



 **ABBREVIATIONS**

**A**

AA	aplastic anaemia
AAG	autoimmune atrophic gastritis
AAS	atomic absorption spectrometry
ACD	anaemia of chronic disease
ACS	acute chest syndrome
ADA	adenosine deaminase
ADP	ALA dehydratase deficiency porphyria
AHP	acute hepatic porphyrias
AIHA	autoimmune haemolytic anaemia
AIP	acute intermittent porphyria
AK	adenylate kinase
ALA	delta aminolaevulinic acid
ALA-S	delta aminolaevulinic acid synthase
eALA-S	erythroid delta aminolaevulinic acid synthase
ALC	absolute lymphocyte count
ALL	acute lymphoblastic leukaemia
AML	acute myeloblastic leukaemia
ANA	antinuclear antibody
ANC	absolute neutrophil count
ARC	absolute reticulocyte count
AS	sickle cell trait
ASS	acute splenic sequestration
ATG	antithymocyte globulin

**B**

BFU-E	burst forming unit-erythroid
bHLH	basic helix-loop-helix
BM	bone marrow
BMFS	bone marrow failure syndromes
BMP	bone morphogenic protein
BMT	bone marrow transplantation
2,3-BPG	2,3-bisphosphoglycerate
BPGM	bisphosphoglycerate mutase
BTK	Bruton's tyrosine kinase
BW	body weight

**C**

CBT	cord blood transplantation
CDA	congenital dyserythropoietic anaemia
CEP	congenital erythropoietic porphyria
CERA	continuous erythropoietin receptor activator
CFU-E	colony forming unit-erythroid

CHA	chronic haemolytic anaemia
CHAP	Chicago Health and Aging Project
CHC	hereditary cryohydrocytosis
CHF	chronic heart failure
CHr	reticulocyte haemoglobin content
CIDA	cytolysis, inflammation, dysmetabolism and alcoholism
CIS	cytokine-inducible SH2 protein
CLT	clotrimazole
CML	chronic myeloid leukaemia
CMML	chronic myelomonocytic leukaemia
CMV	cytomegalovirus
COPD	chronic obstructive pulmonary disease
COX	cytochrome C oxidase
CRG	Cochrane Review Group
CRP	C-reactive protein
CP	Chuvash polycythaemia
CPO,	coproporphyrinogen oxidase
CS	Caesarian section
CsA	cyclosporine A
CT	computerised tomography

**D**

DBA	Diamond-Blackfan anaemia
DFO	deferroxamine
DFP	deferiprone or L1
DFX	deferasirox
DHPLC	denaturing high pressure liquid chromatography
DHS	dehydrated hereditary stomatocytosis
DHTR	delayed haemolytic transfusion reaction
DIHA	drug-induced immune haemolytic anaemia
DISC	death-inducing signalling complex
DLCO	diffusing capacity of the lung for carbon monoxide
DLI	donor lymphocyte infusion
DMT1	divalent metal transporter 1
DR	death receptor
DRTA	distal renal tubular acidosis
DVT	deep vein thrombosis

**E**

eADA	erythrocyte adenosine deaminase
eALA-S	erythroid delta aminolaevulinic acid synthase
EAN	epiphyseal aseptic necrosis
EDTA	ethylene diamine tetra-acetic acid

EEC	endogenous erythroid colonies
EKLF	erythroid Kruppel-like factor
ELISA	enzyme-linked immunosorbent assay
EM	electron microscopy
Emp	erythroblast macrophage protein
EMH	extramedullary haematopoiesis
Epo	erythropoietin
EPO	erythropoietin gene
EpoR	erythropoietin receptor
EPP	erythropoietic protoporphyria
ESA	erythropoiesis stimulating agent
ET- 1/3	endothelin-1/3
ET	essential thrombocythaemia
EtOH	ethanol
ETS	external transcribed spacer

**F**

FDA	US Food and Drug Administration
FECH	ferrochelatase
FGF	fibroblast growth factor
FIH	factor inhibiting HIF
FLVCR	feline leukaemia virus C receptor
FOG-1	Friend of GATA-1
FP	familial pseudohyperkalaemia

**G**

GA	gestational age
Gfi-1B	growth factor independence 1B
GH	growth hormone
GI	gastrointestinal
Glu	glucosamine
GLUT1	glucose transporter-1
GSH	glutathione
GSK3	glycogen synthetase kinase-3
GMP	good manufacturing practice
GPA/C/D	glycophorin A/C/D
G6PD	glucose 6 phosphate dehydrogenase
GPI	glycosyl-phosphatidyl-inositol
GPI	glucose phosphate isomerase
GR	glutathione reductase
GS	glutathione synthetase
GT	gene therapy
GvHD	graft-versus-host disease

**H**

Hb	haemoglobin
HBV	hepatitis B virus
HC	hereditary coproporphyria
hCG	human chorionic gonadotropin
HCP1	haem carrier protein 1
Hct	haematocrit
HCV	hepatitis C virus
HD	haemodialysis
HDFN	haemolytic disease of the foetus and newborn
HDW	red cell Hb distribution width
HEMPAS	haemolytic anaemia with a positive acidified serum test
HE	hereditary elliptocytosis
HEP	hepato-erythropoietic porphyria
HES	hypereosinophilic syndrome
HFE	hereditary haemochromatosis protein
HH	hereditary haemochromatosis
HHCS	hyperferritinaemia-cataract syndrome
HIF	hypoxia inducible factor
HLF	HIF-like factor
HIV	human immunodeficiency virus
HJV	haemojuvelin
HK	hexokinase
hMG	human menopausal gonadotrophin
HNSHA	hereditary non-spherocytic haemolytic anaemias
hNTH1	human endonuclease III homolog 1
HPFH	hereditary persistence of foetal haemoglobin
HRI	haem-regulated inhibitor
HRF	HIF related factor
HS	hereditary spherocytosis
HSC	haematopoietic stem cell
HSCT	haematopoietic stem cell transplantation
HSP	heat shock protein
HSs	DNase1 hypersensitive sites
HTR	haemolytic transfusion reactions
HU	hydroxyurea
HUS	haemolytic uraemic syndrome

**I**

IAP	inhibitor of apoptosis
IBD	inflammatory bowel disease
ICI	intra-cavernous injection
IDA	iron deficiency anaemia

IE	idiopathic erythrocytosis
IF	initiation factor
IFN	interferon
Ig I	immunoglobulins
IGF-1	insulin growth factor 1
IH	intracranial haemorrhage
IL	interleukin
IMF	idiopathic myelofibrosis
In	inositol
INF	interferon
IPAS	inhibitory PAS protein
iPS	induced pluripotent stem cells
IPSS	International Prognostic Scoring System
IRE	iron responsive element
IRIDA	iron refractory iron deficiency anaemia
IRP	iron regulatory protein
ISC	irreversible sickle cells
ISC	iron sulfur clusters
IST	immunosuppressive therapy
IUGR	intrauterine growth retardation

**J**

JAK2	Janus tyrosine kinase-2
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**K**

KCC	K-Cl cotransport
KIRs	killer immunoglobulin-like receptors

**L**

LC	liquid chromatography
LCR	locus control region
LDH	lactate dehydrogenase
LIC	liver iron concentration
LINE	repeats long interspersed repeats
LOH	loss of heterozygosity
LPI	labile plasma iron
LPS	lipopolysaccharide
LTC-IC	long-term culture-initiating cells
LU	leg ulcer
LVEF	left ventricular ejection fraction

**M**

MAC	membrane attack complex
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MALDI	matrix-assisted laser desorption ionisation
Man	mannose
MAPK	Ras/mitogen-activated protein kinase
MARE	maf recognition element
MCH	mean corpuscular haemoglobin content
MCV	mean corpuscular volume
MDS	myelodysplastic syndrome(s)
MFS	major facilitator superfamily
MiRNA	micro RNA
MPD	myeloproliferative disease
MPL	thrombopoietin receptor
MPN	myeloproliferative neoplasms
MRA	magnetic resonance angiography
MRI	magnetic resonance imaging
mRNA	messenger ribonucleic acid
MS	mass spectrometry
MtF	mitochondrial ferritin
MUD	matched unrelated donor
MW	molecular weight
<b>N</b>	
NEMO	NFkB essential modulator
NF-E2	nuclear factor - erythroid-derived 2
NGAL	neutrophil gelatinase-associated lipocalin
NHANES	National Health and Nutrition Examination Survey
NMD	nonsense-mediated decay
NO	nitric oxide
NRAMP	natural resistance – associated macrophage protein
NSAID	non-steroidal anti-inflammatory drug
NTBI	non-transferrin-bound iron
<b>O</b>	
OHS	overhydrated hereditary stomatocytosis
<b>P</b>	
PAS	Per Arnt Sim domain
PBG	porphobilinogen
PFCP	primary familial congenital polycythaemia
PCH	paroxysmal cold haemoglobinuria
PCT	porphyria cutanea tarda
PGK	phosphoglycerate kinase
PFK	phosphofructokinase
PFEMP1	<i>P. falciparum</i> erythrocyte membrane protein 1

PHT	pulmonary hypertension
PI	phosphatidylinositol residue
PIG-A	phosphatidyl inositol glucosyltransferase
PI-3K	phosphoinositide-3 kinase
PHD	prolyl hydroxylases
PHT	pulmonary hypertension
PK	pyruvate kinase
PMF	primary myelofibrosis
PMN	polymorphonuclear neutrophil
PNH	paroxysmal nocturnal haemoglobinuria
PP	prolonged priapism
PPIX	protoporphyrin IX
PPOX	protoporphyrinogen oxidase
PRCA	pure red cell aplasia
PS	phosphatidylserine
PTK	protein-tyrosine kinase
PTM	post-translational modification
PUS1	pseudouridine synthase 1 gene
PV	polycythaemia vera
PVSG	Polycythaemia Vera Study Group
<b>Q</b>	
QTL	quantitative trait locus
<b>R</b>	
RA	refractory anaemia
RAEB	refractory anaemia with excess blasts
RAEBt	refractory anaemia with excess blasts in transformation
RACK-1	receptor of activated protein kinase C
RAS	refractory anaemia with ring sideroblasts
RBC	red blood cell
RCMD	refractory cytopenia with multilineage dysplasia
RDW	red cell distribution width
RES	reticuloendothelial system
RhAG	Rh-associated glycoprotein
ROS	reactive oxygen species
<b>S</b>	
SA	sideroblastic anaemia
SAA	severe aplastic anaemia
SCA	sickle cell anaemia
SCD	sickle cell disease
SCF	stem cell factor

SDF	stromal derived factor
SDS-PAGE	sodium dodecyl sulfate-polyacrylamide gel electrophoresis
SF	serum ferritin
SIR	signal intensity ratio
SLE	systemic lupus erythematosus
SMD	systemic mast cell disease
SOCS	suppressor of cytokine signaling
SP	stuttering priapism
SQUID	superconducting quantum interference device
SS	homozygous sickle cell disease
STAT	signal transducer and activator of transcription
sTfR	soluble transferrin receptor

**T**

T4	thyroxine
T-ALL	T-cell acute lymphoblastic leukaemia
TAR	thrombocytopenia with absent radii
TCD	transcranial Doppler
TEC	transient erythroblastopenia of childhood
Tf	transferrin
TfR	transferrin receptor
TGF	transforming growth factor
Thal	thalassaemia
TI	thalassaemia intermedia
TIA	transient ischaemic attacks
TK	tyrosine kinase
TNF	tumour necrosis factor
TPI	triose-phosphate isomerase
TPO	thrombopoietin
TRH	thyrotropin-releasing hormone
TRM	transplant-related mortality
TRMA	thiamine-responsive megaloblastic anaemia
TRV	tricuspid regurgitation velocity
TS	transferrin saturation
TSP	thrombospondin
TSH	thyroid stimulating hormone
TTSP	transmembrane serine protease
TTD	trichothiodystrophy
TTP	thrombotic thrombocytopenic purpura
UIE	urinary iron excretion

**U**

UDPG	uridine 5'-diphospho-alpha-D-glucose
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UROD urodecarboxylase

### **V**

VCAM-1 vascular adhesion molecule-1  
VEGF vascular endothelial growth factor  
VHL von Hippel-Lindau  
VOC vaso-occlusive crisis  
VP variegate porphyria

### **W**

WHO World Health Organisation

### **X**

X-LDPP X-linked dominant protoporphyria  
XLSA/A X-linked sideroblastic anaemia/with ataxia  
XPD xeroderma pigmentosum

### **Z**

ZPP zinc protoporphyrin