

Report of the committee on nomenclature

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The guidelines outlined below are based on the decisions made at an interim meeting of the committee held on April 3-4, 1975. These guidelines, including a list of suggested names for enzyme loci, were approved during a plenary session of the Gene Mapping Workshop in Baltimore, 1975.

Consideration was confined to enzyme nomenclature because the blood group antigens have been dealt with in the standard text by Race and Sanger (1), while participants in the Histocompatibility Testing workshops have handled the terminology problems in that area (2). In general, the guidelines for naming enzymes can also be applied to the plasma proteins.

Guidelines for Genetic Nomenclature of Human Enzymes

Essential to a satisfactory terminology are that it be precise and unambiguous, <sup>that</sup> it clearly distinguish between genotypes and phenotypes, and <sup>that</sup> as far as possible, the symbols used should readily identify the particular enzyme. In addition it should be sufficiently flexible to <sup>which were</sup> permit some unusual symbols/used to designate certain enzymes in the original papers and have subsequently been widely adopted in the literature. It should also be capable of incorporating new discoveries as they are reported.

The following general scheme appears to meet these requirements and is reasonably convenient in practice.

## I. Genotypes

Genotypic symbols, i.e. for loci or alleles, are italicised (or underlined in typescript) to distinguish them clearly from symbols used to designate phenotypes, which are not italicised or underlined.

### A. Loci

1. Loci are designated by letters, either all capitalized (preferred) or just the first letter. Usually two or three letters will suffice, but sometimes four or even five may be required.

Examples: ADA for adenosine deaminase

UMPK for uridine monophosphate kinase

Gd for glucose-6-phosphate dehydrogenase

(Obviously, when lower case letters are used to designate one locus, it is undesirable to use the same letters but in capitals (e.g. GD) to designate another locus.) The letters chosen for locus names are preferably based on the recommended name given by the Enzyme Commission on Nomenclature. However, this is sometimes inconvenient or confusing because of past usage. Thus, GOT is preferred for glutamic-oxaloacetic transaminase, although the E.C. recommended name is aspartate aminotransferase. In some cases, Greek letters are also needed for clarity. Example:  $\alpha$ GAL for  $\alpha$ -galactosidase to distinguish it from  $\beta$ -galactosidase ( $\beta$ GAL).

2. There are often two or more loci coding for different polypeptide chains which are contained in separate enzyme proteins having very similar or identical catalytic properties. Such loci are best differentiated by appropriate subscripts.

Examples:

PGM<sub>1</sub>, PGM<sub>2</sub> and PGM<sub>3</sub> for the three phosphoglucomutase loci

ADH<sub>1</sub>, ADH<sub>2</sub> and ADH<sub>3</sub> for the three alcohol dehydrogenase loci

Although numerical subscripts are often most convenient, sometimes because of past usage or ease of identification, letters are preferred to avoid confusion.

Examples:

LDH<sub>A</sub>, LDH<sub>B</sub> and LDH<sub>C</sub> for the three lactate dehydrogenase loci

PGAM<sub>M</sub> and PGAM<sub>B</sub> for the two phosphoglycerate mutase loci which are active in muscle and brain, respectively.

Some enzymes occur in a so-called soluble (or supernatant or cytosol) form and also in a mitochondrial form, with the two forms being catalytically similar but coded at separate loci. In such cases, the use of S and M as subscripts may be less confusing than numerical or alphabetical designations.

Example:

GOT<sub>S</sub> and GOT<sub>M</sub> for the soluble and mitochondrial forms of glutamic-oxaloacetic transaminase.

B. Alleles

Different alleles at the same locus are designated by superscripts.

Example:

$\underline{PGM}_1^1$ ,  $\underline{PGM}_1^2$ ,  $\underline{PGM}_1^3$ ,  $\underline{PGM}_1^4$  etc., for alleles at the  $\underline{PGM}_1$  locus.

The superscripts may be numerical or alphabetical. In rare cases, + and - signs, when used extensively in the past, may be retained.

Example:

$\underline{Gd}^B$ ,  $\underline{Gd}^A$ ,  $\underline{Gd}^{A-}$  for the three alleles at the glu-  
common  
cose-6-phosphate dehydrogenase locus/ in Black popula-  
tions.

In other cases, place names are best used as the allele superscript to avoid confusion.

Example:

$\underline{Gd}^{\text{Mediterranean}}$ ,  $\underline{Gd}^{\text{Canton}}$ ,  $\underline{Gd}^{\text{Athens}}$ ,  $\underline{Gd}^{\text{Seattle}}$

(Abbreviation of the place name may be more convenient.)

So-called "null" or "silent" alleles with little or no associated enzyme activity are best designated by the superscript 0 (i.e. zero), although the letter s may be retained because of common usage.

Examples:

$\underline{PGM}_1^0$ ,  $\underline{E}_1^S$  ("silent" allele of the serum cholinesterase first locus)

When heterogeneity between "null" alleles can be demonstrated, the allele designation should be qualified, as by a place name.

Example:

$\underline{ADA}^0$  Calcutta

C. Examples of Genotypes

The following are some typical examples of genotypes written in accordance with the above recommendations and section D (below).

1. Heterozygote for the two common alleles at the ADA locus:

$$\underline{ADA^1ADA^2} \quad (\text{or } \underline{ADA^1/ADA^2})$$

2. Heterozygotes for one or the other of these common ADA alleles and a "null" allele not separable from other "null" alleles at this locus:

$$\underline{ADA^1ADA^0} \text{ and } \underline{ADA^2ADA^0} \quad (\text{or } \underline{ADA^1/ADA^0} \text{ and } \underline{ADA^2/ADA^0}) ;$$

3. Genotype of an individual heterozygous for the two common alleles of PGM<sub>1</sub>, homozygous for the common allele of PGM<sub>2</sub> and heterozygous for the two common alleles of PGM<sub>3</sub> (3 unlinked loci):

$$\underline{PGM_1^1/PGM_1^2}, \quad \underline{PGM_2^1/PGM_2^1}, \quad \underline{PGM_3^1/PGM_3^2}$$

or

$$\frac{\underline{PGM_1^1}}{\underline{PGM_1^2}} \quad \frac{\underline{PGM_2^1}}{\underline{PGM_2^1}} \quad \frac{\underline{PGM_3^1}}{\underline{PGM_3^2}}$$

D. Linkage and Phase

A slash, either horizontal or semivertical (— or /) separating alleles, implies chromosomal location. The slash may be omitted in designating the genotype at a single locus. However, if two or more loci are involved, a horizontal line is recommended, particularly if the loci are syntenic.

1. Non-syntenic loci may be designated either by an interrupted horizontal line or by individual slashes and separation by commas.

Example:

$$\frac{\underline{ADA}^1}{\underline{ADA}^2} \quad \frac{\underline{PGM}_1^1}{\underline{PGM}_2^2} \quad \text{or} \quad \underline{ADA}^1/\underline{ADA}^2, \quad \underline{PGM}_1^1/\underline{PGM}_2^2$$

2. When the loci are in the same linkage group and the phase is known, the horizontal line is continuous.

Example:

$$\frac{\underline{AMY}_1^A \quad \underline{AMY}_2^B}{\underline{AMY}_1^B \quad \underline{AMY}_2^A} \quad (\text{i.e. } \underline{AMY}_1^A \text{ and } \underline{AMY}_2^B \text{ are in cis position, as are their alleles})$$

3. When the loci are in the same linkage group but the phase is not known, a semicolon is used.

Example:

$$\frac{\underline{AMY}_1^A}{\underline{AMY}_1^B} ; \frac{\underline{AMY}_2^A}{\underline{AMY}_2^B}$$

4. To designate loci which are syntenic but not in the same linkage group, a colon is used.

Example:

$$\frac{\underline{AMY}_2^A}{\underline{AMY}_2^B} : \frac{\underline{PGM}_1^1}{\underline{PGM}_1^2}$$

## II. Phenotypes

- A. The phenotypic designation should have the same letters and subscripts as the locus (but not italicised or underlined), followed by the numerical, alphabetical or other symbol for the alleles, but not as superscripts. In the case of homozygotes for any allele or heterozygotes for a "null" allele, only one allele symbol is used.

Examples:

<u>Genotype</u>	<u>Phenotype</u>
<u>ADA<sup>1</sup>ADA<sup>1</sup></u>	ADA 1
<u>ADA<sup>1</sup>ADA<sup>2</sup></u>	ADA 2-1
<u>ADA<sup>2</sup>ADA<sup>2</sup></u>	ADA 2
<u>ADA<sup>1</sup>ADA<sup>0</sup></u>	ADA 1
<u>ADA<sup>2</sup>ADA<sup>0</sup></u>	ADA 2
<u>PGM<sub>1</sub><sup>1</sup>/PGM<sub>1</sub><sup>2</sup></u> , <u>PGM<sub>2</sub><sup>1</sup>/PGM<sub>2</sub><sup>1</sup></u> , <u>PGM<sub>3</sub><sup>1</sup>/PGM<sub>3</sub><sup>2</sup></u>	PGM <sub>1</sub> 2-1, PGM <sub>2</sub> 1, PGM <sub>3</sub> 2-1

For hemizygotes, heterozygotes and homozygotes of the X-linked phosphoglycerate kinase alleles PGK<sup>1</sup> and PGK<sup>2</sup>,

<u>Genotype</u>	<u>Phenotype</u>
<u>PGK<sup>1</sup></u>	PGK 1
<u>PGK<sup>2</sup></u>	PGK 2
<u>PGK<sup>1</sup>PGK<sup>1</sup></u>	PGK 1
<u>PGK<sup>1</sup>PGK<sup>2</sup></u>	PGK 2-1
<u>PGK<sup>2</sup>PGK<sup>2</sup></u>	PGK 2

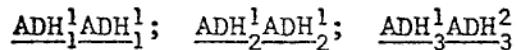
III. Isozyme Subunits

When two or more loci code for different polypeptide chains which occur together as subunits of single isozymes in a set of isozymes, it is useful to designate the subunit structure of the individual isozymes. Greek letters are convenient symbols for the polypeptide chains. A different letter can be used for the peptide product of each locus, by analogy with the  $\alpha$ ,  $\beta$ ,  $\gamma$  and  $\delta$  chains of hemoglobin. Whenever there are two or more alleles at a given locus coding for structurally different forms

of the same polypeptide, superscripts are incorporated which are the same as the superscripts used to designate the corresponding alleles.

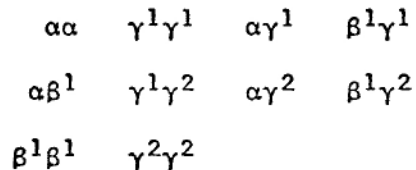
Example:

The three loci of alcohol dehydrogenase, ADH<sub>1</sub>, ADH<sub>2</sub> and ADH<sub>3</sub> are thought to code for three different polypeptide chains:  $\alpha$ ,  $\beta$  and  $\gamma$ . There is evidence for two common alleles at the ADH<sub>2</sub> locus: ADH<sub>2</sub><sup>1</sup> and ADH<sub>2</sub><sup>2</sup>. These alleles code for polypeptides  $\beta^1$  and  $\beta^2$ . There are also two common alleles at the ADH<sub>3</sub> locus: ADH<sub>3</sub><sup>1</sup> and ADH<sub>3</sub><sup>2</sup>, which code for polypeptides  $\gamma^1$  and  $\gamma^2$ . All of the ADH isozymes are dimeric and the subunits interact with each other. In adult liver, all three loci are active. Thus, some of the isozymes are homodimers and some are heterodimers. The heteromeric isozymes contain polypeptides coded by alleles at either the same locus or at different loci. Thus, if an individual has the genotype



the phenotype is ADH<sub>1</sub> 1, ADH<sub>2</sub> 1, ADH<sub>3</sub> 2-1

and in the electrophoretic pattern of a liver extract, there are ten isozymes with the following subunit structures:





## References

1. Race, R.R. and Sanger, R.: Blood Groups in Man. London: Blackwell, 1975 (6th ed.).
2. Svejgaard, A., Hauge, M., Jersild, C., Platz, P., Ryder, L.P., Nielsen, L.S. and Thomsen, M.: The HLA System: An Introductory Survey. Vol. 7 of Monographs in Human Genetics. Basel: S. Karger, 1976.

In the following table, the enzyme name given is usually that recommended in 1972 by the Enzyme Commission.\* When the E.C. name has not been used as the basis for the symbol, or if another name is much more familiar, the E.C. name is given first, and enclosed in brackets. (In a few instances the E.C. name is not given because it is so similar to the more familiar name.) The locus symbol given first is that recommended by this committee. Alternatives are also listed; these are based on systematic or obsolete names which can nearly always be found in the reference.\* The computer symbols in the table are meant to be initial suggestions; they may require individual revision. The final column indicates that the given locus has been reported to be polymorphic in at least one large ethnic group.

\*Enzyme Nomenclature: Recommendations (1972) of the International Union of Pure and Applied Chemistry and the International Union of Biochemistry. Published in 1973 by Elsevier (Amsterdam) and American Elsevier (New York).

Table of Phenotypes for which Information on Chromosomal Assignment is Available

Name of Phenotype	VAM No. ††	E.C. No** (unless not appli- cable = NA)	Locus	Alternatives	Chromosome Assignment or Linkage Group	Computer Symbol †	Polymorphic?
ABO blood group	11030	NA	<u>ABO</u>		9	ABO	Yes
Acid phosphatase-1	17150	3.1.3.2	<u>AcP1</u>		2	ACP-1	Yes
Acid phosphatase-2	20095	3.1.3.2	<u>AcP2</u>		11	ACP-2	
Aconitase, ? mitochondrial	10084	4.2.1.3	<u>Aco</u>		3(P)	ACO	
Aconitase, soluble	10085	4.2.1.3	<u>Aco<sub>S</sub></u>		9(P)	ACO-S	
Adenine phosphoribosyltransferase	10260	2.4.2.7	<u>APRT</u>		16	APRT	
Adenosine deaminase	10270	3.5.4.4	<u>ADA</u>		20	ADA	
Adenosine kinase	10275	2.7.1.20	<u>AdK</u>	<u>AdoK</u>	10(P)	ADK	
Adenovirus-12 chromosome modification site-1	10293	NA	<u>Adv12-CMS-1</u>		1	<u>ADV12-CMS-1</u>	
Adenovirus-12 chromosome modification site-17	10297	NA	<u>Adv12-CMS-17</u>		17	<u>ADV12-CMS-17</u>	
Adenylate kinase-1	10300	2.7.4.3	<u>AK<sub>1</sub></u>		9	AK-1	Yes
Adenylate kinase-2	10302	2.7.4.3	<u>AK<sub>2</sub></u>		1	AK-2	
Adenylate kinase-3	10303	2.7.4.3	<u>AK<sub>3</sub></u>		9(P)	AK-3	
Amylase, pancreatic	10465	3.2.1.1	<u>Amy<sub>2</sub></u>	<u>Amy<sub>P</sub></u>	1	AMY-2	Yes
Amylase, salivary	10470	3.2.1.1	<u>Amy<sub>1</sub></u>	<u>Amy<sub>S</sub></u>	1	AMY-1	Yes
Aniridia, type II (Baltimore)	10620	NA			1(L)	AN-2	
$\alpha_1$ -antitrypsin	10740	?	<u>Pi</u>		2(I) or 12(L)	PI	
Anti-viral protein	10745	NA	<u>AVP</u>		21	AVP	

\*\* Footnotes on last page.

Name of Phenotype	VAM No.	E.C. No. (unless not appli- cable = NA)	Locus	Alternatives	Chromosome Assignment or Linkage Group	Computer Symbol	Polymorphic?
Auriculo-osteodysplasia B factor (see properdin factor B)	10900	NA	<u>AOD</u>		1(L)	AOD	
Cataract, zonular pulverulent	11620	NA	<u>Cae</u>		1	CAE	
Chido blood group	11043	NA	<u>Ch</u>		6	CH	
Citrate synthase, mitochondrial	11895	4.1.3.7	<u>CS</u>		12(P)	CS	
Complement component-2	12060	NA	<u>C2</u>		6	C2	
Complement component-4	12080	NA	<u>C4</u>		6	C4	
Complement component-8	12095	NA	<u>C8</u>		6	C8	
Desmosterol-to-cholesterol enzyme	12565	?	<u>DCE</u>	<u>D:CE</u>	20	DCE	
Diphtheria toxin sensitivity	12615	NA	<u>DTS</u>		5(P)	DTS	
Dombrock blood group	11060	NA	<u>Do</u>		1(L)	DO	
Duffy blood group	11070	NA	Fy		1	FY	
Echo 11 sensitivity	12915	NA	<u>EL1S</u>		19(L)	EL1S	
Elliptocytosis-1	13050	NA	<u>EL<sub>1</sub></u>		1	EL-1	
Enolase-1	17243	4.2.1.11	<u>Eno<sub>1</sub></u>	<u>PPH<sub>1</sub></u>	1	ENO-1	
Enolase-2	13136	4.2.1.11	<u>Eno<sub>2</sub></u>	<u>PPH<sub>2</sub></u>	12(P)	ENO-2	
Esterase activator	13325	?			4(P) or 5	ES-ACT	
Esterase-A4	13322	3.1.1.1	<u>EsA<sub>1</sub></u>	<u>Es-A<sub>1</sub></u>	11	EsA4	
Esterase D	13328	3.1.1.1	<u>EsD</u>		13	ESD	Yes
Factor B (see properdin factor B)							
Formylglycinamideribotide (FGAR) amidotransferase	10255	?	<u>adeB</u>		4(P) or 5	ADEB	

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$\alpha$ - <u>L</u> -fucosidase mitochondrial	23000	3.2.1.51	<u><math>\alpha</math>Fuc</u>		1	A-FUC	Yes
Fumarate hydratase(fumarase),/ Fumarate hydratase (fumarase), soluble/	13685 13686	4.2.1.2 4.2.1.2	<u>FH<sub>m</sub></u> <u>FH<sub>s</sub></u>	<u>FH</u> <u>FH<sub>1</sub></u>	1	FH-2, FH-M FH-1, FH-S	
Galactokinase	23020	2.7.1.6	<u>GalK</u>	<u>GK</u> , <u>GAK</u>	17	GK	
Galactose <sup>+</sup> activator	13703		<u>Gal<sup>+</sup>-Act</u>		2(P)		
Galactose-1-phosphate uridyltrans- ferase	23040	2.7.7.12	<u>GalT</u>	<u>Gt</u> , <u>Gal-1-PUT</u>	3	GAPUT, GALT	Yes
$\alpha$ -galactosidase (Fabry disease)	30150	3.2.1.22	<u><math>\alpha</math>Gal</u>		X	A-GAL, A-GAL	
Glucose-6-phosphate dehydrogenase	30590	1.1.1.49	<u>Gd</u>	<u>G6PD</u>	X	G6PDH, G6PD	Yes
$\beta$ -Glucuronidase	25322	3.2.1.31	<u><math>\beta</math>Gus</u>	<u><math>\beta</math>-Glcu</u> , <u><math>\beta</math>-Gcu</u>	7(I) or 9(I)	GUS, B-GLCU	
Glutamate- $\gamma$ -semialdehyde synthetase	13825	?	<u>GSS</u>	<u>GSASyt</u> , <u>GSAS</u>	10(P)	GSS, GSASYT	
Glutamate oxaloacetic transaminase-1	13818	2.6.1.1	<u>GOT<sub>S</sub></u>	<u>GOT-1</u> , <u>GOT<sub>1</sub></u>	10	GOT-1	
Glutathione reductase	13830	1.6.4.2	<u>GSR</u>		8(P)	GSR	Yes
Glyceraldehyde-3-phosphate dehy- drogenase	13840	1.2.1.12	<u>GAPDH</u>	<u>GAPD</u>	12(P)	GAPDH, GAPD	
Glyoxylase I	13875	4.4.1.5	<u>GLO<sub>1</sub></u>	<u>GLY-1</u> , <u>GlX-1</u>	6	G-1, GLO-1	Yes
Gm immunoglobulin types (also see immunoglobulin heavy chains)	14710- 14719	NA			12 (I)	Gm	Yes
Guanylate kinase-1	13927	2.7.4.8	<u>GuK<sub>1</sub></u>	<u>GuK<sub>1</sub></u> , <u>GUMPK<sub>1</sub></u>	1	GMPK-1, GUMPK-2	
Guanylate kinase-2	13928	2.7.4.8	<u>GuK<sub>2</sub></u>	<u>GuK<sub>2</sub></u> , <u>GUMPK<sub>2</sub></u>	1	GUK-1 & 2	
Hageman factor	23400	NA	<u>HaF</u>		7(P)	HAf	

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Haptoglobin, alpha	14010	NA	<u>Hp</u>		16	A-HP	
Hemoglobin, alpha or beta	14180,14190	NA	<u>Hb</u> , <u>Hb</u>		2(I) & 4(I)	A-HB, B-HB	
Hexokinase-1	14260	2.7.1.1	<u>Hk</u> <sub>1</sub>	<u>Hk</u> <sub>I</sub>	10	HK-1	
Hexosaminidase A	27280	3.2.1.30	<u>Hex</u> <sub>A</sub>	<u>NAGA</u> <sub>A</sub>	15	Hex-A, Hex A	
Hexosaminidase B	14265	3.2.1.30	<u>Hex</u> <sub>B</sub>	<u>NAGA</u> <sub>B</sub>	5	Hex-B, Hex B	
HLA: Major histocompatibility complex	14280,14283,NA 14284, 15785		<u>HLA-A</u> , <u>HLA-B</u> <u>HLA-C</u> , <u>HLA-D</u>	(Several)	6	HLA	
Hypoxanthine-guanine phosphoribo- syltransferase	30800	2.4.2.8	<u>HPRT</u>		X	HGPRT	
Immune response	14685	NA	<u>Ir</u>		6(L)	IR	
Immunoglobulin heavy chains (also see Gm immunoglobulin types)	14710-14719	NA			2(I)	Ig	
Indophenoloxidase (see superoxide dismutase)							
Inosine triphosphatase	14753	3.6.1.19	<u>ITP</u>		20	ITP	
Interferon-1	14757	NA	<u>If</u> <sub>1</sub>		2(P)	IF-1	
Interferon-2	14758	NA	<u>If</u> <sub>2</sub>		5(P)	IF-2	
Isocitrate dehydrogenase-1	14770	1.1.1.42	<u>ICD</u> <sub>S</sub>	<u>IDH</u> <sub>S</sub> , <u>IDH</u> -1	2	ICDH-1, IDH-1	
Isocitrate dehydrogenase, mito- chondrial	14765	1.1.1.42	<u>ICD</u> <sub>M</sub>	<u>IDH</u> <sub>M</sub> , <u>IDH</u> -2	15(P)	ICDH-2, IDH-M	
Lactate dehydrogenase A	15000	1.1.1.27	<u>LDH</u> <sub>A</sub>	<u>LDH</u> -A	11	LDH-A	
Lactate dehydrogenase B	15010	1.1.1.27	<u>LDH</u> <sub>B</sub>	<u>LDH</u> -B	12	LDH-B	
Lecithin-cholesterol acyltrans- ferase	24590	2.3.1.43	<u>LCAT</u>		15	LCAT	

Name of Phenotype	VAM No.	E.C. No. (unless not appli- cable = NA)	Locus	Alternatives	Chromosome Assignment or Linkage Group	Computer Symbol	Polymorphic?
Lethal antigen	15125-15127	NA	<u>a1</u> , <u>a2</u> , <u>a3</u>		11	AL	
Malate dehydrogenase-1	15420	1.1.1.37	<u>MDH<sub>S</sub></u>	<u>MOR<sub>S</sub></u> , <u>MOR-1</u> , <u>MDH-1</u>	2	MDH-1	
Malate dehydrogenase, mitochondrial	15410	1.1.1.37	<u>MDH<sub>M</sub></u>	<u>MOR<sub>M</sub></u> , <u>MOR-2</u> , <u>MDH-2</u>	7	MDH-2	
Malic enzyme-1	15425	1.1.1.40	<u>ME<sub>S</sub></u>	<u>MOD<sub>S</sub></u> , <u>MOD-1</u> , <u>ME-1</u>	6	ME-1	
Mannosephosphate isomerase	15455	5.3.1.8	<u>MPI</u>		15	MANPI, MPI	
β2-microglobulin	10970	NA	<u>β2M</u>		15	B-2M	
MNSs blood group	11130	NA	<u>MNSs</u>		2(L)	MNS	
Nail-patella syndrome	16120	NA	<u>NP<sub>a</sub></u>	<u>NP</u>	9	NPA	
Nucleoside phosphorylase	16405	2.4.2.1	<u>NP</u>		14	NP	
P blood group	11140	NA	<u>P</u>		6(L)	P	
Pepsinogen	16970	3.4.23 <sup>*</sup>	<u>Pg</u>	<u>Pg-5</u>	6	PEPSG, Puc	Yes
Peptidase A	16980	3.4.11. <sup>*</sup>	<u>PepA</u>		18	PEPA	Yes
Peptidase B	16990	3.4.11. <sup>*</sup>	<u>PepB</u>		12	PEPB	
Peptidase C	17000	3.4.11. <sup>*</sup>	<u>PepC</u>		1	PEPC	Yes
Peptidase D	17010	3.4.13.9	<u>PepD</u>		19(P)	PEPD	Yes
Phosphoglucomutase-1	17190	2.7.5.1	<u>PGM<sub>1</sub></u>		1	PGM-1	Yes
Phosphoglucomutase-2	17200	2.7.5.1	<u>PGM<sub>2</sub></u>		4(P)	PGM-2	Yes
Phosphoglucomutase-3	17210	2.7.5.1	<u>PGM<sub>3</sub></u>		6	PGM-3	Yes

Name of Phenotype	VAM No.	E.C. No. (unless not appli- cable = NA)	Locus	Alternatives	Chromosome Assignment or Linkage Group	Computer Symbol	Polymorphic?
6-phosphogluconate dehydrogenase	17220	1.1.1.44	<u>PGD</u>	<u>6PGD</u>	1	6PGD	Yes
Phosphoglycerate kinase	31180	2.7.2.3	<u>PGK</u>		X	PGAK, PGK	
Phosphohexose isomerase	17240		<u>PHI</u>		19	PHI	
Phosphopyruvate hydratase (see enolase)							
Phosphoribosyl glycineamide synthetase	13844	6.3.4.13	<u>GAPS</u>		21(P)	GAPS	
Polio sensitivity	17385	NA	<u>PVS</u>		19	PVS	
Properdin factor B	13847	NA	<u>Bf</u>		6	B	
Pyrophosphatase, inorganic	17903	3.6.1.1	<u>PP</u>		10	PP	
Pyruvate kinase-3 (M2)	17905	2.7.1.40	<u>PK</u> <sub>M2</sub>	<u>PK</u> <sub>III</sub> , <u>PK</u> <sub>3</sub>	15	PK-M2, PK3	
Retinoblastoma-1	18020	NA	<u>Rb</u> <sub>1</sub>		13(L)	RB <sub>1</sub>	
Rhesus blood group	11170	NA	<u>Rh</u>		1	RH	
Ribosomal RNA	18045	NA	NA		13, 14, 15, 21, 22	R-RNA	
Rodgers blood group	11171	NA	<u>Rg</u>		6	RG	
5S RNA gene(s)	18042	NA	<u>RN5S</u>		1	RN5S	
Scianna blood group	11175	NA	<u>Sc</u>		1(L)	SC	
Sclerotylosis	18160	NA	<u>Tys</u>		2(L)	TYS	
Serine hydroxymethyltransferase (glycine + A auxotroph complet- ing)	13845	2.1.2.1	<u>SHMT</u>		12(P)	SHMT	
Spherocytosis, Denver type	18290	NA	<u>Sph</u> <sub>1</sub>		8(L) or 12(L)	Sph-1	



Name of Phenotype	VAM No.	E.C. No. (unless not appli- cable = NA)	Locus	Alternatives	Chromosome Assignment or Linkage Group	Computer Symbol	Polymorphic?
Superoxide dismutase-1	14745	1.15.1.1	<u>SOD<sub>S</sub></u>	<u>IPO-A</u> , <u>SOD-A</u> , <u>SOD-1</u>	21	SOD-1	
Superoxide dismutase-2	14746	1.15.1.1	<u>SOD<sub>M</sub></u>	<u>IPO-B</u> , <u>SOD-B</u> , <u>SOD-2</u>	6	SOD-2	
SV40-T antigen	18680	NA	<u>SV40-T</u>		7(P)	SV40-T	
Testis determining factor	--	NA	<u>TDF</u>		Y	TDF	
Thymidine kinase, mitochondrial	18829	2.7.1.75	<u>TK<sub>m</sub></u>	<u>TK<sub>2</sub></u>	16(P)	TK <del>M</del>	
Thymidine kinase, soluble	18830	2.7.1.75	<u>TK<sub>S</sub></u>	<u>TK<sub>1</sub></u>	17	TK <del>S</del>	
Triseposphate isomerase	19045	5.3.1.1	<u>TPI</u>		12	TPI	
Tryptophanyl-tRNA synthetase	19105	6.1.1.2	<u>TrpRS</u>		14	TRPRS	
Uridyl diphosphate glucose pyro- phosphorylase	19175	2.7.7.9	<u>UGPP</u>		1(P)	UGPP	
Waardenburg syndrome	19350	NA	<u>WS<sub>1</sub></u>		9(L)	WS-1	
Xeroderma pigmentosum, Egyptian	27870	NA	<u>XP<sub>E</sub></u>		9(L)	XP-E	
X-linked species (or surface) antigen	31345	NA	<u>SAX</u>		X	SAX	
Y histocompatibility antigen	--	NA	<u>H-Y</u>		Y	H-Y	

\* Enzyme Nomenclature: Recommendations (1972) of the International Union of Pure and Applied Chemistry and the International Union of Biochemistry, 1973, Elsevier (Amsterdam) and American Elsevier (New York).

† When within the capability of the computer, lower case should be used as in the locus symbols.

†† Number assigned to locus in McKusick's Mendelian Inheritance in Man (4th ed., 1975 with additions).