Human aldolase B cDNA detects a Pvu II RFLP in healthy individuals

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SOURCE AND DESCRIPTION OF CLONE: pA3, a 1.6 Kb full-length cDNA coding for human aldolase B, isolated from a human liver library cloned in pAT153/Pvu II (Paolella et al., 1984).

POLYMORPHISM: Hybridization to Pvu II digested human genomic DNA generates four bands of about 6.5, 2.1, 1.9 and 1.0 Kb in most individuals. An additional 2.3 Kb band is present in some individuals (see Figure, where the 2.3 Kb band is present in individuals 109, 15 and 20 out of the 10 reported; the 6.5 and 1.0 Kb bands are omitted for reasons of space).

FREQUENCY: Studied in 52 unrelated individuals, all from Naples (Italy) and surroundings. 29% of them showed the additional 2.3 Kb band. No homozygosity for this band was found.

NOT POLYMORPHIC FOR: Pst I, Sac I, Bam HI, Bgl II and Hind III in at least 8 unrelated individuals tested.


MENDELIAN INHERITANCE: Demonstrated in one family. The mother and the son are heterozygotes, whereas the father is a homozygote.

PROBE AVAILABILITY: Available for collaboration.

OTHER COMMENTS: Hereditary fructose intolerance is due to a lack of aldolase B activity in liver. Although we observed no 2.3 kb homozygotes, no sign of partial digestion was seen in heterozygote individuals.


ACKNOWLEDGEMENTS: This study was supported by the Ministero della Pubblica Istruzione and the Consiglio Nazionale delle Ricerche, Rome, Italy. Special acknowledgement is given to Progetto Finalizzato Ingegneria Genetica e Basi Molecolari delle Malattie Ereditarie.