Three new phenotypes of human red cell acid phosphatase: ACP1FA, ACP1GA, and ACP1GB

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Summary. Three new phenotypes of human erythrocyte acid phosphatase (ACP1) have been detected and found to be unique by direct comparison with previously identified ACP1 variants. One of these new electrophoretic variants, labeled as ACP1FA, has been detected in the Hispanic population of California. The electrophoretic variants identified as ACP1GA and ACP1GB have been detected in a black family in North Carolina. A family study has shown that ACP1FA is transmitted as an allele of ACP1.

Introduction

Human erythrocyte acid phosphatase (ACP1) polymorphism was first described by Hopkinson et al. (1963, 1964) as being under the control of three codominant, autosomal alleles ACP1A, ACP1B, and ACP1C. These three alleles code for five common phenotypes identified as ACP1A, ACP1B, ACP1BA, ACP1CA, and ACP1CB with the homozygous phenotype ACP1C being uncommon. Subsequent work by other investigators has identified additional variant alleles of ACP1 which can be differentiated by either electrophoretic mobility differences or by quantitative differences in enzymatic activity.

Rare ACP1 alleles whose gene products can be differentiated by electrophoretic mobility differences include ACP1D (Karp and Sutton 1967), ACP1E (Sorensen 1975), ACP1R (Giblett and Scott 1965), and ACP1R1 (Yoshihara and Mohrenweiser 1980). The ACP1D and ACP1E alleles are extremely rare; whereas ACP1R alleles have been reported to have gene frequencies between 0.02 and 0.236 in several South African black populations (Jenkins and Corfield 1972) and 0.023 in the North Carolina black population (Nelson 1984). The allele ACP1R1 is restricted to the primitive Ticuna Indian Tribe of South America with a gene frequency of 0.11 (Yoshihara and Mohrenweiser 1980).

Two ACP1 alleles which have been differentiated by quantitative variations in enzymatic activity include ACP1GU and ACP1A1, which although restricted to the Guaymi Indians of Panama and Costa Rica, reaches a gene frequency of 0.132 (Tanis et al. 1977; Mohrenweiser and Novotny 1982), and ACP1E (Herbick et al. 1970).

This report deals with the detection of three new variant phenotypes of ACP1 which were identified by differences in electrophoretic mobility. These new variant phenotypes, designated as ACP1FA, ACP1GA, and ACP1GB, have been directly compared to the electrophoretic ACP1 variants identified to date. Family studies have shown that ACP1C is transmitted as an allele of ACP1. Unfortunately family studies were not possible for the individual having the phenotype ACP1FA.

Materials and methods

Most blood samples were collected by venipuncture into vacuum tubes containing no preservatives or anticoagulants. The ACP1FA sample was collected by venipuncture into vacuum tubes containing EDTA. After removal of the plasma, a portion of the packed red cells was pipetted onto a piece of clean white cotton sheeting. Bloodstains prepared in this manner were stored at -20°C until tested. Lyophilized powders were prepared from triple washed packed red cells to preserve enzymatic activity over prolonged periods of time.

Electrophoresis was performed using two methods. The first employed at 10% starch gel 20 x 20 x 0.1 cm in size using a 0.1 M trisodium citrate-0.29 M sodium dihydrogen phosphate buffer (pH 5.7) as described by Nelson (1982). The second method used a 10% starch gel 20 x 20 x 0.2 cm in size with a 0.15 M trisodium citrate-0.245 M sodium dihydrogen phosphate buffer (pH 5.9) as modified from Wraxall and Emes (1976). The ACP1 isozyme bands were visualized on the gel by overlaying the entire surface of the gel with a piece of Whatman 3MM chromatography paper soaked with a 0.003 M 4-methyl-umbelliferyl phosphate solution in a 0.05 M citric acid/sodium hydroxide buffer as described by Wraxall and Emes (1976). The fluorescent bands were visualized using long wave ultraviolet light, and weakly staining isozyme bands were intensified by removing the paper overlay and pouring 0.5 M sodium hydroxide on the surface of the gel.

Results and discussion

Each ACP1 allele produces two isozyme bands; one of which has a greater staining intensity than the other. The isozyme bands produced by ACP1A are electrophoretically unique.
However the more intensely staining (major) and less intensely staining (minor) isozyme bands produced by $ACP_1^B$ and $ACP_1^C$ overlap. Hence identification of the phenotypes produced by $ACP_1^B$ and $ACP_1^C$ are made by a determination of the relative banding intensities as well as by migrational differences. To more easily identify specific isozyme bands in this report; the more intensely staining isozyme band is given the same letter identifier as the allele which produces it and the weaker staining isozyme band is given the same letter identifier as the allele which encodes it followed by a prime ('). For example, $ACP_1^A$ produces two isozyme bands; the more intensely staining anodal band is identified as the $a$ band and the weaker staining cathodal isozyme band is identified as the $a'$ band (Fig. 1).

A composite photograph and schematic diagram illustrating the new $ACP_1$ variants in the citrate-phosphate pH 5.7 buffer system is shown in Fig. 2. Also shown in Fig. 2 are phenotypes which represent all other previously identified $ACP_1$ electrophoretic variant alleles. The new $ACP_1$ phenotypes have been labeled in Fig. 2 as $ACP_1FA$, $ACP_1GA$, and $ACP_1GB$. The $ACP_1FA$ was detected by W.K. Carlton in Fresno, California in a Hispanic male and the $ACP_1GA$ was detected by E.A. Smith in Durham, North Carolina in a black male. The $ACP_1GB$ phenotype originated from the mother of the $ACP_1GA$ individual.

The isozyme banding pattern of the heterozygous $ACP_1FA$ phenotype is composed of the $a$ and $a'$ bands as well as two additional isozyme bands; the minor $f'$ band which does not migrate away from the origin and the major $f$ band which migrates towards the cathode. The banding pattern of the heterozygous phenotype $ACP_1GA$ also demonstrates the $a$ and $a'$ bands, as well as an intense $g$ band which migrates to the same position as the $b'$ and $c$ isozyme bands, and a weaker $g'$ band which migrates slightly toward the cathode from the