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Evidence for a "New" Allele at the Esterase D (E.C. 3.1.1.1.) Locus

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Summary. An apparently new EsD gene product (EsD*Düsseldorf) was detected by use of horizontal agarose gel electrophoresis (AGE), starch gel electrophoresis (SGE), and isoelectric focusing (IEF). The observed phenotype EsD (1-Düsseldorf) can be distinguished from any known EsD type.

Key words: EsD, new variant – Blood groups, EsD

Zusammenfassung. Eine neue Variante des Isoenzymsystems EsD wird beschrieben. Das Bandenmuster unterscheidet sich nach elektrophoretischer Trennung nur wenig vom Typ EsD (2-1). Das neue Genprodukt imponiert wenig anodal gegenüber dem des Allels EsD*2. Eine sichere Unterscheidung ist mit Hilfe der isoelektrischen Fokussierung möglich. Familienstudien waren nicht möglich.

Schlüsselwörter: EsD, neue Variante – Blutgruppen, EsD

The erythrocyte enzyme EsD (E.C. 3.1.1.1.) located on chromosome 13 [3] was discovered by Hopkinson et al. [6] who described two common codominant alleles EsD*1 and EsD*2. Rare additional variants have been reported: EsD*3, EsD*4, EsD*5, and EsD*6[2] [2, 4, 10–12]. Eriksen and Dissing [5] observed a variant allele, which was named EsD*Copenhagen. The existence of a silent allele EsD*0 has also been reported by Marks et al. [9], Sparkes et al. [13], Koziol and Stéphién [7], Basler and Henke [1]. The following report describes the findings of a "new" variant (provisoriously named EsD*Düsseldorf or EsD*D) in the red cells of a white male child.

Materials and Methods

The specimen for testing was drawn in sodium citrate (3.8% w/v). EsD was initially phenotyped by conventional electrophoresis in starch gel and in agarose gel using the technique of Kühnl et al. [8]. Stroma-free hemolysates were used. After observing an isozyme pattern, which appeared to be *slightly* unusual, it was decided to focuse the specimen concerned.

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2 The existence of a "new" allele called EsD*7 has been reported recently (Siege M, Schwehn B (1983) Ärztl Lab 29:288–290)

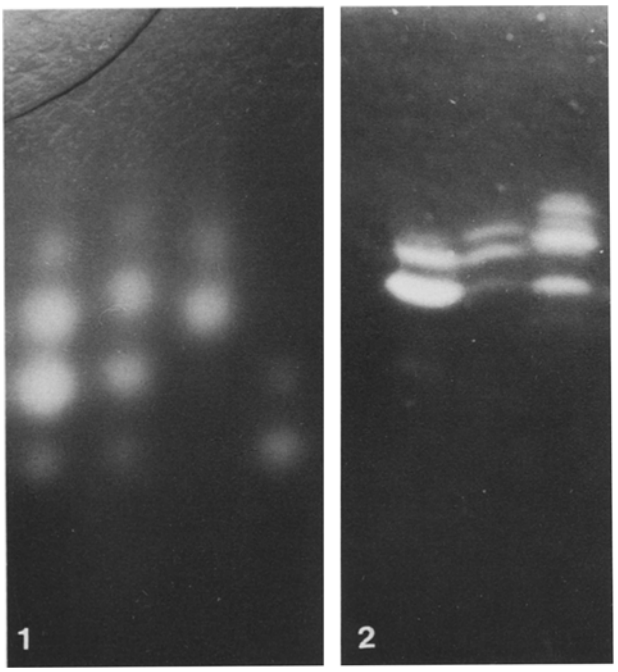

Fig. 1. Isozyme pattern of EsD phenotypes after SGE (from left to right): EsD 2-1, D-1, 2, 1

Fig. 2. EsD phenotypes after IEF (from left to right): EsD 2-1, 5-1, D-1

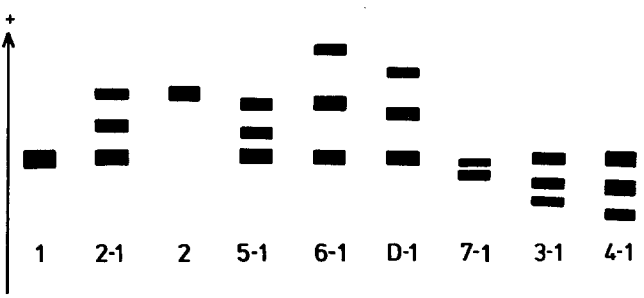

Fig. 3. Schematic drawing of EsD phenotypes after AGE (only the main bands are presented). The banding pattern of EsD (Cph-1) is reported to be very similar to that of EsD (3-1) but is displaying a slightly faster anodic mobility (5)

Acrylamide gels Ampholine PAG-plates, pH range 4–6.5, were obtained from LKB. The Pharmacia 'FBE 3000' and the 'ECPS 2000/300' apparatuses were used throughout this study. Electrofocusing was carried out across the width of the gel on a cooling plate through which a water/ethanol mixture was circulated at 5° C. Filter paper strips which connected the gel to the 'cathodal' and 'anodal' electrodes were soaked in 0.1 *M* alanin and 0.1 *M* glutamic acid in 0.5 *M* H_3PO_4, respectively. Focusing was continued for 150 min during which time the voltage was increased stepwise from 200 to 1,200 V. Plates were stained using a filter paper overlay technique according to that used for starch or agarose gels. The staining mixture was the same as that described by Hopkinson et al. [6].

The individual carrying the new phenotype was a healthy white 3-year-old boy. Both the drawing of the blood sample and the testing were repeated 3 months after the first finding. The child's mother was also included into the test.

Results and Discussion

Figure 1 shows the isozyme pattern of EsD (1-D) after SGE among other common phenotypes. The main EsD*D isozyme was of slightly faster electrophoretic mobility than the common EsD*2 isozyme. *Fresh* samples of EsD (1-D) display a ratio of enzyme activity of 1 : 2 : 1, which cannot be seen in Fig. 1.

By means of IEF, the abnormal pattern of EsD (1-D) can undoubtedly be distinguished from those of EsD (2-1) and EsD (5-1), as Fig. 2 shows. The mother of the "affected" child is carrying type EsD (1). Unfortunately, a study of the family was not possible, but we believe that there can be only very little doubt that we are dealing with a new variant at the EsD locus.

Additionally, we tried to demonstrate the phenotype pattern in a diagram illustrating all isozyme patterns known so far (Fig. 3).

To our knowledge, this variant is the ninth EsD-allele described so far. We refrained from hastily naming it EsD*9, because we think this should be restricted to a nomenclature conference. In our opinion the EsD*7 allele should become EsD*8 because EsD*Cph was described earlier.

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