Chapter 1
General Concepts of Disease:
Principles of Diagnosis

Learning Objectives

• Define:
  – Disease
  – Lesions
  – Organic and functional disease
  – Symptomatic and asymptomatic disease
  – Etiology
  – Pathogenesis

• Categories of human disease
• Types of diagnostic tests and procedures

Characteristics of Disease (1 of 3)

• Disease: disturbance of body structure or function
• Lesions: well-defined, characteristic structural changes in organs and tissues as a result of disease
  • Organic disease
    – Associated with structural changes
    – Gross examination
    – Histologic examination
  • Functional disease
    – No morphological abnormalities yet body functions are profoundly disturbed
Characteristics of Disease (2 of 3)

- **Pathology**: study of disease
  - Pathologist: physician who specializes in diagnosing and classifying diseases by studying the morphology of cells and tissues
  - Clinician: physician/health care professional that cares for patients
- **Symptoms**: subjective manifestations such as pain or weakness
- **Signs**: physical findings or objective manifestations such as swelling or redness

Characteristics of Disease (3 of 3)

- Symptomatic disease: with symptoms and/or signs
- Asymptomatic disease: no signs or symptoms
  - Distinction between asymptomatic and symptomatic depends on extent
  - Early stages of disease, usually asymptomatic
  - If not treated, progresses to symptomatic
- **Etiology**: cause of disease
- Etiologic agent: agent responsible for causing disease
- **Pathogenesis**: process of development of disease
- **Pathogen**: any microorganism that causes disease

Classifications of Disease (1 of 3)

- Congenital and hereditary diseases
  - Developmental disturbances
  - Causes: genetic abnormalities; abnormalities in chromosome number or distribution; intrauterine injury; interaction of genetic and environmental factors
  - Hemophilia (hereditary), German measles (congenital)
Classifications of Disease (2 of 3)

• Inflammatory diseases: Body reacts to injury through an inflammatory process
  – Bacteria or microbiologic agents: sore throat
  – Allergic reaction: hay fever
  – Autoimmune diseases: SLE, diabetes type 1
  – Unknown etiology
• Degenerative diseases
  – Tissue or organ degeneration as a result of aging or breakdown
  – Arthritis, atherosclerosis

Classifications of Disease (3 of 3)

• Metabolic diseases: Disturbance in metabolic process in body
  – Diabetes, hyper- or hypothyroidism, fluid and electrolyte imbalance
• Neoplastic diseases: Uncontrolled cell growth
  – Benign: lipoma
  – Malignant: Lung cancer
• Basis of classification
  – 1. Similarity of lesions
  – 2. Similarity of pathogenesis
• Diseases with similarities may not necessarily be closely related

Health and Disease

– Good health: more than the absence of disease
  – Condition in which body and mind function efficiently and harmoniously as an integrated unit
• Traditional medicine: goal is to cure or ameliorate disease.
• Modern medicine: advances relieve suffering and advance human welfare but do not guarantee good health.
Continuum of Health and Disease

- Everyone is somewhere between the midpoint and good health
- Good health requires active participation, assuming responsibility for one’s health
  - Eat properly, exercise, avoid harmful excesses such as overeating, smoking, heavy drinking, or using drugs
  - Use one’s mind constructively, express emotions appropriately, nurture a positive mental attitude

Principles of Diagnosis

- Diagnosis: determination of nature and cause of illness
  - Clinical history
  - Physical examination
  - Differential diagnosis
- Prognosis: eventual outcome of disease
- Treatment
  - Specific treatment – directed at underlying cause
  - Symptomatic treatment – alleviates symptoms but does not influence course of disease

Clinical History (1 of 2)

- History of current illness
  - Severity, time of onset, and character of patient’s symptoms
- Medical history
  - Details of general health and previous illnesses that may shed light on current problems
- Family history
  - Health of patient’s parents and family members; diseases that run in families
Clinical History (2 of 2)

- Social history
  - Patient’s occupation, habits, alcohol and tobacco consumption, general health, current problems
- Review of symptoms
  - Symptoms other than disclosed in history of present illness, suggesting other parts of the body affected by disease

Physical Examination

- Physical examination
  - Systematic examination of patient, with emphasis on parts of body affected by illness
  - Abnormalities noted correlated with clinical history
- Differential diagnosis
  - Consideration of various diseases or conditions that may also explain patient’s symptoms and signs
  - Diagnostic possibilities narrowed by selected laboratory tests or other diagnostic procedures
  - Opinion of medical consultant may be sought

Screening Tests

- Screening tests for detection of disease
  - Detect early asymptomatic diseases amenable to treatment to prevent or minimize late-stage organ damage
- Screening for some genetic diseases
  - Screen for carriers of some genetic diseases transmitted from parent to child as either dominant or recessive trait
  - Identifying carriers allows affected persons to make decisions on future childbearing or management of current pregnancy
  - Example: recessive gene for sickle cell anemia in 8% of Black population
Requirements for Effective Screening

• A significant number of persons must be at risk for the disease in the group being screened.
• A relatively inexpensive noninvasive test must be available to screen for the disease that does not yield a high number of false-positive or false-negative results
• Early identification and treatment of the disease will favorably influence course of disease.

Diagnostic Tests and Procedures (1 of 8)

• Clinical laboratory tests
• Tests of electrical activity to measure electrical impulses associated with bodily functions and activities
  – ECG: measures serial changes in electrical activity of the heart in various phases of the cardiac cycle
  – EEG: measures electrical activity of brain; brain waves
  – EMG: measures electrical activity of skeletal muscle during contraction and at rest

Diagnostic Tests and Procedures (2 of 8)

• Radioisotope (radionuclide) studies: evaluate organ function by determining rate of uptake and excretion of substances labeled with a radioisotope
• Endoscopy
  – To examine interior of body using rigid or flexible tubular instruments equipped with lens and light source
  – To perform surgery formerly done through large abdominal incisions
Diagnostic Tests and Procedures (3 of 8)

• Ultrasound
  – Mapping echoes produced by high-frequency sound waves transmitted into body; echoes reflect change in tissue density, producing images
Diagnostic Tests and Procedures
(4 of 8)

• X-ray
  – Principle: use of high-energy radiation waves at lower doses to produce images to help diagnose disease
  – Can penetrate through tissues at varying degrees depending on tissue density
  – Act on a photographic film or plate (roentgenogram) as the rays leave the body

X-ray of colon with radiopaque barium sulfate
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X-ray of gallbladder: gallstones appear as radiolucent (dark) within a radiopaque bile (white)
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Opened gallbladder, same patient
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Diagnostic Tests and Procedures (5 of 8)

- Computed tomographic (CT) scans
  - Principle: radiation detectors record amount of X-rays or ionizing radiation absorbed by body and feed data into a computer that reconstructs the data into an image
Diagnostic Tests and Procedures  
6 of 8

• Magnetic resonance imaging (MRI)
  – Principle: computer-constructed images of body based on response of hydrogen protons in water molecules when placed in a strong magnetic field
Diagnostic Tests and Procedures (6 of 7)

• MRI: advantages over CT scan
  – Does not use ionizing radiation
  – Can detect abnormalities in tissues surrounded by bone, such as spinal cord, orbit, skull
  – Bone interferes with scanning because of its density but does not produce an image in MRI because of its low water content

Diagnostic Tests and Procedures (6 of 6)

• Cytologic and histologic examinations
  – Papanicolau (Pap) smear: identifies abnormal cells in fluids or secretions; for recognizing early changes that may be associated with cervical and other cancers
  – Biopsy: tissue samples obtained for histologic examination to determine abnormal structural and cellular patterns accompanying disease

Discussion

• Explain the requirements for an effective screening.
• Differentiate:
  – Symptomatic versus specific treatment
  – Sign versus symptom
  – Symptomatic versus asymptomatic disease
  – Diagnosis versus prognosis