UNIT-I

Introduction and History of Diagnostics

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What is Diagnostics

Diagnostics is defined as the identification of diseases by the examination of symptoms & signs & by other investigations an opinion or conclusion so reached.

A diagnostic assessment is an intensive clinical and functional face to face evaluation of a recipients mental health, developmental disability, or substance abuse condition that results in the issuance of a diagnostic/assessment report with a recommendation regarding whether the recipient meets target population criteria, & includes an order for enhanced benefit services that provides the basis for the development of an initial person centered plan.

Diagnostic Criteria

The term diagnostic criteria designates the specific combination of signs, symptoms & test results that the clinician uses to attempt to determine the correct diagnosis.

They are standards, normally published by international committees, & they are designed to offer the best sensitivity & specificity possible, respect the presence of a condition, with the state-of-the-art technology.

Types of Diagnostic Testing

Clinical diagnostic:

A diagnosis made on the basis of medical signs & patient reported symptoms, rather than diagnostic tests.

Laboratory diagnosis:

A diagnosis based significantly on laboratory reports or test results rather than the physical examination of the patient.

for instance, a proper diagnosis of infectious diseases usually requires both an examination of signs & symptoms, as well as laboratory characteristics of the pathogen involved.

Radiology diagnosis:

A diagnosis based primarily on the results from medical imaging studies. magnetic resonating imaging(mri) are common radiological diagnosis.

Diseases

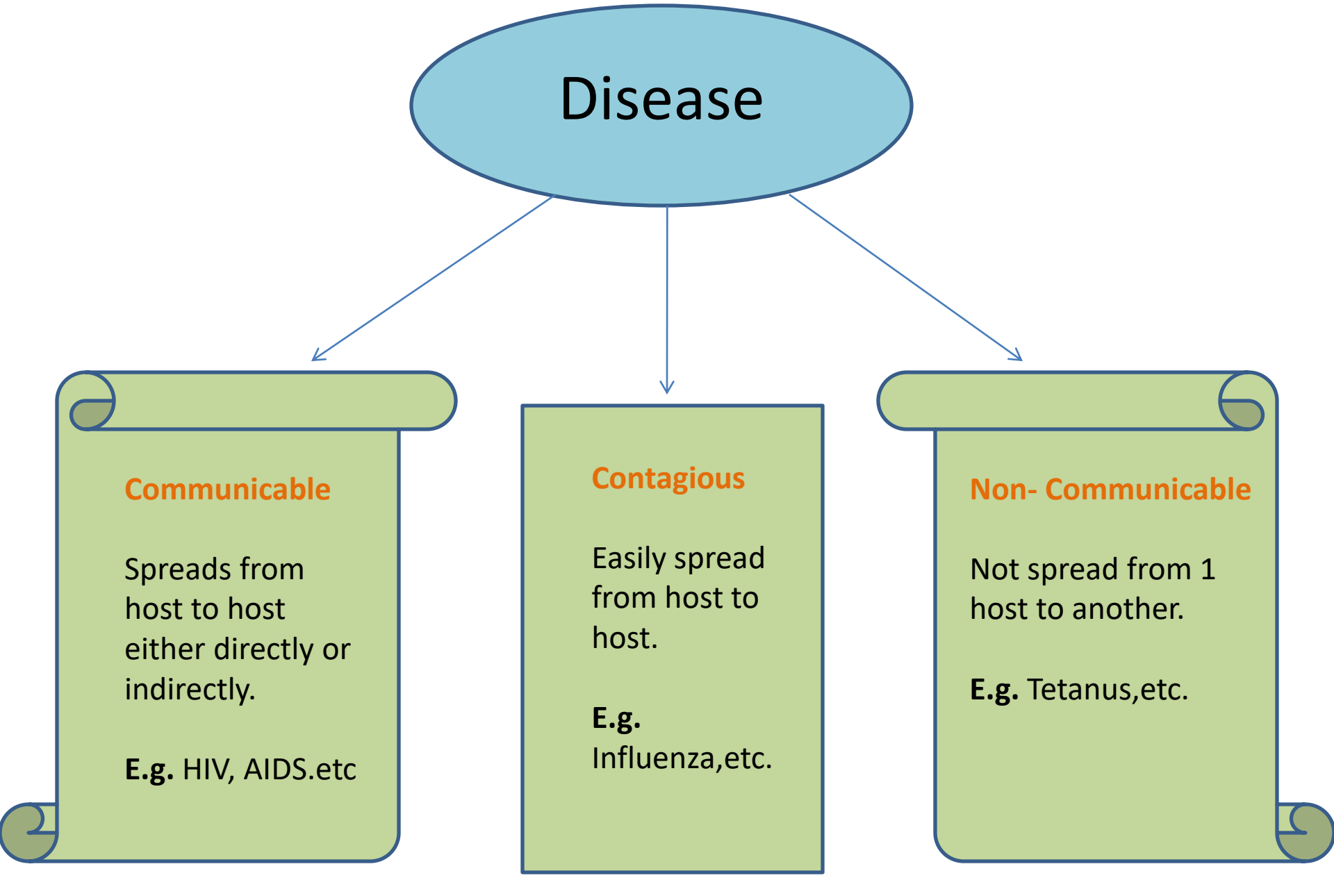
Disease is a state of change of health resulting from an Infection.

Disease is a condition of body or some parts of body or organ of body in which its function are disrupted.

I**nfection**: The invasion of bodily tissue by pathogenic microorganisms that proliferate, resulting in tissue injury that can progress to disease.

Factors influencing composition of microbiomes are:

- ✓Nutrients
- ✓Temperature
- ✓pH
- ✓Oxygen
- ✓Salinity
- ✓Sunlight
- ✓Host Defence
- ✓Individual Criteria's (Age, Sex, Geography, Health, Stress)


Disease
Communicable
Spreads from host to host either directly or indirectly.
E.g. HIV, AIDS.etc
Contagious
Easily spread from host to host.
E.g. Influenza,etc.
Non- Communicable
Not spread from 1 host to another.
E.g. Tetanus,etc.

INFECTIOUS DISEASES

•Infectious diseases kill more people worldwide than any other single cause. They are caused by germs.

•Person can get infected by touching, eating, drinking or breathing something that contains a germ.

•Vaccines, proper hand washing and medicines can help prevent infections.

•Germs are tiny living things that are found everywhere- in air, soil & water. Germs can also spread through animal and insect bites and sexual contact.
There are four main kinds of germs-

Bacteria
Viruses
Fungi
Protozoa

Types of Infection

Primary infection: Initial infection with an organism to host constitutes primary infection.

Secondary infection: When in a host whose resistance is lowered but pre-existing infection, a new organism may set up a new infection.

Local infection: Infection that is limited to a defined area or single organ with symptoms that resemble inflammation(redness, tenderness & swelling)

Systemic infection: Infection that spreads to whole body resulting in a septicemia.

Acute infection: It appears suddenly or lasts for a short time
e.g. URI

Chronic infection: May occur slowly over a long period & may last months to years.

Latrogenic infection: Infection resulting due to therapeutic and diagnostic procedures.

Factors affecting Microbial Pathogenicity

- ✓Host Susceptibility.
- ✓Host Resistance.
- ✓Presence of Bacterial Virulence Factors.
- ✓Presence of Host-mediated Pathogenesis.
- ✓Ability for Intracellular Growth.

Disease Occurrence

Incidence: No. of people that develop a disease in a given time period. i.e. rate of disease spread.

Prevalence: Total no. of cases in a given population at a given time. Indications that how long a disease affects the population.

Sporadic: Occurs only occasionally in given population.

Endemic: Constantly present in a population.

Epidemic: Acquired by many people in a given area in a short time period.

Pandemic: Worldwide epidemic.

Disease Prolongation

Acute: Rapid but short time. E.g. Influenza.

Chronic: Slow progression but long duration. E.g. TB

Subacute: Intermediate between acute and chronic. E.g. Sclerosing encephalitis.

Latent: Causative agent remains inactive, Can reactivate to produce symptoms. E.g. Herpes virus.

Disease caused by Infectious Agents

Production of poisons, such as toxins and enzymes, that destroy cells and tissues.

Direct invasion and destruction of host cell.

Triggering responses from the host's immune system which leads to disease signs and symptoms.

Disease is transmitted by many ways-

- **Contact transmission** (Direct, indirect, Droplet)
- **Vehicle transmission** (Airborne, Waterborne, Foodborne)
- **Vector Transmission** (Mechanical, Biological)
- **Nosocomial Infections** (Formites, Used apparatus, staff to patient)

Disease Progression

Incubation Period

- Before signs and symptoms appear.

Prodromal Period

- Early, mild, non- specific symptoms appear.

Period of Illness

- Specific symptoms seen which may be severe and immune system overcomes pathogen or patient dies.

Period of Decline

- Symptoms subside but may be vulnerable to secondary infections.

Period of Convalescence

- Patient's body returns to pre-disposing state.

Genetic Disorders

Genetic disorders or inherited disorders is a disease that is caused by an abnormality in an individual's DNA.

Abnormalities can arise due to mutations or addition/deletion of an entire chromosomes or set of chromosomes.

Most inherited disorders are quite rare and found in one person from every thousands or millions.

Inherited disorders are mainly of 2 types-

- Allosomal Disorder
- Autosomal Disorder

Allosomal Disorder

Inherited disorders related to sex chromosomes are called allosomal disorders.

Allosomes are referred to as sex chromosomes.

E.g.

- **Kleinfelter's syndrome**

This disorders takes place due to nondisjunction of the X chromosomes. At the time of conception the sperm containing both X and Y combines with an egg containing X chromosome which may contribute the extra X chromosome in male child (XXY).

In this males with some development of breast tissues, little body hair present, typically tall, have small testes, and are infertile due to absence of sperms.

- **Turners syndrome**

This disorder takes place due to absence of an entire sex chromosome. This is in case of female child due to monosomy X (X0).

It is associated with underdeveloped ovaries, short stature, webbed, broad chest, lack of secondary sexual characteristics and are sterile.

Autosomal Disorder

Disorders related to autosomes are referred to as autosomal disorder.

Autosome is a chromosome but it is not a sex chromosome and are in pairs.

We humans have a diploid genome which contains 22 pairs of autosomes and one pair of allosome making total 46 chromosomes.

E.g.

- **Downs syndrome**

This is caused due to nondysjunction of the 21^{st} chromosomes. Individual has a trisomy (3-21^{st} chromosomes). A person has small ears, small flattened nose, small mouth, tongue appears larger, short neck, small hands with short finger, mentally retarded.

•**Haemophilia**

It is a hereditary disorder that impairs the body's ability to control blood clotting or coagulation as a result if an individual suffering from this disorder gets cut then bleeding will not stop due to deficiency of clotting factors. It is recessive X-linked genetic disorder.

'Haemophilia A' has deficiency of clotting factor VIII.
'Haemophilia B' has deficiency of clotting factor IX.

•**Sickle cell anaemia**

It is an inherited, chronic disease in which the RBCs become crescent shaped. It occurs due to mutation in the haemoglobin gene. As a result they function abnormally and cause small blood clots with arise of recurrent painful sickle cell crises.

Thank you....