Symptoms, Signs & Syndromes

A medical glossary

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Preface

The symptoms of a disease and the physical manifestations (or signs) of the underlying pathological process are the starting points for the diagnosis of the majority of illnesses. It is the authors' strong belief that the professions ancillary to medicine, nurses and physiotherapists in particular, are provided by their close and enduring contact with patients in the ward with an invaluable opportunity to observe the changing pattern of human disease.

With this in mind we have chosen to describe those symptoms and signs and their underlying physio-pathological causes, where these are known, which are the manifestations of the commoner diseases.

It has long been medical custom that when a number of symptoms and signs occur together a name is given to them (commonly the name of the physician who first saw the association); this association is called a syndrome.

The choice of subject matter no doubt reflects our own interests; we hope these interests will be shared by the reader.

We would like to thank Mrs P. Docherty, Mrs K. Simpson and Miss M. Dunwell without whose unremitting labour this glossary could not have been produced.

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BETTY CHAMPNEY
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How to use this glossary

The entries are arranged in alphabetical order, as in an ordinary dictionary. Each entry starts with a definition of the term, followed by an explanation, if this is known, of the particular phenomenon and a description of its characteristic features.

The reader may be unfamiliar with some of the terms used in the explanations. For ease of cross-reference, those terms printed in capital letters have their own full entry at the appropriate point in the glossary.

Abdominal pain

Abdominal pain is a symptom of many conditions. It may be acute or chronic, constant or intermittent, continuous or colicky and it may or may not be associated with readily recognizable signs of peritoneal irritation.

(1) Conditions within the abdomen giving rise to abdominal pain include:
   (a) Diseases of hollow organs, e.g. duodenal ulceration, chronic cholelithiasis, strangulation, or virus infections such as mesenteric adenitis.
   (b) Diseases involving the peritoneum including chemical or bacterial peritonitis.
   (c) Vascular conditions such as mesenteric arterial or venous thrombosis, or dissecting aneurysm.
   (d) Sudden 'distension' of the capsule of the liver as in right sided heart failure.
(2) Conditions outside the abdomen giving rise to abdominal pain:
   (a) REFERRED PAIN, originating in the heart, diaphragm or chest wall.
   (b) Metabolic causes of pain in which the mechanism by which abdominal pain is produced is not entirely clear, including diabetic KETOSIS, PORPHYRIA, URAEMIA.
   (c) Toxic causes including lead poisoning.
(3) Neurogenic pain caused by diseases of the spinal cord or nerve roots, e.g. spinal cord tumours or virus infections such as herpes.
(4) Psychogenic pain of which one of the most classical is MUNCHAUSEN'S SYNDROME.

Abdominal swelling

Generalized abdominal swelling is usually a sign of:

(1) An accumulation of abdominal fat.
(2) Gas in the intestinal tract.
(3) Fluid in the peritoneal cavity—ASCITES.
(4) Faeces in the colon.
(5) Pregnancy, giant ovarian cysts or massive fibroids.

The symptom, in contrast to the actual presence of abdominal swelling, is not uncommon in neurotic patients who will often complain of a sensation of tightness requiring the loosening of constricting clothing.

(1) Fat. Abdominal swelling due to adipose tissue is usually part of a generalized OBESITY. Rarely is it a sign of the hormonal derangement produced by CUSHING's disease or syndrome in which abnormal quantities of adrenocortical hormones are secreted. In this condition excessive fat normally accumulates on the trunk and proximal parts of the limbs.

(2) Gas accumulates in the intestinal tract in excessive quantities in the presence of mechanical or paralytic obstruction. The former is normally associated with vomiting and intestinal COLIC whereas the latter may be relatively painless despite the gross abdominal distension which occurs.

(3) ASCITES. The accumulation of excessive quantities of fluid in the peritoneal cavity which must be distinguished from the swelling produced by a large ovarian cyst or occasionally a full bladder.

(4) Faeces. An excess of faeces sufficient to cause generalized abdominal distension is rare. It may, however, occur in 2 conditions.

   (a) Hirschsprung's disease, a congenital condition in which the parasympathetic nerve supply to the rectum and possibly the colon is absent so that the bowel fails to empty. In the majority of affected babies this condition becomes evident soon after birth when the abdominal distension becomes obvious and the baby fails to pass meconium.

   (b) Idiopathic megacolon, a condition in which there is no demonstrable anatomical abnormality of the bowel. This disorder begins usually at 3 to 4 years of age and is believed to be caused by some psychological upset during potty training.

(5) The presence of a fetus. The generalized abdominal swelling caused by full-term pregnancy may be

mimicked by the presence of large fibroids of the uterus although the presence of a fetal heart beat serves to distinguish between the two conditions.

Abscess

A collection of pus in a cavity lined by granulation tissue. Clinically abscesses of bacterial origin are often described as 'hot' when caused by pyogenic bacteria and 'cold' when caused by infection with organisms such as the *Mycobacterium tuberculosis*. The former are associated with fever, the overlying skin is redder and warmer than its surroundings and the abscess is painful. A 'cold' abscess causes none of these symptoms and if the overlying skin is affected at all it is cold to the touch and often bluish in colour. Either type may 'point' on a skin or epithelial surface whereupon the 'hot' abscess discharges its contents onto the surface and is usually cured, whereas a 'cold' abscess usually forms a SINUS.

The pus of a 'hot' abscess consists of bacteria, living and dead, dead tissue cells, polymorphonuclear leucocytes, proteolytic enzymes and a variety of chemical antibacterial substances including antibodies. The pus of a 'cold' abscess is described as caseous or cheesy because it is thicker and creamy yellow in colour. It contains living and dead mycobacteria, many lymphocytes and a fluid content rich in fat.

Whilst the majority of abscesses are caused by bacterial infection, non-bacterial agents such as chemicals are occasionally responsible. This type of abscess is normally sterile but may become infected to become a typical 'hot' abscess.

Acetonuria

The presence of acetone in the urine. The commonest cause of acetonuria is a lack of the hormone insulin. The result is a decrease in the rate of breakdown of glucose which is normally used by the tissues as their main source of energy. When this occurs the body begins to use fat and protein to provide its energy supply but in the absence of sufficient insulin these compounds are incompletely broken down. The result is the production of chemical substances known as ketone bodies, one of which is acetone. Of greater physiological importance

is the production also of acetoacetic acid which when present in excessive quantities in the bloodstream causes ACIDOSIS. The presence of acetone and/or acetoacetic acid in the urine can be detected by the use of 'Acetest' tablets.

Acetone also appears in the urine in starvation when there is no disturbance of insulin secretion. This is again due to the body tissues having to use substances other than glucose to maintain the energy requirements of the body.

Achlorhydria

The absence of hydrochloric acid from the gastric juice caused by the disappearance of the acid-secreting parietal cells from the mucosal lining of the stomach. Achlorhydria occurs in pernicious anaemia even when potent stimulating agents such as histamine or pentagastrin are used. It is also common in patients suffering from cancer of the stomach, but unlike pernicious anaemia, the administration of histamine or pentagastrin still causes the production of acid.

Acholuric jaundice

A type of JAUNDICE in which the skin and tissues become yellow but there is no increase in the amount of bile pigment in the urine which therefore remains of normal colour.

The underlying cause of this type of jaundice is always an excessive destruction of erythrocytes which leads to the release into the circulation of increasing quantities of haemoglobin from which the bile pigments are formed. Should the quantity formed be greater than the liver cells can excrete, jaundice occurs. The urine, however, remains a normal colour because unconjugated bile pigment, that is pigment which has not passed through a liver cell, is insoluble in water and therefore cannot be excreted in the urine by the kidney.

Excessive erythrocyte destruction (haemolysis) occurs in:

(1) Conditions associated with alterations in shape of the erythrocyte. The classical example of this is hereditary spherocytosis in which the red cells are no longer biconcave but spherical. This alteration in shape causes these cells to be more fragile than normal and they are there-

fore broken down more easily and after a shorter time in the circulation than are normal erythrocytes.

(2) Conditions associated with alterations in the chemical composition of haemoglobin such as sickle cell anaemia in which the red cells twist and alter in shape and are destroyed when exposed to low oxygen tensions.

(3) Exposure of the erythrocytes to antibodies. If anti-erythrocyte antibodies are present in the circulation, the red cells are haemolysed, as after a mis-matched blood transfusion.

(4) Miscellaneous causes of excessive haemolysis include thermal burns in which the red cells are destroyed by heat and SEPTICAEMIA in which the red cells are destroyed by the various bacterial toxins.

In acholuric jaundice the stools and the urine remain normal in colour because there is no obstruction to the flow of bile pigment from the liver into the duodenum. When the condition is chronic as in hereditary spherocytosis, stones composed of bile pigment are finally deposited in the gall bladder. In the course of time these may later cause obstructive jaundice.

Acidosis

An increase in the acidity of the blood, the chemical reaction of which is normally kept constant by regulatory mechanisms involving the lungs and the kidneys.

The symptoms of acidosis include:

(1) DYSPNOEA due to stimulation of the respiratory centre in the brain stem by the increasing acidity of the blood. Increased depth and rate of respiration blows off carbon dioxide and, therefore, tends to lower the concentration of carbonic acid in the blood.

(2) Mental confusion and loss of consciousness occur when the condition becomes serious, hence the COMA of uncontrolled diabetes.

(3) Since many of the causes of acidosis are also associated with abnormal levels of circulating potassium, cardiac irregularities including CARDIAC ARREST are not uncommon.

The causes of acidosis are commonly divided into two:

(1) Respiratory acidosis which occurs in patients suffering from severe pulmonary disease such as chronic bronchitis in which carbon dioxide cannot be eliminated from the lungs. The gas remaining in the circulation forms carbonic acid.

(2) Metabolic acidosis which may be caused by:
   (a) The excessive production of acidic materials in the body as in the condition of diabetic KETOSIS.
   (b) Severe renal damage in which the kidneys are unable to excrete the acidic materials formed during normal metabolic processes. It is this latter type of acidosis which is complicated by a rise in the concentration of potassium in the blood to levels which are incompatible with normal cardiac function.
   (c) An excessive loss of alkaline substances from the body as a result of severe DIARRHOEA or from small intestinal FISTULAE.

Acromegaly

A sign of over-secretion of growth hormone by the anterior lobe of the hypophysis cerebri after fusion of the epiphyses has occurred. Compare this to GIGANTISM. An excess of this hormone results in generalized hyperplasia of the tissues with the result that the bones of the extremities, jaw and face enlarge, producing the characteristic features of the acromegalic which include prominent ridges above the eyes, a protruding lower jaw and enlargement of the hands. The disease is associated with osteoporosis which results in loss of strength in the bones so that the vertebral bodies collapse and a KYPHOSIS develops. Loss of libido and diabetes mellitus are also common.

Agranulocytosis

The disappearance of the polymorphonuclear white blood cells from the circulation, often accompanied by a pancytopenia in which all the cellular elements of the blood, i.e. erythrocytes and platelets also disappear. The condition should be suspected when a patient suddenly develops a reduced resistance

to infection. This occurs because the disappearance of the granulocytes removes one of the main defences of the body against bacterial invasion. Commonly severe fever and ulceration of the gums, oral mucosa and throat occur, later followed by fungal infection.

The diagnosis is easily made by full blood count, differential white count and an examination of the bone marrow.

Among the many causes of agranulocytosis are the toxic effects of many therapeutic agents including:

(1) Phenylbutazone, used in the treatment of arthritis.
(2) Thiouracil, used in the treatment of thyrotoxicosis.
(3) Chloramphenicol, used in the treatment of bacterial infections.
With these drugs there is seldom a relationship between the amount of the drug which has been received and the occurence of agranulocytosis.
(4) The alkylating agents such as thiotepa used in the treatment of malignant disease. The incidence and severity of the agranulocytosis following the administration of these drugs is related to the dose.

Albinism

Albinism is a sign of an inherited enzyme deficiency. Although pigment cells are present in normal numbers, the pigments which normally colour the skin and eyes cannot be produced due to absence of one specific enzyme known as tyrosinase. The result is an individual with a pale skin, white hair and pink eyes. Albinos are rare, occurring 1 in 10 to 20 000 births.

Albright's syndrome

A syndrome, the cause of which is unknown, associated with marked deformity of the skeleton, precocious puberty in girls and skin pigmentation. During the growing period prior to closure of the epiphyses, bone is replaced by dense fibrous tissue which is a poor substitute. The bones in this condition are then incapable of supporting the body and the weight-bearing bones bend leading to severe bowing of the lower limbs.

Albuminuria see PROTEINURIA

Aldrich's syndrome

A hereditary disorder transmitted by a sex-linked gene which affects only boys in whom PURPURA, THROMBOCYTOPENIA, eczema and recurrent skin infections occur. The possible cause is a failure of IMMUNITY to develop.

Alkalosis

A condition in which the blood is more alkaline than normal—that is the pH is raised above the normal value of 7.4.

Alkalosis is either metabolic or respiratory:

(1) Metabolic alkalosis occurs when there is an excess of alkali in the blood brought about by:
   (a) The ingestion of excessive quantities of alkali, usually the result of the treatment of duodenal ULCERATION. In this case the patient may develop NAUSEA, VOMITING and ANOREXIA.
   (b) The loss of acid from the body by excessive vomiting or gastric aspiration as the gastric juice contains hydrochloric acid. Alkalosis due to this cause is nearly always associated with the signs of DEHYDRATION and TETANY, with the development of a positive CHVOSTEK'S SIGN.

(2) Respiratory alkalosis is caused by any condition which produces a persistent increase in the depth and rate of respiration (hyperventilation). This leads to the excessive extraction of carbon dioxide from the blood in the lungs which produces a fall in the level of carbonic acid in the blood. Respiratory alkalosis is common in hysteria and at high altitudes when respiration is stimulated by the lack of oxygen.

The principal result of alkalosis of respiratory orgin is to precipitate the onset of TETANY, even though the serum calcium level remains normal.

Alkaptonuria

The presence in the urine of homogentisic acid. If such urine is allowed to stand it turns brown or black in colour. The presence of such a pigment is due to an error in protein metabol-

ism. The condition is rare and is inherited through a recessive gene. Normally there is no evidence of the condition until middle age when about this time a breakdown product of homogentisic acid begins to be deposited in the sclerotics of the eye. Deposition of the same pigment in cartilage, tendons and ligaments leads to darkening of the cheeks and nose and ARTHRITIS.

Allergy

A sign of an altered reaction by the tissues to antigens which, in this particular case, are known as allergens.

There may be a subtle difference between ALLERGY and ANAPHYLAXIS (hypersensitivity) since the latter implies an excessive reaction to an antigen, but most authorities use the two terms synonymously. Two types of allergy are recognized, the distinction being made according to the speed with which the reaction begins. In the first type, the reaction occurs almost immediately, whereas in the second it is delayed for 24 to 48 hours.

The immediate type of reaction is due to interaction between the allergen and antibodies already circulating in the blood stream, whereas in the delayed type the reaction is the result of a reaction between the allergen and the cells of the tissue.

Colloquially, the term allergy is applied to the reaction which follows the inhalation of pollen or dust, the eating of certain foodstuffs such as shellfish or strawberries, contact with plants such as the primula, or materials such as furs, and the response to the administration of certain drugs, for example, penicillin. The reaction varies—when the allergen is inhaled, rhinitis, hay fever or asthma occur; skin contact produces URTICARIA and the injection of an allergen may produce a generalized anaphylactic reaction resulting in death.

Amblyopia

Blurring of vision. In childhood amblyopia is usually caused by difficulty in fusing the separate visual images which pass to the brain from the eyes. To overcome this difficulty, the image from one eye is suppressed in order to prevent DIPLOPIA (double vision), but as a result visual perception is reduced.

Later, because of the disuse following suppression, the visual pathway deteriorates. Amblyopia of one eye may also occur if there is a high refractive error.

In an adult, amblyopia may be caused by a number of conditions affecting the cells of the retina or the optic nerve. These include disseminated sclerosis, which damages the nerve and poisons such as quinine, methylated spirits and tobacco, which damage the retina. Amblyopia may also be a symptom of HYSTERIA.

Amenorrhoea

Absence of menstruation is divided into two types.

(1) False amenorrhoea in which menstruation is taking place, but the outflow is obstructed, for example by an imperforate hymen.

(2) True amenorrhoea which may be divided into physiological and pathological varieties:

  (a) The physiological types occur:

    (i) Prior to puberty and during adolescence due to the absence of hormonal stimulation.

    (ii) In pregnancy, amenorrhoea occurs due to the continuous production of large quantities of the hormones oestrogen and progesterone by the chorion.

    (iii) During lactation, when amenorrhoea results from lack of formation of the hormones known as gonadotrophins.

    (iv) During the menopause, when the ovaries cease to react to the gonadotrophic stimulus.

  (b) Pathological types. Since the maintenance of the menstrual cycle depends on the proper functioning of the hypothalamus, the anterior lobe of the pituitary gland, the ovary and the uterus, any disturbance of this chain may result in amenorrhoea.

Amenorrhoea is, therefore, observed in FROHLICH'S SYNDROME, in conditions causing injury or disease of the midbrain such as encephalitis or meningitis, and in cerebral conditions such as depressive illnesses. Disease of the pituitary, usually in the