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# Esterase D Phenotypes in North-eastern Japan

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**Summary.** Esterase D (EsD) was typed in 2,367 unrelated healthy blood donors from Yamagata and Miyagi districts, north-eastern area of Japan. The gene frequencies observed were: $EsD^1$=0.642, $EsD^2$=0.358. In these blood donors two rare EsD phenotypes, EsD 3–2 and a new variant, were found. As for this new variant, the mode of hereditary transmission was discussed by family study.

**Key words:** Enzyme Polymorphism, Esterase D – Esterase D

**Zusammenfassung.** Esterase D Phänotypen von 2,367 unverwandten Blutspendern in Yamagata und Miyagi, einer nordöstlichen Gegend Japans wurden stärkegel-elektrophoretisch untersucht. Die Häufigkeit der Allele war: $EsD^1$=0.642, $EsD^2$= 0.358. In diesen Stichproben wurden zwei seltene Phänotypen, EsD 3–2 und eine neue Variante, beobachtet. Der Vererbungsmodus der neuen Variante wurde auf Grund der Blutuntersuchungen der Familienmitglieder diskutiert.

**Schlüsselwörter:** Blutgruppen, Esterase D – Esterase D, Polymorphismus

Using starch gel electrophoresis, Esterase D (EsD) of one individual can be separated into various isozymes, and different zymograms can be obtained from different individuals. There are three common phenotypes, EsD 1, EsD 2–1 and EsD 2, in all populations, and they are determined by two codominant autosomal genes, $EsD^1$ and $EsD^2$. Besides these three common phenotypes, a very rare type, EsD 3–1, which is determined by another codominant autosomal gene $EsD^3$, has already been reported by Bender and Frank (1974) and Rittner and Müller (1975).

The present paper deals with the EsD phenotype and gene frequencies in the north-eastern area of Japan, and two rare types, EsD 3–2 and a new variant type, which were found among the blood donors in this area. The new variant type did not belong to EsD 3–1, EsD 3–2, nor others, and the mode of inheritance of this variant type was investigated through family study.

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**Table 1.** EsD phenotype and gene frequencies in the Miyagi and Yamagata population

| | Phenotype frequencies | | | | | | Gene frequencies | |
|---|---|---|---|---|---|---|---|---|
| | 1 | 2–1 | 2 | 3–2[a] | variant[a] | total | $EsD^1$ | $EsD^2$ |
| observed | 989 | 1061 | 315 | 1 | 1 | 2367 | 0.642 | 0.358 |
| expected | 974.8 | 1087.1 | 303.1 | | | 2365.0 | | |

[a] excluded from the calculation of gene frequency
$\chi^2 = 1.039$ $p > 0.25$

**Table 2.** EsD gene frequencies from different parts of Japan and Hessen, Germany

| Area | Number | $EsD^1$ | $EsD^2$ |
|---|---|---|---|
| Miyagi and Yamagata | 2365 | 0.642 | 0.358 |
| Mie [Ishimoto et al. 1974] | 847 | 0.650 | 0.350 |
| Kyoto [Nishimukai et al. 1976] | 353 | 0.637 | 0.363 |
| Hessen [Kühnl et al. 1974] | 510 | 0.8882 | 0.1118 |

## Materials and Methods

Blood was drawn from 2,367 healthy blood donors living in Yamagata and Miyagi districts, the north-eastern area of Japan. Blood was also drawn from other four members of the family of the blood donor who showed a new variant type. After washing with physiological saline, red cells were lysated at $-18°C$. The red cells stored in glycerin in a refrigerator were found to keep EsD activity as well as fresh red cells.

Electrophoretic separation in starch gels was performed according to the method by Hopkinson et al. (1973) with discontinuous Tris-citrate-borate-lithium hydroxide buffer system at pH 7.2. Electrophoresis was carried out for about 5 hr at $4°C$ at about 10 V/cm. The isozymes were developed according to Hopkinson et al. (1973) using 4-methyl-umbelliferyl acetate as substrate.

## Results and Discussions

EsD phenotypes and gene frequencies among 2,367 blood donors from Yamagata and Miyagi districts are shown in Table 1. EsD gene frequencies in Yamagata and Miyagi are compared with those in other parts of Japan [Ishimoto et al. 1974, Nishimukai et al. 1976] and Hessen/Germany [Kühnl et al. 1974] in Table 2. As shown in Table 2, there is little if any variation among these different parts of Japan but the $EsD^2$ gene frequency in Japan are observed to be considerably higher than that of Germany.

In these blood donors, the first rare type was found in a man. It is demonstrated together with EsD 1, EsD 2–1 and EsD 2 in Fig. 1. The enzyme bands of this rare type appeared at the sites of EsD 3 and EsD 2 and it was considered exactly as EsD 3–2. The second rare type was found in a young woman. It is shown together with EsD 1, EsD 2–1 and EsD 2 in Figure 2. The enzyme bands of this second rare type appeared at the sites of EsD 1, EsD 2 and EsD 3. It could not be determined whether it was EsD 3–1 or EsD 3–2, and it seemed likely to be such a new variant as EsD 3–1–2. By her family study, EsD phenotypes of her father, mother, sister and brother

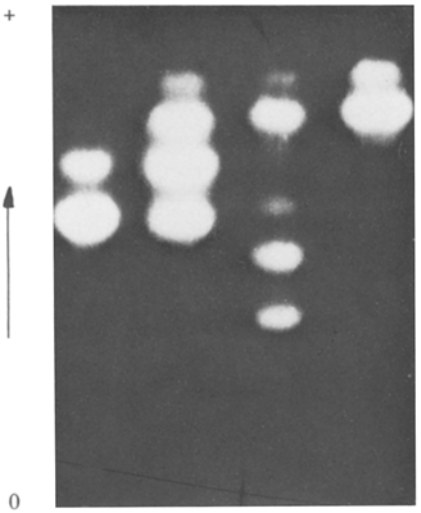


**Fig. 1.** EsD phenotypes; from left to right EsD 1, EsD 2–1, EsD 3–2 and EsD 2

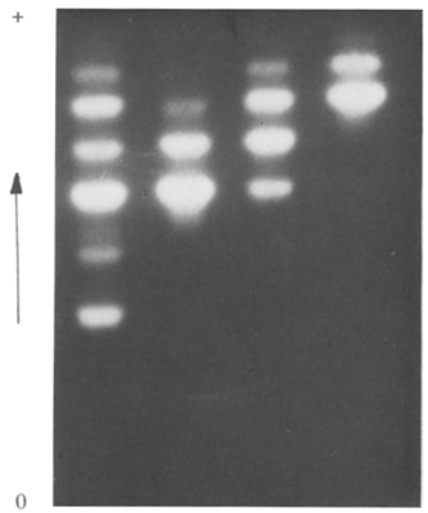


**Fig. 2.** EsD variant type together with three common EsD phenotypes; from left to right EsD variant type, EsD 1, EsD 2–1 and EsD 2

were examined. Their EsD phenotypes are shown in Fig. 3 together with EsD 1, EsD 2–1 and EsD 2. As shown in Figure 3, her mother's EsD phenotype was exactly the same as her. Her father's EsD phenotype was EsD 2–1 and her sister's was also EsD 2–1. On the other hand, her brother's EsD phenotype had distinct bands at the sites of EsD 3 and EsD 1 and was diagnosed as EsD 3–1. EsD phenotypes of two daughters and a son from the parents of EsD 2–1 and EsD variant type were EsD 2–1, EsD variant type and EsD 3–1, respectively. It is very difficult to understand the exact mode of the hereditary transmission of this variant type, but a reasonable explanation may be obtained by assuming that her mother has a gene $EsD^2$ and another gene $EsD^{3\cdot1}$, a completely linked gene between $EsD^3$ and $EsD^1$ within a cistron. According to this hypothesis, it is considered that the proband inherited the linked gene $EsD^{3\cdot1}$

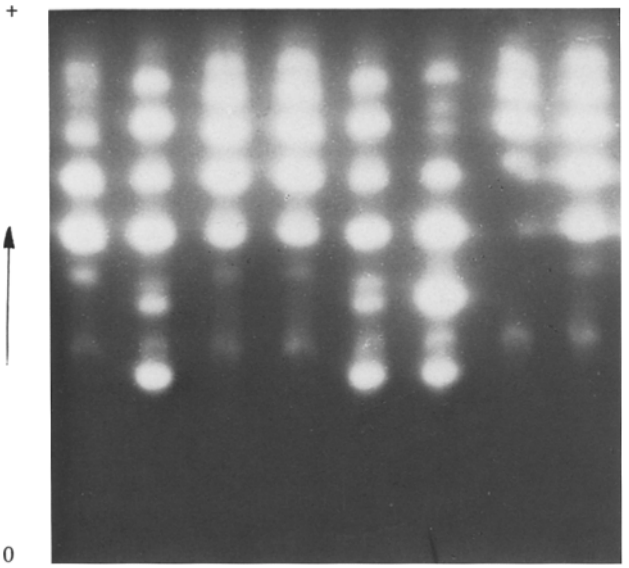


**Fig. 3.** EsD phenotypes of the members of family with EsD variant type together with three common EsD phenotypes; from left to right EsD 1, mother (EsD variant type), father (EsD 2–1), sister (EsD 2–1), proband (EsD variant type), brother (EsD 3–1), EsD 2 and EsD 2–1.

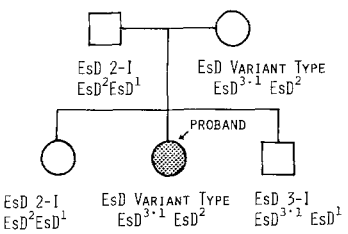


**Fig. 4.** Hereditary transmission of the EsD variant. EsD phenotypes and assumed genotypes of each members of family are listed in the pedigree

from her mother and the $EsD^2$ gene from her father, and her sister inherited the $EsD^2$ gene from her mother and the $EsD^1$ gene from her father. On the other hand, her brother inherited the linked gene $EsD^{3\cdot1}$ from his mother and the $EsD^1$ gene from his father. From such a mode of inheritance, both her and her mother's EsD genotypes are assumed to be $EsD^{3\cdot1}/EsD^2$ and her brother's to be $EsD^{3\cdot1}/EsD^1$. The pedigree is shown in Figure 4, which lists the observed EsD phenotypes and the assumed genotypes of each member of the family. The fact that EsD 2–1, EsD variant type and EsD 3–1 were born from the parents of EsD 2–1 and EsD variant type is explained without contradiction by assuming a linked gene $EsD^{3\cdot1}$. Therefore, the possibility of the presence of a linked gene $EsD^{3\cdot1}$ is suggested and this variant type of EsD may be considerd as EsD 3–1–2.

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