OVERVIEW OF PATHOLOGY AND ITS RELATED DISCIPLINES

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Summary

Pathology is the science of disease. It deals with deviations from normal body function and

structure. Many disciplines are involved in the study of disease, as it is necessary to understand the complex causes and effects of various disorders that affect the organs and body as a whole. This article outlines the major disorders, the different disciplines involved in their study and the various diagnostic methods employed. Included in this article are three selected topics of special interest, being good representatives of general pathologic processes and organ related disorders.

1. Introduction

Pathology is the science of disease (abnormalities in structure and function). It is the study of abnormal anatomy, abnormal histology, abnormal biochemistry and abnormal physiology. It comprises a knowledge and understanding of functional and structural changes in disease states, from the molecular level, right up to the effects on the patient and hence is important in providing the link between basic biologic as well as environmental sciences and the practice of medicine (Figure 1).

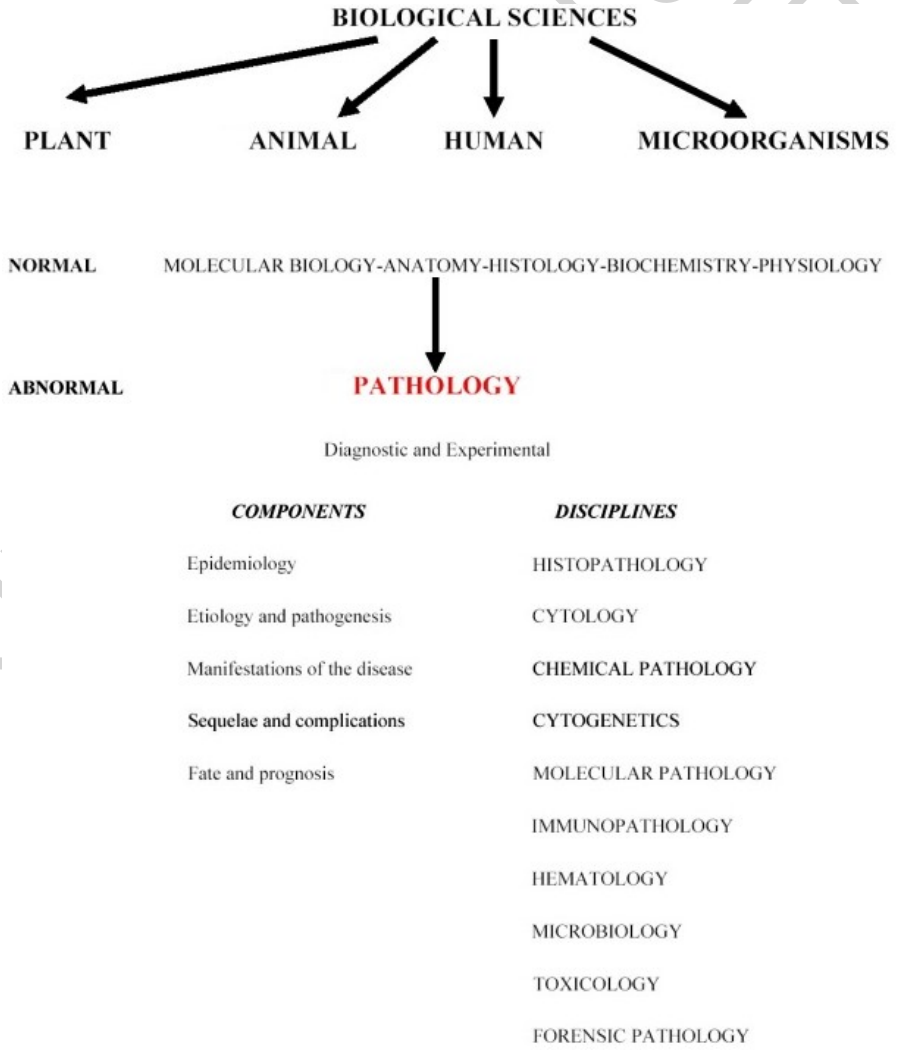


Figure 1: Pathology and its related disciplines in the hierarchy of biological sciences

The ultimate goal of pathology is identifying the causes of a disease thereby paving the way to its treatment or its prevention

1.1 Pathology coverage

The approach towards studying a disease in pathology follows the following lines (Figure 1):

- Definition and epidemiology: name of the disease, its *incidence, prevalence* in a population and its geographic distribution.
- Etiology and pathogenesis: (*cause* and *mechanism):* deals with injurious agents that have caused a particular disease and how they came about to produce the damage.
- Manifestations: Pathology entails the description of any deviation from normal on the tissue as well as the body level: a) Organ morphology (*gross pathology and microscopic pathology*): deals with changes in the appearance of an organ (gross or macroscopic) and on the level of the tissue forming that organ (microscopic). b) Body reaction: Is the effect of damage in a particular organ and other body systems which are involved in the process and this includes the clinical picture
- Sequelae and complications: Involves the course or progress of the disease and its unhealthy effects.

Prognosis and fate: includes the disease outcome or anticipated course
Good prognosis: cure or recovery
Bad prognosis: progression of disease (morbidity) or death (mortality) complications (unhealthy effects), occurring at the site of disease and away from the site (spread to other organs).

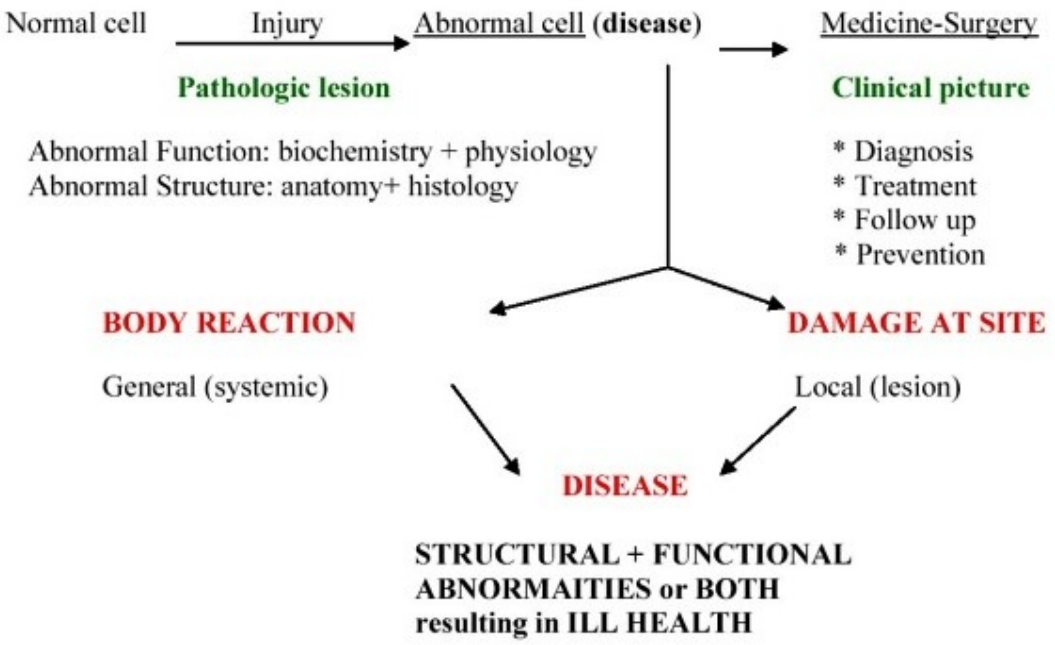


Figure 2: Effects of injury on the normal cell and emergence of a disease

1.1.1 Etiology and Pathogenesis of a Disease

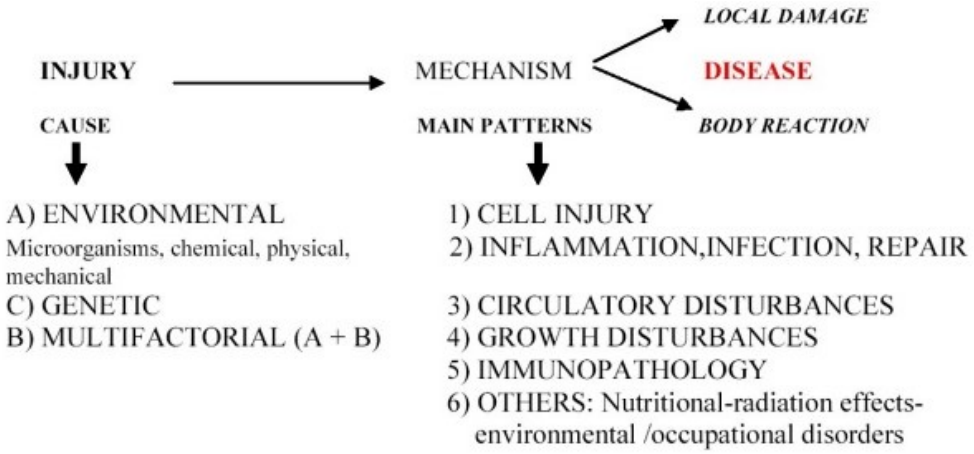
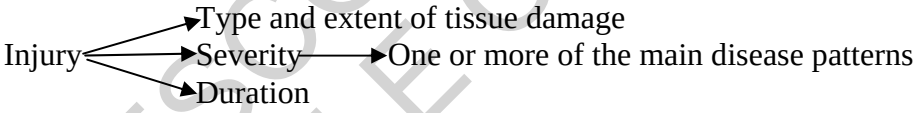


Figure 3: Causes and main patterns of disease

In some cases *etiology* is vague, or not yet known, such conditions are termed primary disorders, *idiopathic*, essential, spontaneous or cryptogenic. When a disease *etiology* is unclear, but known to occur commonly in certain groups of people sharing same occupation or concentrated in a certain geographic location or is even related to having a common habit, these common factors act as clues to the occurrence of a disease and is hence termed *risk factors***.**

1.1.2 Manifestations of Disease (Lesions)



Very mild of short duration: No change (body can overcome injury) OR just a functional disturbance like pain

Very mild and long duration, Moderate injury with short duration Or severe but very early (at start): Functional abnormality with or without *ultrastructural* change (changes at the level of cell organelles)

Severe of short duration or Mild or moderate and prolonged: Functional and morphological Depending on the above factors and the tissue type affected, injury commonly results in the following cellular changes:

- Reversible changes e.g. degenerative changes in cells (sick cells which have a chance of recovery)
- Irreversible changes: e.g. necrosis (cell death)

1.1.3 Phases Of A Disease Process (Course)

Phase I, Functional disturbance stage: At this level of affection, samples obtained from body fluids reveal mostly functional changes, which can be diagnosed by hematology, chemical pathology or by molecular and genetic methods

Phase II, Functional and early morphologic changes: At this level, cellular changes may be diagnosed from body fluids by hematology, chemical pathology and cytology or in tissue by biopsy and submitted for molecular and genetic methods as well as electron *microscopy*

Phase III, Functional and morphologic changes: diagnosed from body fluids submitted to hematological, cytological, molecular and genetic evaluation or tissue *biopsy*, to be evaluated like phase II of diseases, by microscopic examination in addition to gross organ or tissue evaluation

1.2 Physician's approach to patient

The clinician or attending physician starts by :

1. History taking: to determine epidemiological data and symptoms of the patient
2. Clinical examination: to identify signs of the disease discovered on examination
3. Provisional diagnosis and differential diagnosis is based on signs and symptoms
4. Final diagnosis: usually after requesting special investigations or sample taking

a. Investigations: imaging, x-ray, ultrasound, CT scan etc.
b. Laboratory work out: pathologists and other laboratory specialties

5. Treatment is either curative (cures) or palliative (can't cure but helps symptom relief).Treatment depends on *diagnosis* and *prognosis*. Follow up even after recovery, may be required in some cases, as with recurrent tumors.

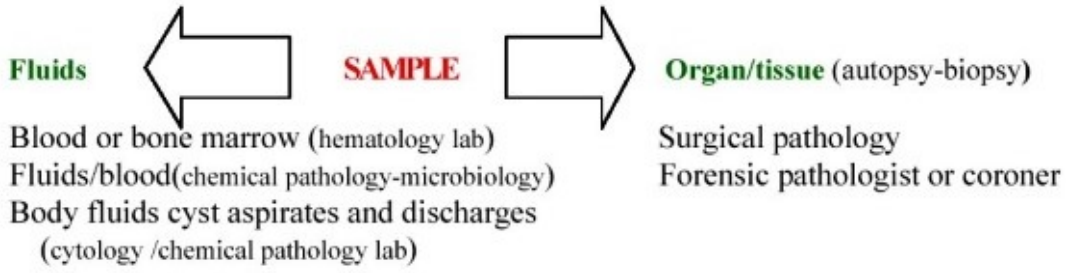


Figure 4: Types of Pathology samples

1.3 Types of pathologists and affiliated specialties

Surgical pathologist, diagnostic pathologist or histopathologist: Deals with surgically removed tissues from living patients (*biopsy*) and also with *autopsy* samples, to determine cause of death.

Cytologist or cytopathologist: Is responsible for making a *diagnosis* based on cell morphology and deals with all fluid samples, discharge, fine needle, lavage and brush samples.

Forensic pathologist: Deals with *autopsy* samples in medico- legal cases.

Hematopathologist: Handles the *diagnosis* of blood and bone marrow samples.

Chemical pathologist: Measures and interprets substances present in blood body fluids and tissues.

Immunopathologist: Investigates immune responses
Other disciplines working in close conjunction and associated with the pathologist include:

Cytogenetisist: Responsible for investigation of genetic and chromosomal abnormalities.
Microbiologist: Detects and identifies viruses, bacteria, fungi and parasites

1.4 Role of pathologist

Is to address the problem rather than its effect

This can be achieved by:

1. Diagnostic pathology: includes surgical biopsy material as well as body fluid cytology samples. Examination of this material is important for making the final diagnosis, monitoring of treatment and follow up of patients. Once clinician receives the Pathology report he starts to treat his patient accordingly, based on the final diagnosis (Figure 4)
2. Autopsy: Examination of the body of a patient after death is important for:

- Determining cause of death
- Procure statistical information about a disease and determining mortality and morbidity rates
- Auditing the accuracy of the clinical diagnosis and treatment methodology
- Education of medical profession teams, undergraduates and postgraduate students.
- Supplies researchers with material necessary for studying cause and mechanisms of particular disease entities.

Performance of an autopsy requires a midline incision from the neck to symphysis pubis, enabling the pathologist to remove thoracic and abdominal organs. The brain is also removed by making a skin flap and opening the cranium. All organs are weighed and described grossly. Small pieces of tissue are taken from normal and abnormal looking areas for microscopic examination, chemical pathology analysis and microbiologic cultures. In cases where autopsy is needed, but the deceased's family has refused permission for autopsy, the ultimate limited autopsy can be performed without actually disfiguring the body by means of fine needle or core biopsy sampling (a wider bore of needle). This technique is good for assessing liver disease especially hepatitis B & C. It is also used in highly infectious cases, where risk of infection may outweigh the need for autopsy (Figure 4.)

3. Experimental pathology: Deals with observation of effects on experimental animal models or in a tissue culture setting.

2. Pathology and its related disciplines

Diagnostic and experimental pathology involve obtaining scientific and biological knowledge from fluids or tissue material of patients, experimental animal models and cell culture studies. In diagnostic pathology the analysis is made at the level of the disease itself i.e. cause, mechanism and effects of the disease on organ systems or organs. Experimental pathology on the other hand deals with observation of effects on experimental animal models or in a tissue culture setting.

Surgical pathology, histopathology, cellular or anatomic pathology: Involves the *diagnosis* of a disease based on examination of the tissues both macroscopically and microscopically.

Cytology: Involves the *diagnosis* of a disease based on examination of body fluids and small tissue fragments.

Hematology: Is the study of diseases of blood and bone marrow
Chemical pathology: Entails the investigation of metabolic disturbances by changes in the concentration of substances in body fluids and to a lesser extent in tissue.

Microbiology: Study and *diagnosis* of infectious diseases and the organisms responsible for them. It encompasses a wide range of causative agents as bacteria, viruses, fungi and parasites.

Molecular pathology: This entails the study of the biochemical based changes in a single nucleotide in genomic *DNA* resulting in a defective gene product, which may produce a *lesion* in some disorders as congenital diseases and cancer.

Cytogenetics: Involves the study of abnormal chromosomes and genes responsible for certain diseases i.e. (karyotyping).

NB In cytogenetics The *karyotype* is examined using a blood sample to determine numerical (aneuploidy or polyploidy) or structural abnormalities in chromosomes, as deletions and ring chromosome, translocations, centric fragment, isochromosome and inversion, whereas in *molecular pathology* examination of specific areas of *DNA* on the chromosome is performed.

Immunopathology: Entails the study of disorders of the body defense system.

Toxicology and Forensic pathology: The former studies the changes produced secondary to introduction of a known toxin into the body and the latter encompasses the whole scope of pathology applied to legal purposes.

2.1 Cytology

Is the science of cell structure: Compared to histology, the diagnostic criteria are few and depend solely on nuclear and cytoplasmic features of cells. Unlike histology, where the cellular features as well as the pattern and the relationship of the cells with their surroundings are all taken into consideration, this technique does not offer such a

possibility. Despite these diagnostic limitations, *cytology* is a *rapid - inexpensive - non invasive* method of patient evaluation and it is rapidly gaining precedence over many diagnostic techniques because it is:

Easy: technique does not require a surgical facility and most samples can be taken anywhere, even at home.

Rapid: can take many samples in a short time

Inexpensive: requires few personnel and minimal equipment

Repeatable: Non invasive method which offers no inconvenience to the patient

Uses:

Screening: Early detection of premalignant and early invasive neoplasia e.g. Cervico-vaginal PAP smears, breast carcinoma screening programs, urine screening, and sputum screening for bronchogenic carcinoma

Initial diagnosis

Follow up and monitoring of treatment

Research and tissue culture material

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Biographical sketch

Dr. Soheir Mahfouz, born in Cairo, Egypt 1950, is a medical graduate from Cairo University. She pursued her postgraduate studies and training in Kasr El Ainy Teaching Hospital, which is the main teaching hospital for Cairo University. She was appointed in the Pathology Department, Cairo University as Pathology instructor 1976-1980, Assistant lecturer 1980-1984, Lecturer 1984-1989, Assistant professor of pathology and acting responsible for the Cytology Unit 1989-1994. She was appointed as Professor of Pathology in 1994.

In 1998 she became Head of Pathology University of 6th of October Faculty of Medicine, where she served from 1998-2001. She then became Head of Cytology in 2003 at the Pathology Department, Cairo University.

Dr Mahfouz was also Electron Microscopy Representative: EM unit at NEMROCK (Kasr El Ainy Center of Nuclear Medicine and Radiation Oncology) 1985-1993 and member in the project of earlier detection and brachytherapy management of cancer cervix uteri: 1986-1989, sponsored by the International Atomic Energy Association IAEA and World Health Organization (WHO).

Professor Mahfouz is a member of the Egyptian Association of Laboratory Medicine, Paleopathology Association and a member of the Egyptian Society of Pathologists as well as a member of both the International and Arab division of the International Academy of Pathology. At present (2006) she is a member of the Curriculum development team and on the committee for undergraduate curricula development, in addition to being the acting responsible for the Histopathology MRC Path program of the Royal college of Pathologists in Egypt.