Define the common terms used to describe disease, such as lesions, organic and functional disease, symptomatic and asymptomatic disease, etiology, and pathogenesis.

List the major categories of human disease.

Explain the approach that a practitioner uses to make a diagnosis and decide on a patient’s treatment.

Describe the various types of diagnostic tests and procedures that can help the practitioner in making a diagnosis and deciding on proper treatment.

Characteristics of Disease

Any disturbance of structure or function of the body may be regarded as disease. A disease is often associated with well-defined, characteristic structural changes, called lesions, that are present in various organs and tissues. One can recognize lesions by examining the diseased tissue with the naked eye, which is called a gross examination, or with the aid of a microscope, which is called a histologic examination. Sometimes histologic examinations are supplemented by specialized studies that evaluate the properties of the cell membranes and the proteins within the cells. A disease associated with structural changes is called an organic disease. In contrast, a
functional disease is one in which no morphologic abnormalities (morph 5 structure or shape) can be identified even though body functions may be profoundly disturbed. However, as we develop new methods for studying cells, we can sometimes identify previously unrecognized abnormalities that disturb cell functions. Consequently, many of the traditional distinctions between organic and functional disease are no longer as sharply defined as in the past.

Pathology is the study of disease, and a pathologist is a physician who specializes in diagnosing and classifying diseases primarily by examining the morphology of cells and tissues. A clinician is any physician or other health practitioner who cares for patients. A disease may cause various subjective manifestations, such as weakness or pain, in an affected individual. These are called symptoms. A disease may also produce objective manifestations, detectable by the clinician, which are called signs or physical findings. In many diseases, the quantity of blood cells in the circulation may change, and so may the biochemical constituents in the body fluids. These alterations are reflected as abnormal laboratory test results.

A disease that causes the affected individual no discomfort or disability is called an asymptomatic disease or illness. A disease is often asymptomatic in its early stages. If the disease is not treated, however, it may progress to the stage where it causes subjective symptoms and abnormal physical findings. Therefore, the distinction between asymptomatic and symptomatic disease is one of degree, depending primarily on the extent of the disease.

The term etiology means cause. A disease of unknown etiology is one for which the cause is not yet known. Unfortunately, many diseases fall into this category. If the cause of a disease is known, the agent responsible is called the etiologic agent. The term pathogenesis refers to the manner by which a disease develops, and a pathogen is any microorganism, such as a bacterium or virus, that can cause disease.

Classifications of Disease

Diseases tend to fall into several large categories, although the diseases in a specific category are not necessarily closely related. Rather, the lesions produced by the various diseases in a category are morphologically similar or have a similar pathogenesis. Diseases are conveniently classified in the following large groups:

1. Congenital and hereditary diseases
2. Inflammatory diseases
3. Degenerative diseases
4. Metabolic diseases
5. Neoplastic diseases

Congenital and hereditary diseases are the result of developmental disturbances. They may be caused by genetic abnormalities, abnormalities in the numbers and distribution of chromosomes, intrauterine injury as a result of various agents, or an interaction of genetic and environmental factors. Hemophilia, the well-known hereditary disease in which blood does not clot properly, and congenital heart disease induced by the German measles virus are examples of diseases in this category.

Inflammatory diseases are those in which the body reacts to an injurious agent by means of inflammation. Many of the diseases characterized by inflammation, such as a sore throat or pneumonia, are caused by bacteria or other microorganismic agents. Others, such as “hay fever,” are a manifestation of an allergic reaction or a hypersensitivity state in the patient. Some diseases in this category appear to be caused by antibodies formed against the patient’s own tissues, as occurs in some uncommon diseases classified as autoimmune diseases. The etiology of still other inflammatory diseases has not been determined.

Degenerative diseases, the primary abnormality is degeneration of various parts of the body. In some cases, this may be a manifestation of the aging process. In many cases, however, the degenerative lesions are more advanced or occur sooner than would be expected if they were age related, and they are distinctly abnormal. Certain types of arthritis and “hardening of the arteries” (arteriosclerosis) are common examples of degenerative diseases.

Metabolic diseases

The chief abnormality seen in metabolic diseases is a disturbance in some important metabolic process in the body. For example, the cells may not be utilizing glucose normally, or the thyroid gland may not properly regulate the rate of cellular metabolism. Diabetes, disturbances of endocrine glands, and disturbances of fluid and electrolyte balance are common examples of metabolic diseases.

Neoplastic diseases

Neoplastic diseases are characterized by abnormal cell growth that leads to the formation of various types of benign and malignant tumors.

Health and Disease: A Continuum

Health and disease may be considered two extremes of a continuum. At one extreme is severe, life-threatening, disabling illness with its corresponding major effect on the physical and emotional well-being of the patient. At the other extreme is ideal good health, which may be defined as a state of complete physical and mental well-being. The healthy person is emotionally and physically capable of leading a full, happy, and productive life that is free of anxiety, turmoil, and physical disabilities that limit activities. Between these two extremes are many gradations of health and disease, ranging from mild or short-term illness that limits activities to some extent through moderate good health that falls short of the ideal state. The midpoint in this continuum may be considered a “neutral” position in which one is neither ill nor in ideal good health.

In this continuum, most of us are somewhere between midposition and the ideal state. The goal of traditional medicine is to cure or ameliorate disease. This is accomplished by various means, ranging from administering an antibiotic to cure an infection to very complex “high-technology” treatments such as kidney transplants and
heart surgery. The advances of modern medicine have done much to relieve suffering
and advance human welfare, but modern medicine does not guarantee good health.
Health is more than an absence of disease; it is a condition in which body and mind
function efficiently and harmoniously as an integrated unit. Consequently, we must
make an active part in achieving good health by assuming some responsibility for our
own physical and emotional well-being. This means practicing such common sense
measures as eating properly, exercising moderately, and avoiding harmful excesses
such as overeating, smoking, heavy drinking, or using drugs, which can disrupt phys-
cical or emotional well-being. Taking responsibility for one’s health also requires us-
ing one’s mind constructively, expressing emotions, and feeling good about oneself.
Positive mental attitudes are essential for good health because negative feelings may be
reflected in disturbed bodily functions that are manifested as disease.

**Principles of Diagnosis**

The determination of the nature and cause of a patient’s illness by a physician or
other health practitioner is called a diagnosis. It is based on the practitioner’s eval-
uation of the patient’s subjective symptoms, the physical findings, and the results of
various laboratory tests, together with other appropriate diagnostic procedures.
When the practitioner has reached a diagnosis, he or she can then offer a prognosis:
an opinion concerning the eventual outcome of the disease. Then a course of treat-
ment is instituted.

**THE HISTORY**

The clinical history is a very important part of the evaluation. It consists of several parts:

1. The history of the patient’s current illness
2. The past medical history
3. The family history
4. The social history
5. The review of systems

The history of the present illness elicits details concerning the severity, time of
onset, and character of the patient’s symptoms. Many diseases have characteristic
symptoms. The patient’s description of the oppressive substernal pain of a heart at-
tack or the pain and urinary disturbances associated with a bladder infection, for
example, may provide very helpful information that suggests the correct diagnosis.
The past medical history provides details of the patient’s general health and previ-
ous illnesses. These data may shed light on the patient’s current problems as well.
The family history provides information about the health of the patient’s parents
and other family members. Some diseases, such as diabetes and some types of heart
disease, tend to run in families. The social history deals with the patient’s occupa-
tion, habits, alcohol and tobacco consumption, and similar data. This information
may also relate to the patient’s general health and current problems. The review of
systems inquires as to the presence of symptoms other than those disclosed in the
history of the present illness; such symptoms might suggest disease affecting other
parts of the body. For example, the practitioner inquires about such symptoms as
pain or burning on urination, which suggest an abnormality of the urinary tract, and
coughing, shortness of breath, or chest pain, which may indicate disease of the res-
piratory system. In this way, possible dysfunctions of other organ systems are eval-
uated by systematic inquiry.

**THE PHYSICAL EXAMINATION**

The physical examination is a systematic examination of the patient. The practi-
tioner places particular emphasis on the part of the body affected by the illness, such
as the ears, throat, chest, and lungs in the case of a respiratory infection. Any ab-
normities detected on the physical examination are correlated with the clinical his-
tory. At this point, the practitioner begins to consider the various diseases or conditions
that would fit with the clinical findings. Sometimes, more than one possible diagno-
sis needs to be considered. In a differential diagnosis the practitioner considers a
number of diseases that are characterized by the patient’s symptoms. For example,
if a patient complains of shortness of breath and abnormalities are detected when
the lungs are examined with a stethoscope, the practitioner may consider both chronic
lung disease and chronic heart failure in the differential diagnosis.

Often the practitioner can narrow the list of diagnostic possibilities and arrive
at a correct diagnosis by using selected laboratory tests or other specialized diagnos-
tic procedures. In difficult cases, the clinician may also wish to obtain the opinion
of a medical consultant, who is a physician with special training and experience in
the type of medical problem presented by the patient.

**TREATMENT**

After the diagnosis has been established, a course of treatment is initiated. There are
two different types of treatment: specific treatment and symptomatic treatment.
A specific treatment is one that exerts a highly specific and favorable effect on
the basic cause of the disease. For example, an antibiotic may be given to a patient
who has an infection that is responsive to the antibiotic, or insulin may be given to
a patient with diabetes. Symptomatic treatment, as the name implies, makes the pa-
tient more comfortable by alleviating symptoms but does not influence the course
of the underlying disease. Examples are the treatment of fever, pain, and cough by
means of appropriate medications. Unfortunately, there are no specific treatments
for some diseases. Consequently, the clinician must be content with treating the man-
ifestations of the disease, without being able to influence its ultimate course.

**Screening Tests for Disease**

**PURPOSE AND REQUIREMENTS FOR EFFECTIVE SCREENING**

Many diseases that respond to treatment are asymptomatic initially. If untreated,
however, the disease often progresses slowly, causing gradual but progressive organ
damage until eventually the person is seriously ill with far advanced organ damage
cased by the disease. Unfortunately, treatment of late-stage disease is often much
less effective and may not be able to restore the function of the organs that have been
damaged. Had the disease been identified and treated in its early asymptomatic stage,
persons in a population carry a recessive gene that can be detected by relatively simple screening tests, identifying carriers allows the affected persons to make decisions regarding future childbearing or management of a future pregnancy. One high-incidence recessive gene for which screening is available is the sickle hemoglobin gene, which occurs in about 8 percent of the black population. A child born to two carriers of the sickle hemoglobin gene who receives the sickle hemoglobin gene from each parent will develop a severe anemia called sickle-cell anemia. The sickle hemoglobin gene and its clinical manifestations are considered in Chapter 14. Other examples of genetic diseases for which screening is available are described in Chapter 9.

### Groups Suitable for Screening

Screening tests should target a group of persons in whom there is a relatively high frequency of disease, and tests should also target the age group in whom the disease is likely to be present. If the disease, for example, has its onset in middle age, then screening adolescents and children in the target group would not be productive.

#### Suitable Screening Tests

Screening a group of persons for a disease in its early asymptomatic stage requires some type of test that can identify some characteristic manifestation of the disease, such as high blood sugar in the case of diabetes, or the presence of blood in the stool in the case of a colon tumor. A test used for screening should be reasonably inexpensive and should have few false-positive results (test is positive when no disease is present) and few false-negative results (test is negative when disease is present). If the test produced a large number of false-positive results in the group being screened, many persons with false-positive test results would have to undergo more extensive and sometimes invasive testing, as well as a comprehensive medical evaluation, only to find that the test result was a “false alarm” and that they did not have the disease. The other hand, less sensitive screening tests would yield an excess of false-negative tests, and many persons who actually had the disease would not be detected.

#### Benefits of Screening

Screening test results should provide some benefit to the person being screened. Generally, there is no point in screening for a disease if no treatment is available to arrest the progression of the disease.

Examples of widely used cost-effective screening tests for disease include urine tests to detect glucose in the urine as a screening test for diabetes, tests to detect blood in the stools to screen for colon tumors, Papnicolaou smears (Pap tests) to screen for abnormalities in the epithelium of the uterine cervix that predispose to cancer, and breast x-ray examinations (mammograms) to screen for very early breast cancer at a stage when it can be treated most effectively.

### Screening for Genetic Disease

Screening tests can also be used to screen for carriers of some genetic diseases that are transmitted from parent to child as either dominant or recessive traits. When many persons in a population carry a recessive gene that can be detected by relatively simple screening tests, identifying carriers allows the affected persons to make decisions

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**The image contains a page from a book discussing clinical laboratory tests and procedures.**

**Clinical laboratory tests**

Clinical laboratory tests have many uses. They can be used to determine the concentration of various constituents in the blood and urine, which are frequently altered...
The ability of the thyroid gland to concentrate and utilize radioactive iodine is used as a measure of thyroid function and can also be used to detect tumors within the thyroid gland. One may administer a radioactive material that is filtered out or concentrated in a tissue or organ and then measure the radioactivity by radiation detectors applied to the exterior of the body and connected to a computer. For example, specially processed albumin labeled with a radioisotope may be administered intravenously as a measure of pulmonary blood flow. The material is filtered out and retained in the lungs as the blood flows through them. If blood flow to a part of the lung is inadequate for any reason, less radioactivity is recorded in that area. This technique is frequently used to detect the presence of blood clots in the lung that impede blood flow to parts of the lung. Phosphorus-containing isotopes are concentrated in the skeletal system. If there are deposits of tumor in bone, the isotopes are concentrated around the tumor deposits and can be easily identified (FIGURE 1-1). Radiopaque materials injected intravenously can also be used to evaluate blood flow to heart muscle and to identify areas of damaged heart muscle.

**GENERAL CONCEPTS OF DISEASE**

**CHAPTER 1**

**DIAGNOSTIC TESTS AND PROCEDURES**

**TESTS OF ELECTRICAL ACTIVITY**

Several different tests measure the electrical impulses associated with various bodily functions and activities. These include the electrocardiogram (ECG), the electroencephalogram (EEG), and the electromyogram (EMG). The most widely used of these tests is the ECG. Electrodes attached to the arms, legs, and chest are used to measure the serial changes in the electrical activity of the heart during the various phases of the cardiac cycle. The ECG also identifies disturbances in the heart rate or rhythm and identifies abnormal conduction of impulses through the heart. Heart muscle injury, such as occurs after a heart attack, can also be recognized by means of characteristic abnormalities in the cardiogram. The EEG measures the electrical activity of the brain, often called brain waves, by means of small electrodes attached to different areas in the scalp. Brain tumors, strokes, and other abnormalities of cerebral structure or function may cause altered brain wave patterns that are detected by this examination. The EMG measures the electrical activity of skeletal muscle during contraction and at rest. Abnormal electrical activity is often encountered in various inflammatory or degenerative diseases involving the skeletal muscles. The test is performed by inserting a needle into the muscle that is being studied. The speed at which a nerve conducts impulses can also be measured by means of electrodes taped to the surface of the skin over the nerve being tested. Abnormal conduction of nerve impulses, encountered in some diseases, can be identified by such studies.

**RADIOISOTOPE (RADIONUCLIDE) STUDIES**

The function of various organs can be evaluated by administering a substance labeled with a radioactive material called a radioisotope. Specially designed radiation detectors then measure the uptake and excretion of the labeled substance. For example, in certain types of anemia, one measures the absorption and excretion of radioisotope-labeled vitamin B12, which is a vitamin required for normal blood formation. The ability of the thyroid gland to concentrate and utilize radioactive iodine is used as a measure of thyroid function and can also be used to detect tumors within the thyroid gland. One may administer a radioactive material that is filtered out or concentrated in a tissue or organ and then measure the radioactivity by radiation detectors applied to the exterior of the body and connected to a computer. For example, specially processed albumin labeled with a radioisotope may be administered intravenously as a measure of pulmonary blood flow. The material is filtered out and retained in the lungs as the blood flows through them. If blood flow to a part of the lung is inadequate for any reason, less radioactivity is recorded in that area. This technique is frequently used to detect the presence of blood clots in the lung that impede blood flow to parts of the lung. Phosphorus-containing isotopes are concentrated in the skeletal system. If there are deposits of tumor in bone, the isotopes are concentrated around the tumor deposits and can be easily identified (FIGURE 1-1). Radiopaque materials injected intravenously can also be used to evaluate blood flow to heart muscle and to identify areas of damaged heart muscle.

**ENDOSCOPY**

An endoscopy, or endoscopic examination (endo 5 within 1. akopes 5 examine), is an examination of the interior of the body by means of various types of rigid or flexible tubular instruments that are named according to the part of the body they are designed to examine. These instruments have a system of lenses for viewing and a light source to illuminate the region being examined. An endoscopy, for example, is used to examine the interior of the esophagus, a gastroscope to examine the stomach, and a bronchoscope to examine the trachea and major bronchi. An instrument for viewing the interior of the bladder is called a cystoscope. A sigmoidoscope is a rigid tube used to examine the rectum and the sigmoid colon, and a colonoscope is a flexible tube that can be used to examine the entire length of the colon. An instrument called a laparoscope is used to visualize the pelvic organs. It is inserted into the abdominal cavity through a small incision in the umbilicus.

**ULTRASOUND**

Ultrasound is a technique for mapping the echoes produced by high-frequency sound waves transmitted into the body. Echoes are reflected wherever there is a change in the density of the tissue. The reflected waves are recorded on sensitive detectors, and images are produced. This method is widely used to study the uterus during pregnancy because it does not require the use of potentially harmful radiation and poses no risk to the fetus (FIGURE 1-2). The technique can be used to determine the position of the placenta and the fetus within the uterus; it can also identify some fetal abnormalities and detect twin pregnancies. Ultrasound is also used to study the structure and function of the heart valves. The procedure can detect valve abnormalities and
bronchi can be visualized by instilling a radiopaque oil into the bronchi. The oil forms a thin film on the bronchial mucosa and delineates the contours of the bronchi. This procedure is called a bronchogram (FIGURE 1-4).

One uses the same principle to visualize the urinary tract. A radiopaque substance is injected into a vein and is excreted in the urine as the blood flows through the kidney, outlining the contour of the urinary tract. This is called an intravenous pyelogram (IVP) (FIGURE 1-5). Another method is to introduce the dye directly into both ureters through tubes that are inserted into both ureters by means of a cystoscope introduced into the bladder. This procedure is called a retrograde pyelogram. To visualize the gallbladder, the patient ingests tablets of radiopaque material that is absorbed into the circulation, excreted by the liver in the bile, and concentrated in the gallbladder. Gallstones can be identified because they occupy space in the gallbladder and cause irregularities in the radiopaque material concentrated there (FIGURE 1-6).

X-RAY EXAMINATION

X-ray examinations are conducted in many ways, but the basic principle is the same for all types of x-ray studies. X-rays are passed through the part of the body to be examined, and the rays leaving the body expose an x-ray film. The extent to which the rays are absorbed by the tissues as they pass through the body depends on the density of the tissues. Tissues of low density, such as the air-filled lungs, transmit most of the rays, and thus, the film exposed to x-rays passing through them appears black. Tissues of high density, such as bone, absorb most of the rays; the film remains unexposed and appears white. Tissues of intermediate densities appear as varying shades of gray. The x-ray image produced on the film is called a radiograph or roentgenogram. The same basic principle is used to obtain x-ray films of the breast. This procedure is called a mammogram. The applications and limitations of the mammogram procedure are considered in Chapter 16 in the section on diseases of the breast.

Although the linings of internal organs such as the intestinal tract, urinary tract, bronchi, fallopian tubes, and biliary tract have little contrast, they can be examined by administering a dense radiopaque substance called contrast medium. It coats and adheres to the lining of the structure being examined and enhances its visibility. To examine the interior of the gastrointestinal tract, for example, one gives the patient a suspension of barium sulfate to swallow or administers it as an enema. The opaque barium coats the lining of the intestinal tract, and an abnormality in the lining shows on the film as an irregularity in the column of barium (FIGURE 1-3). The lining of the

identify blood clots that sometimes form on the heart valves in association with infection of the valve (described in Chapter 13). Ultrasound can determine the thickness of the ventricular walls and septum and the size of the ventricular chambers during systole and diastole. Ultrasound can identify gallstones in the gallbladder and abnormalities in the prostate suspicious for prostate cancer. The technique has many other applications in medicine.
One can also use contrast material to study the flow of blood in large arteries and to identify areas of narrowing or obstruction. This procedure is called an arteriogram or angiogram (angio = blood vessel). A small flexible catheter is inserted into a large artery in the arm or leg and advanced into the aorta until it is positioned at the opening of the artery that is to be examined. Radiopaque material is then injected through the catheter. It mixes with the blood, and its flow through the vessel is followed by means of a series of x-ray films. If the vessel is narrowed by disease, the film will show areas in which the column of opaque material is narrowed. A complete obstruction of the vessel appears as an interruption of the column. Arteriography is often used to detect narrowing or obstruction of the coronary arteries or of the carotid arteries in the neck, which carry blood to the brain (FIGURE 1-7). Obstruction of the pulmonary arteries by blood clots also can be identified by arteriography. In this case, the catheter used to inject the radiopaque material is inserted into a large vein in the arm, threaded up the vein and through the right side of the heart, and positioned in the pulmonary artery. This same basic method can be used to study the flow of blood through the heart and can detect abnormal communications between cardiac chambers. This type of study is called cardiac catheterization.

**Computed Tomographic Scans**

A computed tomographic scan (CT scan) is performed by a highly sophisticated x-ray machine that produces images of the body in cross section by rotating the x-ray tube around the patient at various levels. The x-ray tube is mounted on a movable frame opposite an array of sensitive radiation detectors that encircle the patient. As the x-ray tube moves around the patient, the radiation detectors record the amount of radiation passing through the body (FIGURE 1-8). In computerized scanning, the amount of radiation absorbed is not read directly on an x-ray film. Instead, the data from the radiation detectors are fed into a computer, which reconstructs the data into an image that reproduces the patient's anatomy as a cross-section picture. The image is displayed on a television monitor and can be recorded on film (FIGURE 1-9). As with x-ray detector rotates to remain opposite the x-ray source
CT scanner
X-ray source generates the beam of X-rays and rotates around patient
Direction of rotation of X-ray source

CT scan of chest. Mediastinum and heart appear white in the center of scan, with less dense lungs on either side. The arrow indicates a lung tumor which appears as white nodule in lung.

CT views of the abdomen at the level of kidneys, revealing a fluid-filled cyst in the kidney (arrow). The cyst appears less dense than surrounding renal tissue. The opposite kidney (right side of photograph) appears normal.
conventional x-rays, dense substances are white and less dense substances appear darker in proportion to the amount of radiation they transmit. The individual organs appear sharply separated from one another because the various parts of the body are separated by planes of fat, which have very low density. These separations increase contrast between adjacent organs. Abnormalities of internal organs that cannot be identified by means of standard x-ray examinations can often be discovered with CT scans. FIGURE 1-10 shows a renal cyst located by CT scan.

MAGNETIC RESONANCE IMAGING

Magnetic resonance imaging (MRI) produces computer-constructed images of various organs and tissues somewhat like CT scans. The device consists of a strong magnet capable of developing a powerful magnetic field, coils that can transmit and receive radiofrequency waves, and a computer, which receives impulses from the scanner and forms them into images that can be interpreted. The MRI scanner with the enclosed magnet and coils appears similar to a CT scanner. The patient lies on a table that is gradually moved into the scanner, as is done in CT scans. The principle of MRI, however, is quite different from that of CT scanning, which uses ionizing radiation to construct images based on the density of tissues. MRI scans, in contrast, depend on the response of hydrogen protons (positively charged particles in the nucleus around which electrons rotate) contained within water molecules when they are placed in a strong magnetic field. Hydrogen protons behave as if they are spinning rapidly about an axis, surrounded by orbiting negatively charged electrons. When subjected to a strong magnetic field, the protons become aligned in the direction of the magnetic field. When a pulse of radiofrequency waves is directed at the protons, they are temporarily dislodged from their orientation, which causes them to wobble. As they return to their original positions, they emit a signal (resonance) that can be measured and used to produce the computer-constructed images. Body tissues, which have a high water content, are a rich source of protons capable of excitation. The intensity of the signals produced is related to the varying water content of body tissues and to the strength and duration of the radiofrequency pulse. Because an MRI does not use ionizing radiation, the patient does not receive radiation exposure. An MRI does expose the patient to strong magnetic fields and radio waves, but this appears relatively safe, on the basis of current knowledge.

Applications

An MRI detects many of the same types of abnormalities detected by a CT, and a CT is superior to an MRI for many applications. An MRI, however, offers distinct advantages over CT in special situations, as, for example, when attempting to detect abnormalities in tissues surrounded by bone, such as lesions in the spinal cord, orbits, or near the base of the skull (FIGURE 1-11). In these locations, bone interferes with scanning because of its density, but it does not produce an image in MRI because the water content of bone is low. MRI also provides a sharp contrast between gray and white matter within the brain and spinal cord, which differ in their water content. For this reason, the technique is useful for demonstrating areas where myelin sheaths of nerve fibers have been damaged, as in a neurologic disease called multiple sclerosis (described in Chapter 26). Further improvements in equipment will undoubtedly increase the usefulness of this diagnostic procedure.

POSITRON EMISSION TOMOGRAPHY

Related to radioisotope studies but much more complex and sophisticated is one of the newest of the diagnostic imaging tests called positron emission tomography (PET), or simply PET scans. Positrons are unique subatomic particles that have the same mass as electrons but carry a positive charge. They are formed when atoms such as carbon, oxygen, or nitrogen are bombarded in a cyclotron with high-energy particles, which breaks down the atomic nuclei and releases the positrons along with other subatomic particles. Positrons escaping from the nuclei collide with negatively charged electrons circling the nuclei, producing radiation that can be detected and measured by sensitive radiation detectors. One uses PET scans to study body functions by injecting into the subject a biochemical compound, such as glucose, that is labeled with a positron-emitting isotope and then assessing the distribution and metabolism of the compound by measuring the radiation produced within the body by the isotope-labeled compound. The radiation output, measured by sensitive radiation detectors, is fed into a computer that constructs computer-generated images similar to those obtained with CT scans. Such studies provide information on the metabolic activities of the organ or tissue being studied, the site within an organ where the compound is being metabolized, and the blood flow to the organ being studied.

Although originally developed for research studies, PET scans are slowly moving from the research laboratory into medical practice. At present, the major clinical applications are to assess biochemical functions within the brain. One can detect and measure changes in brain functions associated with various neurologic diseases such as strokes, brain tumors, Alzheimer’s disease, Parkinson’s disease, and some hereditary de-
generative diseases of the nervous system. The method has also been used to some extent to evaluate changes in blood flow and metabolism in heart muscle after a heart attack. PET scans following intravenous infusion of a labeled glucose compound have also been used to distinguish a benign from a malignant tumor growing within the body, based on the greater glucose uptake and metabolic activity within a malignant tumor, in contrast to the much lower labeled glucose uptake in a benign tumor. This same approach is also used to identify deposits of malignant tumor that have spread throughout the body by demonstrating the increased glucose uptake within the tumor deposits. Many other applications of PET scans are being explored and evaluated.

Although PET scans provide useful information, there are some major drawbacks to their widespread application. They are very expensive procedures and are not widely available. Because positron-emitting isotopes must be produced in a cyclotron, the isotopes produced have a very short duration of activity (half-life), and one must have facilities for incorporating the isotope into the biochemical compound required for the PET scan procedure.

**CYTOLOGIC AND HISTOLOGIC EXAMINATIONS**

Cells covering the surfaces of the body are continually cast off and replaced by new cells. Abnormal cells can often be identified in the fluids or secretions that come in contact with the epithelial surface. This type of examination is called a Papanicolaou smear, or simply Pap smear, after the physician who developed the procedure. It is widely used as a screening test for recognizing early cancer of the uterus and can be used to detect cancers in other locations as well. The Pap smear is discussed in the section on neoplasms in Chapter 10.

Diseased tissues have abnormal structural and cellular patterns that can be recognized by the pathologist. Consequently, it is often possible to determine the cause of a patient’s disease by histologic examination of a small sample of tissue removed from the affected tissue or organ. This procedure is called a biopsy. Samples of tissue can be obtained from any part of the body. Gastroscopes, bronchoscopes, and other instruments used for endoscopic examination, for example, are constructed so that specimens for biopsy can be obtained while the internal organs are being examined. Biopsy specimens can also be taken directly from internal organs such as the liver or kidney by inserting a thin needle through the skin directly into the organ. Samples of bone marrow are obtained in this way, and bone-marrow biopsy is often performed to diagnose blood disease (FIGURE 1-12).

**FIGURE 1-12**

Two samples of bone marrow (adjacent to scale) obtained from pelvic bone by means of a specially designed needle, shown in the upper part of the photograph.

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QUESTIONS FOR REVIEW

1. What are the five major categories of disease?
2. What are the definitions of the following terms: etiology, symptom of disease, sign of disease, diagnosis, and prognosis?
3. How does an organic disease differ from a functional disease?
4. What principal factors does the physician evaluate in arriving at a diagnosis?
5. What is the difference between specific and symptomatic treatment?
6. What are the major categories of diagnostic tests and procedures that can help the practitioner make a diagnosis? Give some examples.
7. What is the difference between an invasive and a noninvasive procedure?
8. What are the basic concepts on which the following procedures are based: Pap smear, x-ray examinations, ultrasound, electrocardiogram, and CT scans?

SUPPLEMENTARY READINGS

Describes principles and applications of this powerful diagnostic tool.

Describes applications and limitations of health screening. Intervention must be able to detect, treat, or prevent the condition it has been designed to deal with. We must not harm the patient in our zeal to prevent disease.

Excellent discussion of risks and benefits of therapeutic procedures, as well as biases. The wish to do everything possible for the patient must not lead to misguided actions.


Decisions about options for treatment of a disease should be a joint effort on the part of both the physician and the patient, and the patient needs to understand that every decision is influenced by uncertainty and risk. The physician can explain the possible risks and benefits of various methods of treatment, but the patient must make the final decision.


An update on the current status of computer-assisted imaging.