

The relationship between antigen and antibody is a permanent bimolecular connection between antigen and antibody. Various non-covalent associations between the epitope (antigenic determinant) and variable area (VH/VL) domain of the antibody are used in the relationship between antigen and antibody.

Chemical Bonds Responsible for the Antigen–Antibody Reaction

Exclusively non-covalent bonds are involved in the association between the Ab-binding site and the epitope in a similar fashion to that in which proteins bind to their cellular receptors or enzymes bind to their substrates. The binding is reversible and high ionic strength or intense pH will inhibit or dissociate it. In Ag-Ab binding, the following intermolecular forces are involved:

1. **Electrostatic bonds:** This stems from the attraction of two protein side chains of oppositely charged ionic groups, such as an ionized amino group (NH_4^+) on a lysine in the Ab and an ionized carboxyl group (COO^-) on an aspartate residue in the Ag.
2. **Hydrogen bonding**: Relatively weak hydrogen bonds between hydrophilic groups (e.g., OH and C=O, NH and C=O, and NH and OH groups) can be formed when Ag and Ab are in very close proximity.
3. **Hydrophobic interactions**: Due to Van der Waals bonding and coalescing in an aqueous environment, hydrophobic groups such as the side chains of valine, leucine, and phenylalanine appear to interact, removing water molecules from their surroundings. As a result, the distance between them reduces, strengthening the

attraction energies involved. It is estimated that this form of interaction leads to up to 50 percent of the overall strength of the Ag-Ab bond.

4. **Van der Waals bonds:** Such forces depend on the interactions between the "electron clouds" surrounding the molecules of Ag and Ab. The relationship of alternating dipoles in two molecules has been compared to one which might occur, alternating in such a way that oppositely directed dipoles would be present in closely applied areas of the Ag and Ab molecules at any given moment.

Any of these non-covalent interactions works over a very short distance (usually around 1 Å), so Ag-Ab interactions are based on a very close antigen-antibody match.

Strength of Ag-Ab interactions

1. **Affinity**

- Combined strength of totalnon-covalent interactions between single Ag- binding site of Ab and single epitope is affinity of Ab for that epitope.
- Low affinity Ab: Bind Ag weakly and dissociates readily.
- High affinity Ab: Bind Ag tightly and remain bound longer.

2. **Avidity**

- Strength of multiple interactions between multivalent Ab and Ag is avidity. Avidity is better measure of binding capacity of antibody than affinity. High avidity can compensate low affinity.

3. **Cross reactivity**

- Antibody elicited by one Ag can cross react with unrelated Ag if they share identical epitopeor have similar chemical properties.

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Competitive Questions:

1. The earliest recorded pandemic – the Black Death was speculated to be _____

a. Plague
b. Rabie
c. Leprosy
d. None of the above

2.VDRL test is an example of
A.Tube test

B.Ring test

C.Slide test

D.none of these

Chapter 6: Genetic disorders

Genetic disorders

There are a number of different types of genetic disorders (inherited) and include:

1. Single gene inheritance
2. Multifactorial inheritance
3. Chromosome abnormalities
4. Mitochondrial inheritance

Symptoms and Signs of Down syndrome

The Down syndrome kid has a trademark look. However, since the phenotype, the way the chromosomes make the infant look, will be markedly different for each patient, each part of the appearance does not need to be present. Popular signs of Down syndrome are:

1. a small head and short neck,
2. flat face, and
3. upward slanting eyes, ears are flat and positioned lower than "normal,"
4. the tongue protrudes and seems to be too large for the mouth,
5. hands tend to be wide,
6. short fingers and there is just a single flexion crease in the palm, and
7. Joints tend to be more flexible and muscles may lack tone.

Single gene inheritance disorders

Mendelian or monogenetic inheritance is also considered single gene inheritance. This form of inheritance is caused by modifications or mutations that arise in a single gene's DNA sequence. Thousands of known single-gene abnormalities are present. Such diseases are considered monogenetic disorders (disorders of a single gene).

Single-gene disorders have different patterns of genetic inheritance, including

- Autosomal dominant inheritance, in which only one copy of a defective gene (from either parent) is necessary to cause the condition;

- Autosomal recessive inheritance, in which two copies of a defective gene (one from each parent) are necessary to cause the condition; and
- X-linked inheritance, in which the defective gene is present on the female, or X-chromosome. X-linked inheritance may be dominant or recessive.

Some examples of single-gene disorders include

1. Cystic Fibrosis,
2. Alpha- And Beta-Thalassemias,
3. Sickle Cell Anemia (Sickle Cell Disease),
4. Marfan Syndrome,
5. Fragile X Syndrome,
6. Huntington's Disease, And
7. Hemochromatosis.

Common multifactorial genetic inheritance disorders

Complex or polygenic inheritance is sometimes called multifactorial inheritance. A combination of environmental factors and mutations in several genes causes multifactorial inheritance disorders. For example, on chromosomes 6, 11, 13, 14, 15, 17, and 22, multiple genes that affect breast cancer susceptibility have been discovered. Multifactorial conditions include several common chronic diseases.

Examples of multifactorial inheritance include

1. Heart Disease,
2. High Blood Pressure,
3. Alzheimer's Disease,
4. Arthritis,
5. Diabetes,
6. Cancer,
7. Obesity.

Chromosomal abnormalities

In the nucleus of each cell, chromosomes, distinct structures composed of DNA and protein, are found. Because chromosomes are the carriers of the genetic material, disease may result from defects in chromosome number or structure. Chromosomal anomalies normally arise because of a cell division problem.

Down syndrome (sometimes referred to as 'Down's syndrome') or trisomy 21, for instance, is a common genetic condition that happens when an individual has three copies of chromosome 21. Many other chromosomal anomalies are present, including:

1. Turner syndrome (45,X0),
2. Klinefelter syndrome (47, XXY), and
3. Cri du chat syndrome, or the "cry of the cat" syndrome (46, XX or XY, 5p-).

Diseases may also occur because of chromosomal translocation in which portions of two chromosomes are exchanged.

Mitochondrial genetic inheritance disorders

Mutations in the non-nuclear DNA of mitochondria are the cause of this form of genetic disease. Mitochondria are tiny circular or rod-like organelles located in the cytoplasm of plant and animal cells that are active in cellular respiration. 5 to 10 circular pieces of DNA may be found in each mitochondrion. Because, during fertilisation, egg cells, but not sperm cells, retain their mitochondria, mitochondrial DNA is often inherited from the female parent.

Examples of **mitochondrial disease** include

1. Leber's hereditary optic atrophy (LHON), an eye disease;
2. myoclonic **epilepsy** with ragged red fibers (MERRF); and
3. Mitochondrial **encephalopathy**, lactic acidosis, and **stroke-like episodes** (MELAS), a rare form of **dementia**.

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Competitive Questions:

1. Which of the following is a type of autosomal recessive genetic disorder?

(a) Haemophilia

(b) Skeletal dysplasia

(c) Sickle cell anaemia

(d) None of the above

2. Which of the following disorder is also called the Royal disease?

(a) Colour blindness

(b) Haemophilia

(c) Sickle cell anaemia

(d) Alzheimer's disease

Chapter 7 Neurological disorders & their clinical management

A modification in a DNA sequence is a mutation. DNA copying errors made during cell division, exposure to ionizing radiation, exposure to chemicals called mutagens, or contamination by viruses may result in mutations.

Neurological diseases face a huge challenge on health around the world. The new figures indicate that the neurological disorders included in the Global Burden of Disease (GBD) Study-and Alzheimer's other dementias, Parkinson's disease, multiple sclerosis, epilepsy, and headache disorders (migraine, headache of tension-type [TTH], and headache of medication-overuse [MOH])-represent 3% of the global disease burden. While this is an apparently small overall number, among the top 50 causes of disability-adjusted life years, dementia, arthritis, migraine, and stroke rank in the top 50 (DALYs)

Parkinson's disease

Parkinson's disease is an insidiously insidious chronic irreversible neurodegenerative condition characterised by the presence of predominantly movement signs (bradykinesia, rest tremor, rigidity, and postural disturbances). It is also associated with a number of non-motor symptoms that, along with late-onset motor symptoms (such as postural instability and crashes, freezing of gait, speech, and swallowing difficulties), are currently one of the most challenging problems facing the practising physician when coping with long-term patients.

Diagnosis

Although there are no conclusive biological or imaging markers, diagnosis is currently made using specific scientific guidelines, such as those established in the United Kingdom by the Brain Bank of the Parkinson's disease Society. These principles are used globally to have a high degree of specificity for a definitive diagnosis. In about 25 percent of patients, scientific pathological trials focused on brain bank material from Canada and the United Kingdom have found that physicians mistakenly identify the disorder. The most prominent factors for misdiagnosis in these trials is the occurrence of critical tremors, vascular Parkinson's disease and atypical Parkinson's disease.

While the diagnosis is made solely on a clinical basis, as previously stated, there are modern diagnostic methods that can be used to focus on the involvement of dopaminergic denervation at the striatal stage, thereby offering clinical diagnosis help. This include fl uorodopa positron emission tomography (FDOPA-PET) and dopamine transporter imaging

using single photon emission tomography with radionucleide tracers (DAT-SPECT). Both approaches are still used as research instruments and not for the normal diagnosis of PD.

The primary Lewy body PD is responsible for most cases of Parkinsonism. 'Parkinsonism-plus' syndromes (including progressive supranuclear palsy, multisystem atrophy, corticobasal degeneration) and secondary parkinsonism's (mainly drug-induced, flunarizine and cinarizine are also significant culprits, particularly in Latin American countries where these medications are often misused to avoid drug-induced, fl unarizine and cinarizine

Treatment

In the development of rational pharmacotherapeutic approaches to PD, the discovery of the dopaminergic defi cit was the main turning point leading to the launch of levodopa and later dopamine agonists. Both subsequently developed medications (dopa-decarboxylase inhibitors, monoamine oxidize inhibitors, catechol-omethyl transferase inhibitors) work indirectly by dopaminergic pathways, with the exception of anticholinergics and amantadine. More recently, functional surgery, developed several years ago as a palliative alternative to PD treatment, has become a major therapeutic choice.

In an effort to interfere at varying stages of the biochemical circuitry of the basal ganglia outside the dopamine agonist receptor, newer advances have been made in the fi eld of PD pharmacotherapy. Drugs working on the receptors of adenosine, glutamate, adrenergic and serotonin are currently being scrutinised as potentially helpful cials at various levels of the disorder.

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Competitive Questions:

1. Alzheimer disease in humans is associated with the deficiency of
A Gamma aminobutyric acid (GABA)
B Dopamine
C Glutamic acid
D Acetylcholine

2. Suspension of attenuated pathogen that stimulates antibody formation is
A Antitoxin
B Vaccine
C Antibiotic
D Serum

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