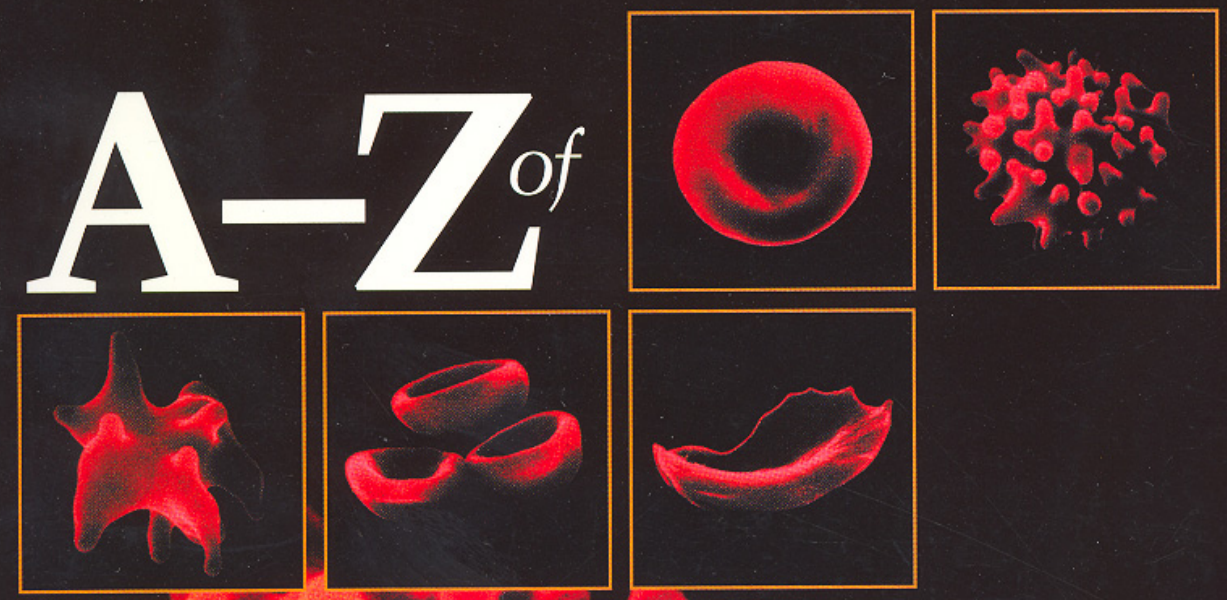
Barbara J. Bain
& Rajeev Gupta

A–Z of Haematology



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A–Z
of Haematology

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Preface

In this A–Z of Haematology we have sought to be as comprehensive as possible, but we have nevertheless given particular emphasis to recent advances in molecular haematology. We have detailed the important genes that have been implicated in haematological neoplasms and in constitutional haematological disorders. Blood transfusion, haemostasis and thrombosis and immunology have not been neglected. We have provided the reader with a complete list of the molecules that have been assigned a Cluster of Designation (CD) number, with descriptions of their functions and patterns of expression in health and disease. Because of the emphasis we have given to the scientific basis of haematology and related disciplines we believe that this work will be useful not only to haematologists but also to research scientists and to biomedical scientists working in diagnostic laboratories. Those working in cancer cytogenetics and immunophenotyping will also find it a valuable repository of relevant knowledge. The very existence of such a book is indicative of the fact that a book still remains a highly convenient reference source. However, for those who wish to seek further information electronically we have provided a list of some of the more useful of the many websites available.

It will be helpful to the reader to know some of the conventions we have followed. All human genes are designated as recommended by the human genome project, in upper case italics with Greek letters being replaced by their Roman equivalent. Approved names are given but where a gene is better known to haematologists by another name, we have mainly used that name in further discussion. We have indicated how gene names (and some protein names) are derived from a longer descriptive phrase by means of bold print plus underlining of the relevant letters, e.g. *PLZF*—**P**romyelocytic **L**eukaemia **Z**inc **F**inger. However, **bold print without underlining** is used for another purpose, to indicate that there is a relevant entry in the book. In order to avoid tedium, words and phrases that are used very frequently, e.g. 'acute myeloid leukaemia' are not generally cross referenced in this manner.

We wish to thank those who have helped with the provision of illustrations: the publisher of the late Professor M. Bessis, Professor D. Catovsky, Dr W. Gedroyc, Miss C. Hughes, Mr R. Morilla, Ms L. Phelan, Ms Julia Pickard and the Cytogenetics Department at Hammersmith Hospital, Professor A. Polliack, Professor Lorna Secker-Walker, The North Trent Cytogenetics Service at Sheffield Childrens Hospital, the Kennedy Galton Institute and the United Kingdom Cancer Cytogenetics Group.

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Online Resources

General haematology

American Society of Hematology **www.hematology.org**

British Society for Haematology **www.blacksci.co.uk/uk/society/bsh**
(use this site to access PubMed, Centers of Disease Control and Institute of Biomedical Science)

European Hematology Association **www.ehaweb.org**

British Committee for Standards in Haematology guidelines **www.bcshguidelines.com/**
(use this site to access *Cells of the Blood, Haematological Malignancy Diagnostic Service* and *Hematology Digital Image Bank*)

Haematologists in Training **www.hit.gb.com/**
(use this site to access *MRC Leukaemia Trials* and an on line medical dictionary through *doctors' guide to internet* and *Guide to Internet Resources on Haematological Malignancies*)

Other general haematology **www.bloodline.net**

Chromosomes, genes and proteins—molecular haematology

Cytogenetics in haematology

Genetics and cytogenetics in Haematology **www.infobiogen.fr/services/chromcancer/**

Online Mendelian Inheritance in Man **www.ncbi.nlm.nih.gov/omim/**

Cardiff Human Gene Mutation Data Base **www.uwcm.ac.uk/uwcm/mg/hgmd0.html**

Sources of probes for molecular genetic studies: Vysis **www.vysis.com/hematology**
and Q-Biogene (previously Oncor) **www.cambio.co.uk/starfish/**

Human proteins website **www.ncbi.nlm.nih.gov/prow**

Websites of antibody manufacturers

http://serotec.oxi.net/asp/index.html

www.bdbiosciences.com

www.vectorlabs.com

Realtime PCR **www.cgr.otago.ac.nz/SLIDES/7700/SLD001.HTM**

Chemokine review **http://www.path.sunysb.edu/courses/syllabus/chemkin.htm**

Cytokine minireviews **http://www.rndsystems.com/asp/g_sitebuilder.asp?BodyId=2**

Haemoglobinopathies and thalassaemias

http://globin.cse.psu.edu

Thrombosis and haemostasis

The International Society on Thrombosis and Haemostasis **www.med.unc.edu/isth/welcome**

The World Federation of Hemophilia **www.wfh.org**

Blood transfusion

American Association of Blood Banks **www.aabb.org**

British Blood Transfusion Society **www.bbts.org.uk**
(use this site to access British blood transfusion guidelines)

National Blood Service **www.blood.co.uk**

Serious Hazards of Transfusion **http://www.shot.demon.co.uk**

Malaria

http://www.rph.wa.gov.au/labs/haem/malaria/

Haematological neoplasms

General **http://cancerweb.ncl.ac.uk/cancernet.html**
(use this site to access an online medical dictionary)

http://www.cancerindex.org/clinks2.htm

The British National Lymphoma Investigation **www.bnli.ucl.ac.uk/**

Lymphoma Forum **www.lymphoma.org.uk/lymphoma.htm**

The Leukaemia Research Fund **www.dspace.dial.pipex.com/lrf-/**

The UK Myeloma Forum **www.ukmf.org.uk**

American Association for Cancer Research **www.aacr.org**

(use this site for access to the five journals published by the AACR)

Abstracts and journals

Entrez PubMed **www.ncbi.nlm.nih.gov/**

Blood **www.bloodjournal.org/**

Haematologica **www.haematologica.it/main.html**

Online flow cytometry cases **www.flowcases.org**

British Medical Journal **www.bmj.com**

Teaching sites

www.hematology.org (click on educational materials)

www.haem.net

http://pathy.med.nagoya-u.ac.jp/atlas/doc/atlas.html

www-medlib.med.utah.edu/WebPath/webpath.html

A

α alpha, the first letter of the Greek alphabet, often used to designate polypeptide chains

α_1 antitrypsin a **serpin** which inactivates **neutrophil elastase**; mutation of the gene encoding α_1 antitrypsin can lead to production of a protein that inhibits coagulation pathway proteases and leads to a bleeding disorder

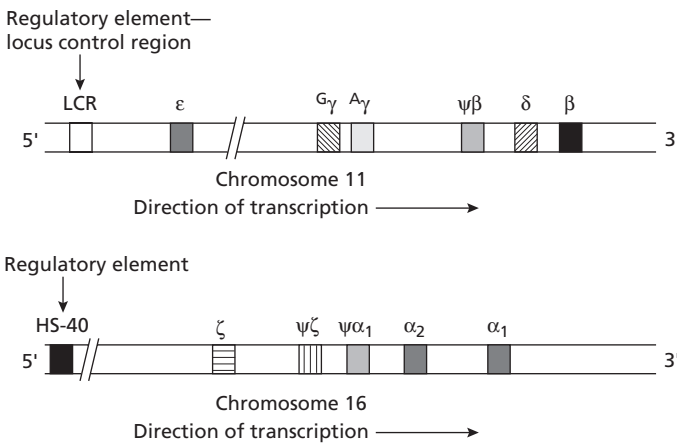
α chain (i) the alpha globin chain which is essential for formation of haemoglobins A, A_2 and F (ii) the heavy chain of immunoglobulin A; two alpha chains combine with two light chains (in a single molecule either kappa or lambda) to form a complete immunoglobulin molecule (iii) part of the αβ T-cell receptor, a surface membrane structure in T lymphocytes which permits antigen recognition

α error a statistically significant difference when no real difference exists; e.g. if the results of two treatment strategies are statistically different with a probability of $P = 0.05$ there is a 1 in 20 chance that there is no real difference

α globin cluster the cluster of genes on chromosome 16 that includes the genes encoding ζ, α2 and α1 chains (Fig. 1)

α globin gene the *HBA* genes, gene map locus 16p13.3, encoding the **α globin chain** of **haemoglobin**; there are two α globin genes, designated α2 and α1, on each chromosome 16

Figure 1 α and β globin gene clusters.
The alpha and beta globin gene clusters on chromosomes 11 and 16 respectively. The β cluster has an upstream locus control region (LCR) and ε, $^{G}\gamma$, $^{A}\gamma$, δ and β genes; there is one pseudogene, ψβ. The α cluster has an upstream H40 regulatory region and ζ, α_2 and α_1 globin chain genes; there are two pseudogenes, ψζ and ψα.



α heavy chain disease a **plasma cell dyscrasia** characterized by secretion of the heavy chain of **immunoglobulin A**

α naphthyl acetate esterase (ANAE) an enzyme belonging to the **non-specific esterase** group of enzymes, strongly expressed in cells of the monocytic and megakaryocytic lineages

α naphthyl butyrate esterase (ANBE) an enzyme belonging to the **non-specific esterase** group of enzymes, strongly expressed in cells of the monocytic lineage

α satellite DNA repeat sequences at the **centromere** of a **chromosome**; the sequences differ between chromosomes, permitting the development of **centromeric probes** that identify different chromosomes

α thalassaemia a group of thalassaemias characterized by deletion or, less often, altered structure and reduced function of one or more of the α globin genes (*see also* **α thalassaemia trait**, **haemoglobin H disease** and **haemoglobin Bart's hydrops fetalis**) (Fig. 2)

α thalassaemia trait a minor haematological abnormality resulting from deletion of one or two of the four α globin genes; includes **heterozygosity** and **homozygosity** for **α^+ thalassaemia**, when one of two α genes on a chromosome is deleted, and heterozygosity for **α^0 thalassaemia**, when both α genes on a single chromosome are deleted (*see* Fig. 2)

A an abbreviation for the purine, adenine

ABC7 a gene, gene map locus Xq13, encoding **A**TP **B**inding **C**assette transporter **7**, a **mitochondrial** protein, mutation of which can cause **sideroblastic anaemia** with spino-cerebellar ataxia

aberrant diverging from normal, e.g. expression of an antigen which is inappropriate for a lineage

abetalipoproteinaemia inherited absence of beta lipoproteins, associated with **acanthocytosis**

ABI1 a gene, **Ab**l-**I**nteractor **1**, gene map locus 10p11.2, which contributes to the *MLL-ABI1* fusion gene in M4 acute myeloid leukaemia associated with t(10;11)(p11.2;q23); *ABI1* encodes spectrin SH3 domain-binding protein 1, which is a widely expressed component of a multi-protein complex that negatively regulates cellular responses to various mitogenic signals

ABL a gene, Abelson murine leukaemia viral oncogene homologue 1, gene map locus 9q34; cellular homologue of *v-abl*, a gene in the **Abe**l**s**on murine leukaemia retrovirus which is involved in some murine leukaemias; encodes a non-receptor tyrosine kinase; *ABL* contributes to:

- the *BCR-ABL* fusion gene in t(9;22)(q34;q11) associated with chronic granulocytic leukaemia and with Philadelphia-positive acute lymphoblastic and acute myeloid leukaemias
- the *ETV6-ABL* fusion gene in chronic myeloid leukaemias, acute myeloid leukaemia and acute lymphoblastic leukaemia associated with t(9;12)(q34;p13) and variant translocations

Both *BCR-ABL* and *ETV6-ABL* are inhibited by the ABL tyrosine kinase inhibitor, imatinib mesylate (STI571)

ABL is amplified by segmental jumping translocations in some patients with therapy-related acute myeloid leukaemia

abnormal localization of immature precursors (ALIP) location of **myeloblasts** and **promyelocytes** in the centre of the intertrabecular space rather than adjacent to trabeculae or surrounding arterioles

ABO blood group system a blood group system in which A and B alleles at the ABO locus at 9q34 encode specific glycosyltransferases that modify a precursor disaccharide (Fig. 3 and Table 1, p. 4); this precursor is part of a glycoprotein or glycolipid which, when unmodified, expresses the H antigen; the O allele does not encode a functional transferase so that homozygosity for O means H is expressed but not A or B; ABO antigens are expressed on all blood cells and many other body cells (see also **Bombay blood group**); ABO chimaerism can result from constitutional mosaic trisomy 9

abortion spontaneous or induced termination of pregnancy before the fetus is viable, e.g. before 28 weeks

Figure 2 α thalassaemias.
The terminology applied to the alpha thalassaemias; most of the alpha thalassaemias result from deletion of one or both alpha genes at a locus and in some cases the zeta gene is also deleted; α^+ thalassaemia indicates that there is one remaining alpha gene at the locus whereas α^0 thalassaemia indicates that both genes at a locus are deleted; in the case of $-\alpha^{3.7}$ the remaining gene at the locus is an α2α1 fusion gene; non-deletional thalassaemia refers to the less common alpha thalassaemias resulting from mutation rather than deletion of an alpha gene, the gene being designated α^T, e.g. α^{Tsaudi}.

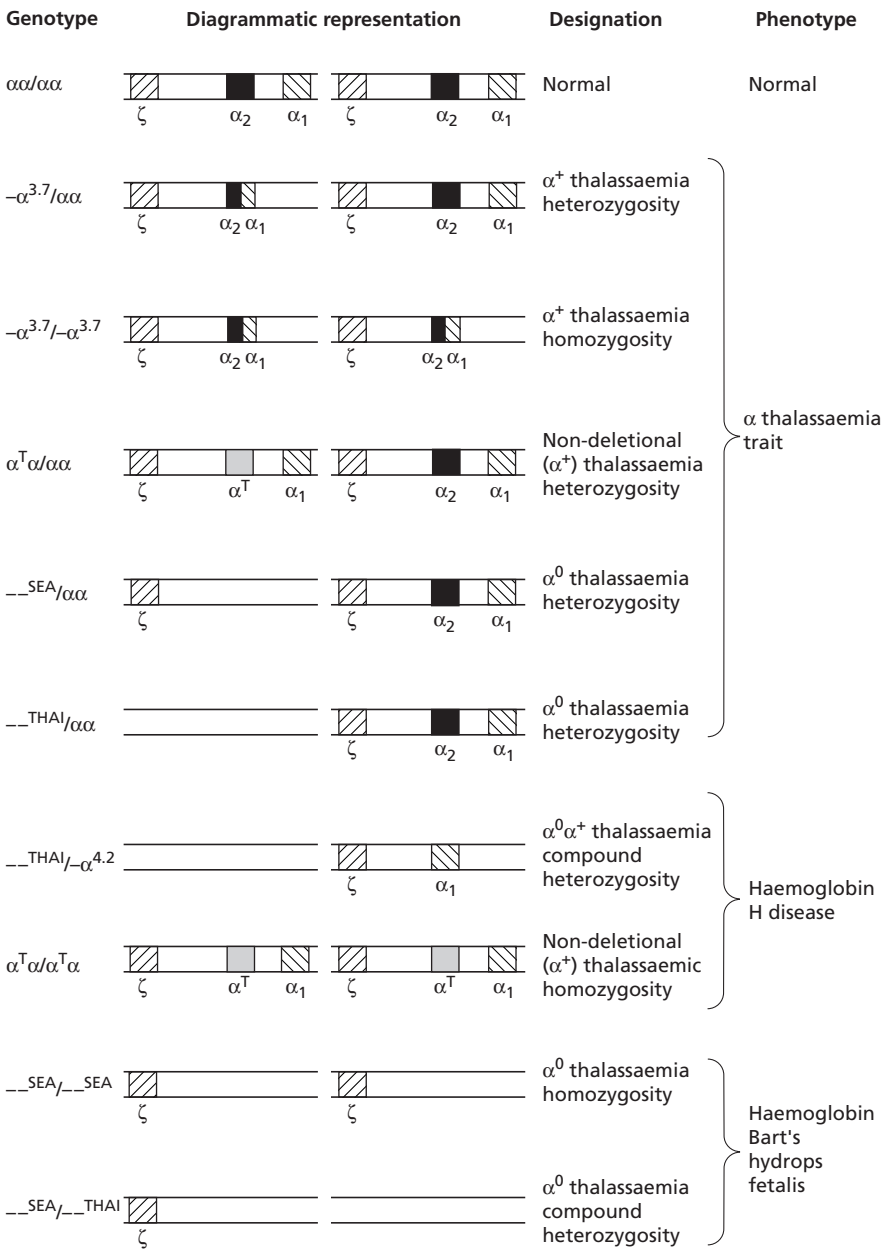
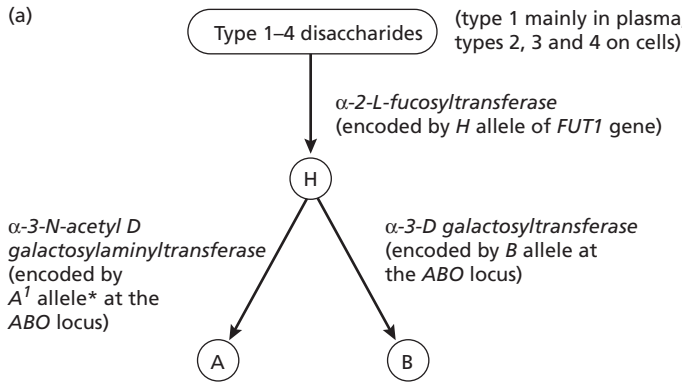


Figure 3 ABO antigens.
The formation of ABO antigens: (a) formation of H antigen and formation of A and B antigens from H; (b) the loci, the alleles and the transferases involved in the formation of ABO antigens. * The A^2 allele encodes a less efficient transferase that does not utilize types 3 and 4 disaccharide; A^3 and A^x also encode less efficient transferases.

(a)



(b)

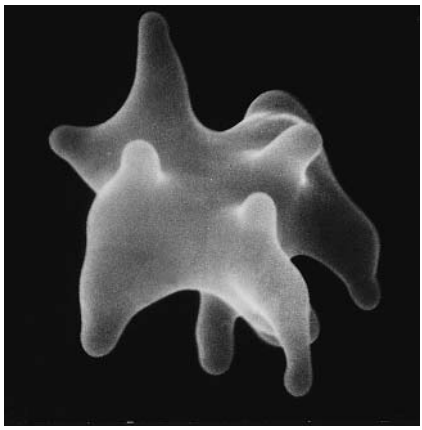
Locus	Allele	Transferase
FUT1	*H*	α-2-L-fucosyltransferase
	h	nil
ABO	*A*	α-3-N-acetyl-D-galactosaminyltransferase
	B	α-3-D galactosyltransferase
	O	nil

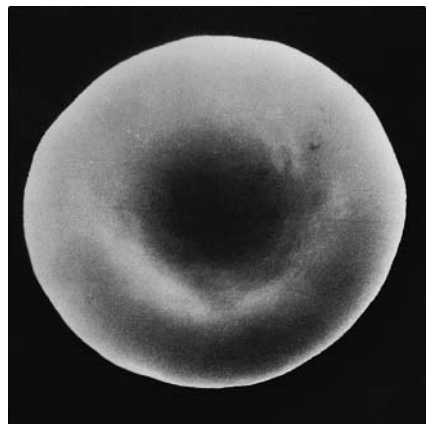
Table 1 Genotypes and resultant phenotypes of the ABO blood group system; the antibodies usually present in individuals of different ABO groups are also shown.

Alleles* at ABO locus	Antigens expressed	Antibodies
AO or AA	A	anti-B
BO or BB	B	anti-A
AB	A + B	nil
OO	nil	anti-A + anti-B

* The A allele may be either A^1 or A^2; A^2 and rare alleles of A encode a less efficient transferase.

Figure 4 An acanthocyte and a discocyte.
Scanning electron micrographs of an acanthocyte and a normal shaped red cell, a discocyte.





absorbance the degree of absorption of light

acaeruloplasminaemia an inherited, autosomal recessive condition, resulting from mutation in the caeruloplasmin gene on chromosome 3q, and consequent deficiency of caeruloplasmin ferroxidase; there is iron overload with low serum iron, normal **transferrin** concentration and moderately elevated **serum ferritin**

acanthocyte an erythrocyte covered with a small number of spicules of variable length, thickness and shape (Fig. 4)

acanthocytosis the presence of **acanthocytes**

accelerated phase a term used to describe a more aggressive phase of **chronic granulocytic leukaemia**

accuracy the closeness of a measured value to the true value

acentric having no **centromere**; acentric chromosomes cannot become attached to the **mitotic spindle** and consequently may not be present in either daughter nucleus

ACHE a gene, gene map locus 7q22, alleles of which encode the Yt^a and Yt^b antigens of the Cartwright blood group system, these antigens being expressed on **GPI**-linked **A**cetyl**ch**oline**e**sterase

achlorhydria absence of gastric acid secretion, a feature of **pernicious anaemia**

acid (i) a hydrogen-containing substance that yields a free hydrogen ion and a cation on dissociation (ii) having a low pH

acidified serum test *see* **acid lysis test**

acid-fast bacillus (AFB) a micro-organism, usually a bacillus of the genus Mycobacteria, um, which, when stained with a Ziehl–Neelsen stain, retains its colour when exposed to acid

acid lysis test a test for **paroxysmal nocturnal haemoglobinuria** and type II **congenital dyserythropoietic anaemia** (Fig. 5)

acidophilic having an affinity for acid dyes such as eosin

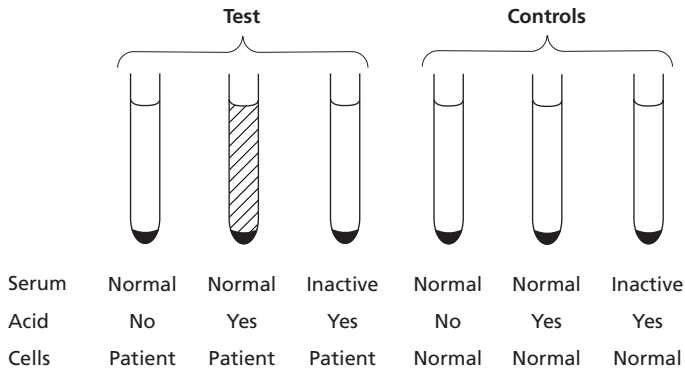
acidosis having a blood pH less than 7.35

acid phosphatase this is a generic term for an enzyme that works optimally at acid pH to release phosphate groups from complex molecules, e.g. from the serine, threonine and tyrosine residues of proteins; they are usually fairly target specific; many lymphoid and myeloid cells have acid phosphatase activity that is demonstrable cytochemically (*see also* **alkaline phosphatase**)

aCML **atypical chronic myeloid leukaemia**

acquired not present at birth; the term generally implies a condition or characteristic that is not inherited

Figure 5 The acid lysis test.
The principle of the acid lysis test (Ham test) is that some of the patient's cells lyse when exposed to acidified fresh normal serum (containing complement), conveying a pink or red colour to the supernatant of the centrifuged test sample; lysis does not occur when the serum has not been acidified or when complement in the serum has been inactivated by prior heating. Normal red cells, which are not susceptible to complement-induced lysis in acidified serum, do not lyse in any of these circumstances.



acquired angio-oedema **angio-oedema** which is not inherited or present at birth, usually caused by an acquired deficiency of **C1 inhibitor**; it can be an autoimmune condition or consequent on a low grade B-cell neoplasm

acquired immune deficiency syndrome (AIDS) an acquired cell-mediated immune deficiency syndrome, consequent on marked reduction of CD4+ T lymphocytes resulting from **HIV** infection

acquired immunity adaptive immunity that is altered by exposure to antigens, dependent on antigen-presenting cells, T lymphocytes and B lymphocytes

acquired Pelger–Huët anomaly acquired hypolobulation of neutrophils or other granulocytes, usually indicative of **myelodysplastic syndrome** or **acute myeloid leukaemia**; the cytological features resemble those of the inherited **Pelger–Huët anomaly**

acrocentric having the **centromere** near one end

ACS2 a gene, Acyl-CoA Synthetase 2, encoding acyl-CoA synthetase 2, gene map locus 5q31 which contributes to an *ETV6-ACS2* fusion gene in myelodysplastic syndrome and acute myeloid leukaemia associated with t(5;12)(q31;p13)

actins an evolutionarily conserved family of intracellular proteins, whose genes exist in multiple copies in all species studied; actin molecules polymerize into intracellular microfilaments that are involved in muscle contraction, cell motility and organelle transport; immunohistochemical demonstration of actin is useful in the diagnosis of rhabdomyosarcoma

activated partial thromboplastin time (aPTT) a coagulation test in which a contact activator, a **partial thromboplastin** and calcium are added to plasma with the clotting time then being recorded; a test of the **intrinsic pathway** of coagulation (*see* Fig. 17, p. 77)

activated protein C resistance resistance to the anticoagulant effect of activated **protein C**, often caused by inheritance of a variant of factor V, **factor V Leiden** (*see* **naturally occurring anticoagulants**)

actuarial survival an estimate of **median survival** made while many patients are still alive

Table 2 WHO criteria for the diagnosis of biphenotypic leukaemia.

Score	B lineage	T lineage	Myeloid
2	cCD79a cIgM cCD22	CD3 (c or Sm) anti-TCR ($\alpha\beta$ or $\gamma\delta$)	MPO
1	CD19 CD10 CD20	CD2 CD5 CD8 CD10	CD117 CD13 CD33 CD65
0.5	TdT CD24	TdT CD7 CD1a	CD14 CD15 CD64

If > 2 points is scored for both myeloid and one of the lymphoid lineages the case is classified as biphenotypic; in the original EGIL recommendations CD117 scored 0.5 rather than 1

c, cytoplasmic; CD, cluster of differentiation; Ig, immunoglobulin; MPO, myeloperoxidase; Sm, surface membrane; TdT, terminal deoxynucleotidyl transferase.

Table 3 A simplified explanation of the French-American-British (FAB) classification of acute myeloid leukaemia (AML).

FAB designation	Description
M0	AML with minimal evidence of myeloid differentiation
M1	AML with granulocytic differentiation but little maturation
M2	AML with granulocytic differentiation and maturation
M3 and M3 variant	Acute hypergranular promyelocytic leukaemia and the hypogranular or microgranular variant form
M4	AML with both granulocytic and monocytic differentiation
M5	AML with monocytic differentiation, either without maturation (M5a or acute monoblastic leukaemia) or with maturation (M5b or acute monocytic leukaemia)
M6	AML with at least half the bone marrow cells being erythroblasts
M7	Acute megakaryoblastic leukaemia

acute basophilic leukaemia an **acute myeloid leukaemia** with prominent basophilic differentiation

acute biphenotypic leukaemia an acute leukaemia in which there is both myeloid and lymphoid differentiation, defined in **the WHO classification** as shown in Table 2

acute eosinophilic leukaemia an **acute myeloid leukaemia** with prominent eosinophilic differentiation

acute erythroleukaemia an **acute myeloid leukaemia** with prominent erythroid differentiation; the **FAB** M6 category of AML (Table 3, *see also* Table 4)

acute hypergranular promyelocytic leukaemia an **acute myeloid leukaemia** characterized by leukaemic cells that are abnormal hypergranular promyelocytes, the FAB M3 category of AML (*see* Tables 3 and 4)

acute leukaemia a leukaemia that, if untreated, leads to death in weeks or months; a leukaemia characterized by continued proliferation with a failure of

Table 4 The WHO classification of acute myeloid leukaemia (AML).

Acute myeloid leukaemia with recurrent genetic abnormalities*
AML with t(8; 21)(q22;q22)/*AML1-ETO* fusion
AML with abnormal bone marrow eosinophils and inv(16)(p13q22) or t(16; 15)(p13;q22)/*CBFB-MYH11* fusion
Acute promyelocytic leukaemia with t(15; 17)(q22;q12)/*PML-RARA* fusion, and variants
AML with 11q23 rearrangement and *MLL* abnormality
AML with multilineage dysplasia†
Following a myelodysplastic syndrome or a myelodysplastic/myeloproliferative syndrome
Without antecedent myelodysplastic syndrome
Therapy-related AML and myelodysplastic syndrome
Alkylating agent-related
Topoisomerase II-inhibitor-related
Other types
AML not otherwise categorized
AML, minimally differentiated (resembles FAB M0)
AML without maturation (resembles FAB M1)
AML with maturation (resembles FAB M2)
Acute myelomonocytic leukaemia (resembles FAB M4)
Acute monoblastic and acute monocytic leukaemia (resembles FAB M5a, M5b)
Acute erythroid leukaemia
Erythroleukaemia (resembles FAB M6)
Pure erythroid leukaemia
Acute megakaryoblastic leukaemia (resembles FAB M7)
Acute basophilic leukaemia
Acute panmyelosis with myelofibrosis
Myeloid sarcoma (granulocytic or monocytic)

* Therapy-related cases may be noted to have one of these abnormalities but are assigned to the category of therapy-related AML.
† Defined as having at least 50% of cells dysplastic in at least 2 lineages.

differentiation so that the dominant cell is a primitive cell referred to as a **blast cell**

acute lymphoblastic leukaemia (ALL) an acute leukaemia in which the predominant cell is a **lymphoblast** of T or B lineage

acute monoblastic leukaemia an **acute myeloid leukaemia** in which the dominant cell is a **monoblast**, the FAB M5a category of AML (*see* Tables 3 and 4)

acute monocytic leukaemia an **acute myeloid leukaemia** in which the leukaemic cells are mainly **promonocytes** and **monocytes** but with at least 20 or 30% of peripheral blood or bone marrow cells being **blast cells**; the FAB M5b category of AML (*see* Tables 3 and 4)

acute myelofibrosis **acute myeloid leukaemia** with reactive bone marrow **fibrosis**; the leukaemic cells are commonly but not always of megakaryocyte lineage

acute myeloid leukaemia (AML) an acute leukaemia in which leukaemic cells belong to any myeloid lineage, i.e. granulocytic, monocytic, erythroid or megakaryocyte lineages, defined in the **WHO classification** as a haematological neoplasm with at least 20% blast cells in the peripheral blood or bone marrow or with a lower percentage of blast cells if there is one of three specified chromosomal rearrangements—t(8;21), inv(16) or t(16;16); AML is further classified as shown in Table 4

acute myelomonocytic leukaemia (AMML) an **acute myeloid leukaemia** in which there is both granulocytic and monocytic differentiation, the FAB M4 category of AML (*see* Tables 3 and 4)

acute non-lymphoblastic leukaemia (ANLL) an alternative designation of **acute myeloid leukaemia**, mainly used in the USA

acute phase reactant one of a number of plasma proteins that rise in concentration in response to acute inflammation or tissue injury, enhancing resistance to infection and promoting tissue repair

acute phase reaction an acute systemic response to infection or inflammation, in which there is a fall in serum albumin, transferrin and iron and a rise in other proteins including C-reactive protein, serum amyloid A protein, factor VIII, fibrinogen and α2 macroglobulin

ADA the gene on 20q that encodes adenosine deaminase, deficiency of which can cause severe combined immunodeficiency

ADAMTS13 a gene at 9q34, encoding a disintegrin-like metalloprotease with thrombospondin type 1 motif, 13, also known as **von Willebrand's factor-cleaving protease**, mutation of which can cause familial **thrombotic thrombocytopenic purpura**

ADCC **antibody-dependent cellular cytotoxicity**

add a cytogenetic abbreviation indicating additional material of unknown origin

Addisonian anaemia *see* **pernicious anaemia**

Addison's disease an illness resulting from chronic failure of the adrenal glands

addresin a tissue-specific **adhesion molecule**

adenine a **purine** base that is a component of both DNA and RNA, pairs with **thymine**

adenocarcinoma carcinoma showing some features of differentiation to glandular structures

adenoma a benign tumour of glandular tissue

adenosine deaminase an enzyme that catalyses the conversion of adenosine to inosine; a deficiency can cause **severe combined immunodeficiency**

adenosine diphosphate (ADP) the nucleotide adenosine, with two attached phosphate moieties; a store of energy; a platelet **agonist**

adenosine triphosphate (ATP) the nucleotide adenosine, with three attached phosphate moieties; an important store of energy

adhesion the process of becoming closely attached to something else

adhesion molecule a molecule that promotes adhesion of cells to each other

adjuvant a substance that non-specifically enhances antigen-specific immune responses

ADP **adenosine diphosphate**

adrenal gland an endocrine gland sited at the upper pole of the kidney

adrenaline *see* **epinephrine**

Adriamycin a trade name for doxorubicin, an anthracycline used in the treatment of lymphomas and carcinomas

adult haemoglobin **haemoglobin A**

adult T-cell leukaemia/lymphoma (ATLL) a subacute neoplasm of T lymphocytes which may have either a leukaemic or a lymphomatous presentation; preceding infection by the **human T-cell lymphotropic virus I** (HTLV-I) is an essential aetiological factor

AE1 a gene, gene map locus 17q21-q22, encoding **S**o**l**ute **C**arrier family **4, A**nion exchanger, member **1** (*SLC4A1*) also known as the **A**nion **E**xchanger **1** and band 3 protein, a component of the red cell membrane (*see* Fig. 64, p. 199); mutation can result in **hereditary spherocytosis, hereditary elliptocytosis** or **Southeast Asian ovalocytosis**; band 3 carries the diego blood group antigens

AF1p a gene, ***A**LL1* **F**usion gene from chromosome **1p**, also known as *EPS15*—**Ep**idermal growth factor receptor pathway **S**ubstrate-**15**, gene map locus 1p32; *AF1p* encodes a phosphorylated protein that is a constitutive component of clathrin-coated pits and is required for clathrin-dependent endocytosis; it contributes to the *MLL-AF1p* fusion gene in some cases of M5 acute myeloid leukaemia

AF1q a gene, ***A**LL1* **F**usion gene from chromosome **1q**, gene map locus 1q21, which contributes to the *MLL-AF1q* fusion gene in some cases of M4 acute myeloid leukaemia; *AF1q* encodes a nuclear protein, the expression of which is usually restricted to the thymus