Z Rechtsmed (1986) 96:159–161

Zeitschrift für
Rechtsmedizin

"New" Phenotypes in the Human Red Cell Isozyme System ADA

J. Henke[1], H. Schweitzer[1], H. Cleve[2], and S. Weidinger[2]

[1] Labor für forensische Blutgruppenkunde, Otto-Hahn-Str. 39, D-4000 Düsseldorf 13, Federal Republic of Germany
[2] Institut für Anthropologie und Humangenetik der Universität, Richard-Wagner-Str. 10/I, D-8000 München 2, Federal Republic of Germany

Summary. Two rare ADA phenotypes were observed in a German mother and her child. These phenotypes may be due to the allele ADA*9 previously found in Bulgaria.

Key words: ADA, rare variant phenotypes – ADA*9

Zusammenfassung. Bei einem Mutter-Kind-Paar aus Deutschland fanden wir zwei seltene ADA-Phänotypen. Wir können die Möglichkeit nicht ausschließen, daß es sich bei dieser Variante um die gleiche handelt, die in Bulgarien gefunden und dem Allel ADA*9 zugeordnet wurde. Eine vergleichende Untersuchung war nicht möglich.

Schlüsselwörter: ADA, seltene Phänotypen – ADA*9

Introduction

Spencer et al. [11] first described inherited variations of the human red cell adenosine deaminase (ADA, E.C.3.5.4.4). They observed three phenotypes ADA 1, ADA 2-1, and ADA 2, which were controlled by two codominant alleles ADA*1 and ADA*2. Subsequently, several rare variants were identified and shown by family studies to represent heterozygous combinations of either ADA*1 or ADA*2 with a rare variant allele (ADA*3, ADA*4, ADA*5, ADA*6, or ADA*7) at the same locus [1, 4–6, 10].

There is also evidence for a silent ADA*0 [2, 3] and furthermore for an allele called ADA*8 [7] which controls an enzyme with reduced activity.

Recently, Nenkov et al. [9] described a "new" phenotype ADA 9-1. A survey of the distribution of ADA alleles in various populations was given by Weissmann et al. [12].

Offprint requests to: J. Henke (address see above)

Material and Methods

ADA isozymes were demonstrated in starch gel, agarose gel, and on cellulose acetate strips, respectively. The techniques were the same as described by Spencer et al. [11] and Martin [8].

The specimen for testing were drawn into sodium citrate (3.8% w/v). Stroma-free hemolysates were used. The individuals carrying the variant phenotypes were a healthy German woman and her daughter. Both were born in the town of Neuss, Northrhine-Westphalia (FRG).

Results and Discussion

Figure 1 shows the observed ADA isozyme patterns after cellulose acetate strip electrophoresis. It is obvious that the main isozyme of the variant phenotypes shows less enzymatic activity than ADA 2 or ADA 1, respectively. It appears that the distance between the variant isozyme band and the ADA 2 band is the same as that between the ADA 2 and the ADA 1 band.

This variant ADA is clearly different from the ADA*7 gene product which is located more closely to the ADA 2 band (Fig. 2). It is still unknown whether this variant is also different from the one that Nenkow et al. [9] described. Un-

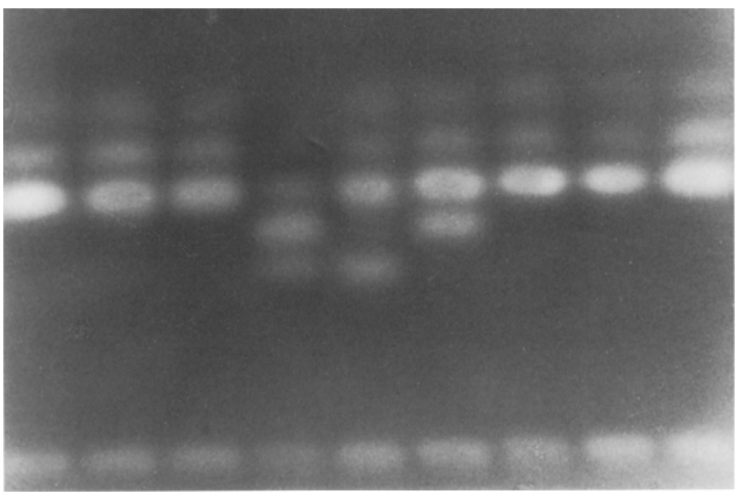

Fig. 1. ADA isozyme patterns after cellulose acetate strip electrophoresis. From left to right ADA 1,1,1,V-2,V-1,2-1,1,1,1

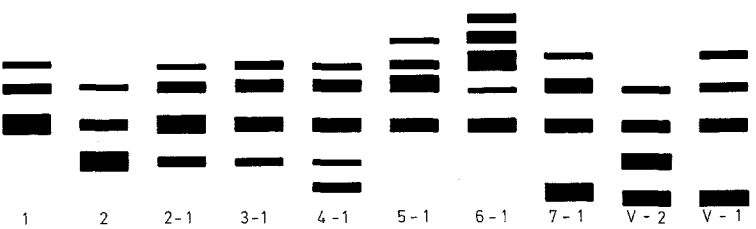

Fig. 2. Diagram showing ADA isozymes in hemolysates of various phenotypes

fortunately, a direct comparison was not possible. Their paper lacks an original photograph of the banding pattern. A schematic drawing of the ADA 9-1 pattern illustrates a similar position after electrophoresis as our variant. The enzymatic activity of the "Bulgarian" variant may, however, be increased.

Conclusions

Firstly, we have found two rare ADA variant phenotypes representing heterozygous combinations with either ADA*1 in the mother or ADA*2 in the child.

Secondly, the mother's phenotype is similar to that described by Nenkov et al. [9] in Bulgaria regarding its electrophoretic mobility. The variant we observed may be different from the Bulgarian type in its enzymatic activity. This difference can be expressed by naming the alleles ADA*9 (Sofia) and ADA*9 (Neuss).

Acknowledgement. We would like to express our thanks to Dr. D. A. Hopkinson (London, England) for his helpful suggestions.

References

1. Berg K, Cleve H, Hofmann G, Schwarzfischer F, Wendt GG, Wischerath H (1975) ADA^7 – A new allele. Vox Sang 29:217–220
2. Brinkmann B, Brinkmann M, Martin H (1973) A new allele in red cell adenosine deaminase polymorphism – ADA^0. Hum Hered 23:603–607
3. Chen SH, Scott CR, Giblett ER (1974) Adenosine deaminase: demonstration of a 'silent' gene associated with combined immunodeficiency disease. Am J Hum Genet 26:103–107
4. Detter JC, Stammatoyannopoulos G, Giblett ER, Motulsky AG (1970) Adenosine deaminase: racial distribution and report of a new phenotype. J Med Genet 7:356–357
5. Dissing T, Knudsen BB (1969) A new red cell adenosine deaminase phenotype in man. Hum Hered 19:375–377
6. Hopkinson DA, Cook PJL, Harris H (1969) Further data on the adenosine deaminase (ADA) polymorphism and a report of a new phenotype. Ann Hum Genet 32:361–367
7. Jenkins T, Rabson AK, Nurse GT, Jant AB, Hopkinson DA (1976) Deficiency of adenosine deaminase not associated with severe combined immunodeficiency. J Pediat 89:732–736
8. Martin W (1983) Kombinierte Darstellung der Systeme AK, ADA und 6-PGD in der Agarosegel-Dünnschicht-Elektrophorese. Ärztl Lab 29:165–166
9. Nenkov N, Popvassilev I, Paunova R, Rackwitz A (1981) Ein neuer Phänotyp im ADA-System. Festschrift der Humboldt Universität zum 60. Geburtstag von O. Prokop, S 32–33
10. Radam G, Strauch H, Vavrusa V (1975) Zur Differenzierung der Varianten 5-1 und 6-1 im Adenosin-desaminasepolymorphismus. Nachweis des neuen Phänotyps 5-2 in der CSSR. Humangenetik 26:151–154
11. Spencer N, Hopkinson DA, Harris H (1968) Adenosine deaminase polymorphism in man. Ann Hum Genet 32:9–14
12. Weissmann J, Vollmer M, Pribilla O (1982) Survey of the distribution of adenosine deaminase and superoxide dismutase markers in different populations. Hum Hered 32:344–356

Received January 14, 1986