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
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

   Computed tomographic scan, CT scan
   © Courtesy of Leonard Crowley, M.D./University of Minnesota Medical School

   CT scan, chest, white nodule on left lung indicates tumor
   © Courtesy of Leonard Crowley, M.D./University of Minnesota Medical School

   CT scan, abdomen at level of kidneys, fluid-filled cysts, right kidney
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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

   MRI, brain, with malformation within brain stem
   © Courtesy of Leonard Crowley, M.D./University of Minnesota Medical School
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