Zygosity diagnosis using polymorphic proteins detected by two-dimensional gel electrophoresis.

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Using two-dimensional gel electrophoresis developed by O'Farrell with some modifications, we have detected 8 new polymorphic proteins and esterase D (ESD) in lymphocyte proteins in one electrophoretogram. Family and population studies indicate that phenotypes of these polymorphic proteins are determined by two common alleles at each autosomal locus. The gene for one of proteins designated LCP1 is closely linked to the gene for ESD, but others are not linked to each other.

To evaluate the efficiency of the polymorphisms in application of zygosity diagnosis, we have analyzed phenotypes of polymorphic proteins in 40 twin pairs (39 pairs of same sexed and one pair of opposite sexed). The phenotypes of 2 out of 8 these proteins were discordant in one pair of opposite sex and 4 like-sexed pairs. In other 35 pairs, phenotypes of 8 polymorphic proteins were perfectly concordant. The Probabilities of monozygosity [Pr(MZ)] calculated from the formular of Essen-Möller were ranged from values of 0.998 to 0.875 in these pairs, using phenotypes of proteins from twin pairs and their parents. The Pr(MZ)s based on the ordinarily used genetic markers such as blood groups, types of serum and isoenzymes, were from 0.998 to 0.850 in them. In addition, the Pr(MZ)s based on combined two data were over 0.99 in these twin pairs. These data suggest that the polymorphisms detected by two-dimensional gel electrophoresis is very useful for the diagnosis of zygosity to similar extent by means of blood groups, types of serum and isoenzymes.