

To The Editor*

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Recent progress in DNA technology, such as the establishment of PCR using allele-specific oligoprimers, made it possible to apply this technique quickly and efficiently in prenatal diagnosis in small laboratories such as ours¹. Recently, we noticed that oligoprimers prepared in the detection of frame shift codon 8, 9 (FSC89) mutation also give positive PCR reaction with the FSC 8 mutation. Dinucleotide (AA) deletion in codon 8 in FSC8 and insertion of G in FSC8,9 create similar shifts in the distal codons as follows:

	8	9	10	11	12	13	14
Normal: 5'	AAG	TCT	GCC	GTT	ACT	GCC	CTG.....3'
FSCB : 5'	—	GTC	TGC	CGT	TAC	TGC	CCT.....3'
C 8,9 : 5'	AAG	GTC	TGC	CGT	TAC	TGC	CCT.....3'

Therefore it was not surprising to detect these mutations by using the same primer matching with the newly created codons. (Fig. 1).

As we have previously reported, FSC 8 mutation is usually associated with thalassemia intermedia². Therefore, differentiation of these two mutations from each other may be critical in genetic counseling and prenatal diagnosis in countries like ours where these two mutations are present. FSC 8 was shown to be associated with haplotype IV (according to Orkin's classification) and a positive recognition site for restriction enzyme Xmn at promotor region of γ_G . Therefore, in the differentiation of these two mutations, searching for an Xmn recognition site or using allele-specific oligoprimers with a 3 end matching with the normal sequence of codon 8 would be helpful.

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This study was partly supported by a grant from the Turkish Scientific and Technical Research Foundation Tag 0757.

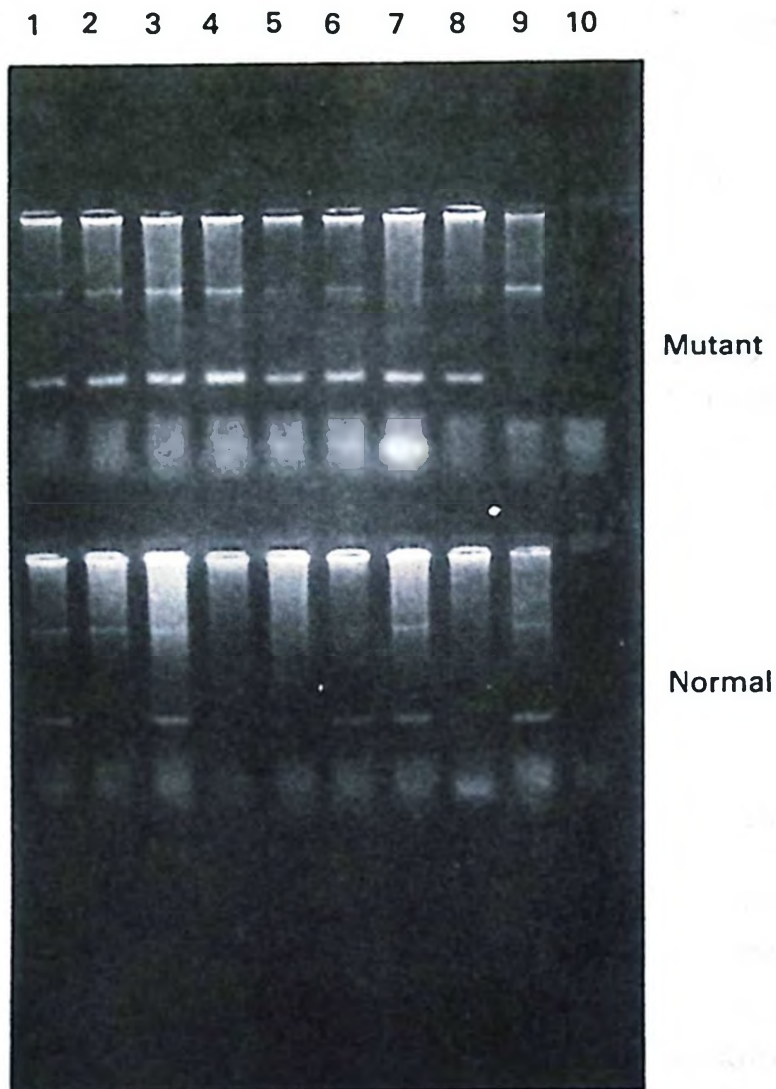


Fig. 1: 1) FSC8 heterozygote. 2) FSC8 homozygote. 3) FSC8 heterozygote. 4) FSC89 homozygote. 5) FSC8 homozygote. 6) FSC89 heterozygote. 7) FSC89 heterozygote. 8) FSC8 homozygote. 9) Control. 10) Blank.

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