

Recommendations for Standardized Human Pedigree Nomenclature






























Significant inconsistencies in the usage of common pedigree symbols lead to inaccurate reporting and poor interpretation of genetic events. Consequently, a Pedigree Standardization Task Force (PSTF) was established by the National Society of Genetic Counselors, and input was solicited from the American Board of Medical Genetics and the American Society of Human Genetics, among others. The article clarifies and standardizes

the symbols to be used to describe almost any familial relationship, and also demonstrates specifically how each symbol should be used. Consistent use of such standardized pedigree nomenclature will reduce the chances for incorrect interpretation of patient, family, medical, and genetic information. It also will improve the quality of patient care and facilitate communication among researchers.

Figure 1
Common Pedigree Symbols, Definitions, and Abbreviations

Instructions:

- Key should contain all information relevant to interpretation of pedigree (eg, define shading)
- For clinical (nonpublished) pedigrees, include:
 - a) family names/initials, when appropriate
 - b) name and title of person recording pedigree
 - c) historian (person relaying family history information)
 - d) date of intake/update
- Recommended order of information placed below symbol (below to lower right, if necessary):
 - a) age/date of birth or age at death
 - b) evaluation
 - c) pedigree number (eg, I-1, I-2, I-3)

	Male	Female	Sex Unknown	Comments
1. Individual	 b. 1925	 30 y	 4 mo	Assign gender by phenotype
2. Affected individual				Key/legend used to define shading or other fill (eg, hatches, dots, etc)
				With ≥2 conditions, the individual's symbol should be partitioned accordingly, each segment shaded with a different fill and defined in legend
3. Multiple individuals, number known				Number of siblings written inside symbol (affected individuals should not be grouped)
4. Multiple individuals, number unknown				"n" used in place of "?" mark
5a. Deceased individual	 d. 35 y	 d. 4 mo		Use of cross (†) may be confused with symbol for evaluated positive (+); if known, write "d." with age at death below symbol
5b. Stillbirth (SB)	 SB 28 wk	 SB 30 wk	 SB 34 wk	Birth of dead child with gestational age noted
6. Pregnancy (P)	 LMP: 7/1/94	 20 wk		Gestational age and karyotype (if known) below symbol; light shading can be used for affected and defined in key/legend
7a. Proband				First affected family member coming to medical attention
7b. Consultand				Individual(s) seeking genetic counseling/testing

Because the information is so pertinent, 2 of the numerous figures in the article are reproduced here to encourage interested readers to obtain a complete copy of the article and the inclusive figures for their own use.













Other important figures in the article include a systematic presentation of pedigree line definitions; assisted reproductive technology symbols and definitions; pedigree symbolization of genetic evaluations/testing information; and a hypothetical clinical pedigree, using recommended nomenclature.

Editor's comment: *Reviewing charts or pursuing the literature about family trees, etc, reveals many inconsistencies in symbols and other designations used in constructing pedigrees. This paper provides needed guidelines for standardization. Pediatric endocrinologists, geneticists, and all pediatricians need to at least understand the symbology used in constructing and reading genetic pedigrees. Obviously, students and residents similarly need this information. It should be incorporated into appropriate teaching programs for all involved medical personnel.*

Bennett RL, et al. *Am J Hum Genet* 1995;56:745-752.

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Figure 2
Pedigree Symbols and Abbreviations for Pregnancies Not Carried to Term

	Male	Female	Sex Unknown	Comments
1. Spontaneous abortion (SAB)	 male	 female	 ECT	If ectopic pregnancy, write ECT below symbol
2. Affected SAB	 male	 female	 16 wk	If gestational age known, write below symbol; key/legend used to define shading
3. Termination of pregnancy (TOP)	 male	 female		Other abbreviations (eg, TAB, VTOP, Ab) not used for sake of consistency
4. Affected TOP	 male	 female		Key/legend used to define shading

Chronic Metabolic Acidosis Decreases Albumin Synthesis and Induces Negative Nitrogen Balance in Humans

Ballmer et al measured the effects of experimentally induced metabolic acidosis on nitrogen balance and protein synthesis in 8 male subjects on a constant metabolic diet. Two different degrees of chronic metabolic acidosis were induced using low-dose NH_4Cl (2.1 mmol/kg body weight; $n=4$) and high-dose NH_4Cl (4.2 mmol/kg body weight; $n=4$) orally for 7 days. Albumin synthesis rates were determined by a labeled phenylalanine technique after an overnight fast. Urinary nitrogen excretion was measured, as well as plasma concentrations of insulin-like growth factor 1 (IGF-1), free thyroxine (fT_4), and triiodothyronine (T_3).

In the low-dose group, a mean pH of 7.375 and a mean bicarbonate level of 19.1 mEq/L were achieved. The plasma albumin concentration did not decrease significantly. Albumin synthesis in 3 of the 4 subjects was slightly lower than during the control period and definitely decreased in the fourth subject. Nitrogen excretion averaged 977 ± 116 mmol/24 h during

the control period and increased, but not significantly, with NH_4Cl administration.

In contrast, plasma albumin concentrations fell significantly in the high-dose group, in whom a mean pH of 7.303 ± 0.053 occurred, in addition to a significantly lower plasma bicarbonate level of 15.1 vs 19.1 mEq/L. Albumin synthesis was significantly lower than during baseline in the high-dose group, and nitrogen excretion increased significantly from $1,012 \pm 180$ mmol/24 h to $1,377 \pm 236$ mmol/24 h ($P < 0.001$). Plasma levels of IGF-1, fT_4 , T_3 , and thyrotropin all showed small but statistically significant declines during acidosis, but only when the low- and high-dose groups were combined.

The authors state that these data demonstrate for the first time that metabolic acidosis in humans decreases albumin synthesis and induces a state of sustained negative nitrogen balance. Thus, as stated by the authors, metabolic acidosis could be an important mediator of negative nitrogen balance,