

RFLP analysis

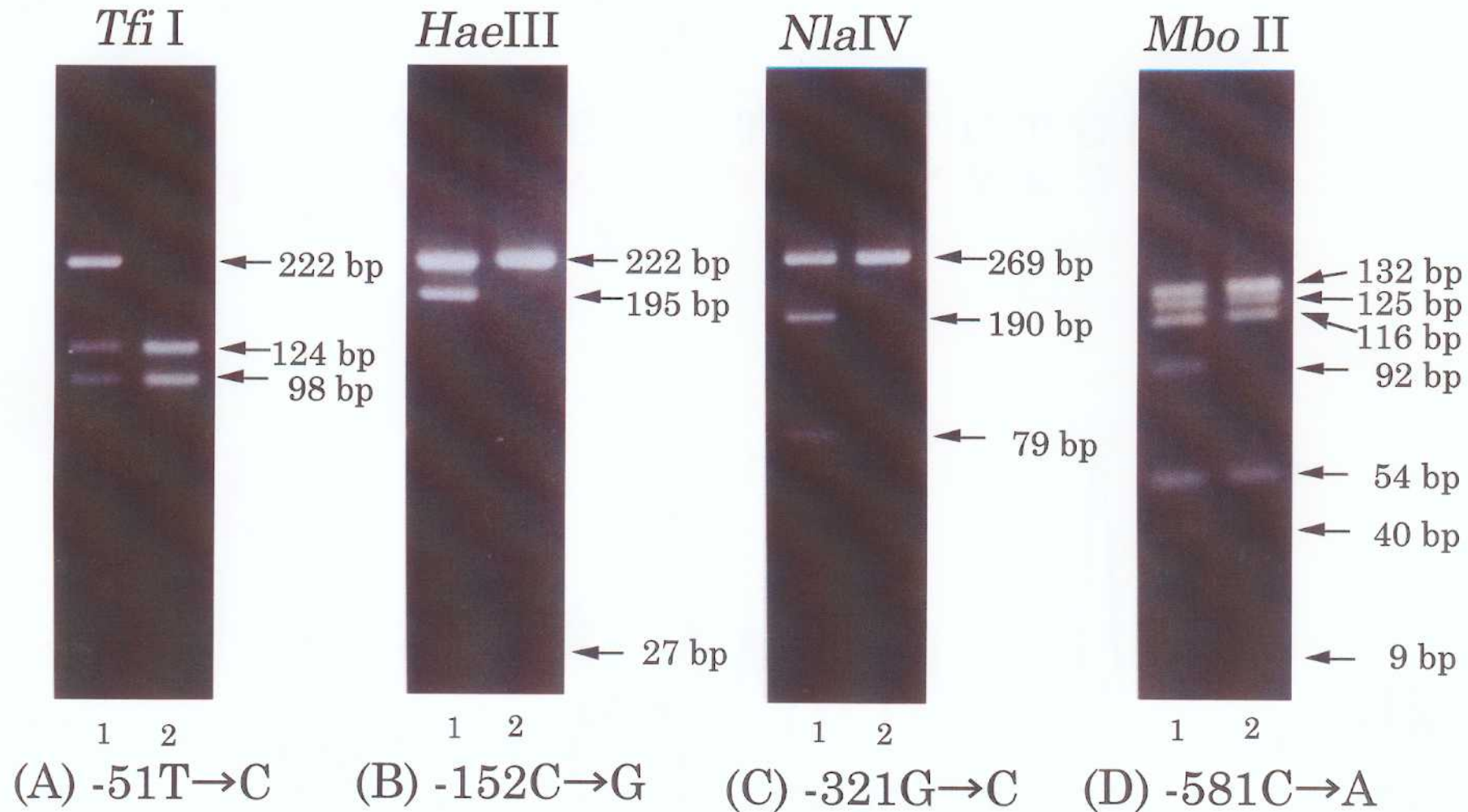


Fig. 1. Figure demonstrates RFLP patterns of PCR products generated with primer sets of 5HT1A-DF and 5HT1A-DR (A), 5HT1A-DF and 5HT1A-DR (B), 5HT1A-CF and 5HT1A-CR (C), and 5HT1A-BF and 5HT1A-BR (D). In the absence of the mutated site, the PCR fragment of 222 bp was cut producing fragments of 124 and 98 bp by *Tfi* I (A). In the presence of the mutated site, each PCR fragment of 222, 269 and 132 bp was cut producing fragments of 195 and 27 bp, 190 and 79 bp, and 92 and 40 bp by *Hae* III (B), *Nla* IV (C) and *Mbo* II (D), respectively. The digested products were visualized by electrophoresis in 3 % agarose gel containing ethidium bromide. Sample 1 shows heterozygous genotype, and sample 2 shows homozygous genotype of the wild type.

Sequencing

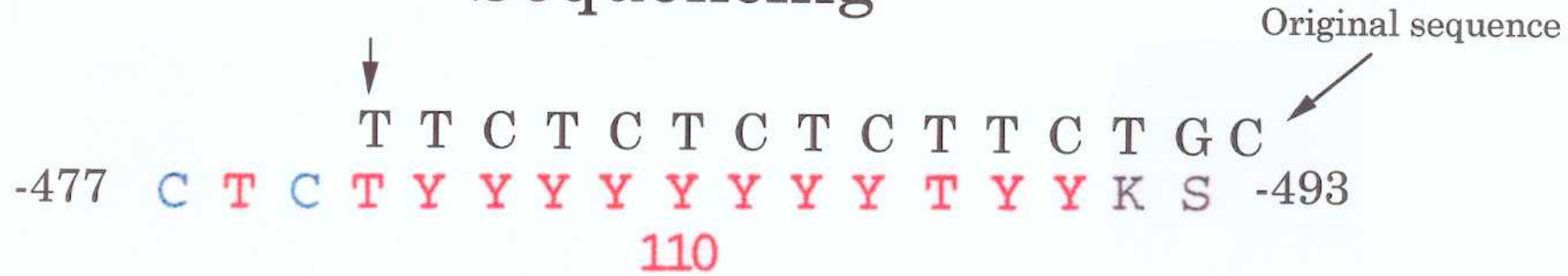


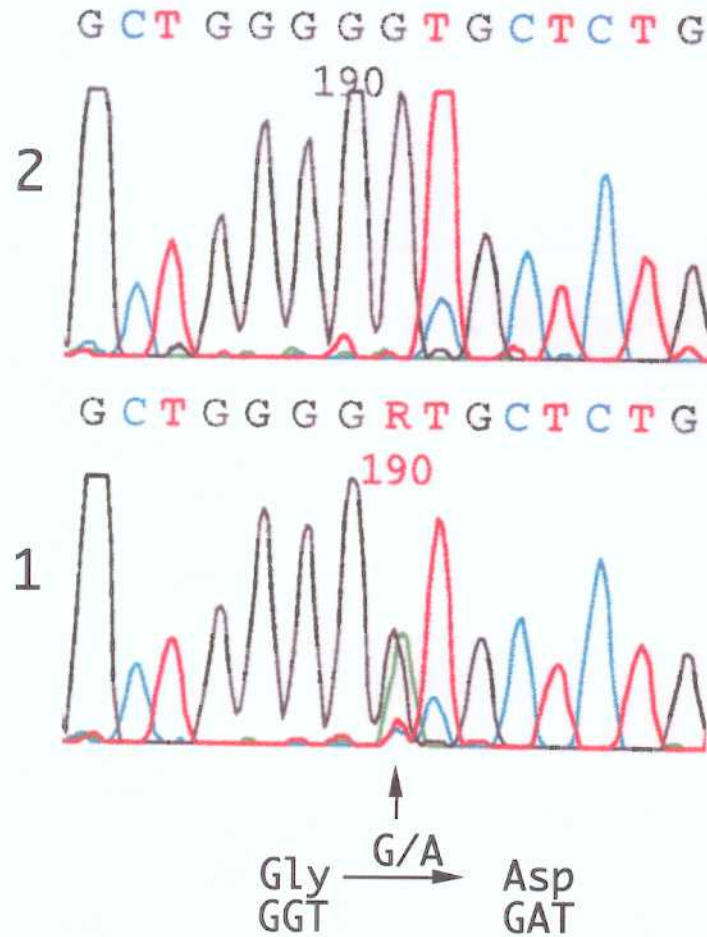
Fig. 2. PCR direct sequencing of PCR product generated with the primer set of 5HT1A-BF and 5HT1A-BR. PCR direct sequencing was performed as described in text. Shown here is reverse sequencing of the nucleotide numbers from -477 to -493. Although original reverse sequence from -477 to -494 is 5'-CTCTTCTCTCTTCTGC, the sequence from -477 to -493 in this sample is 5'-CTCTYYYYYYYYYYKYS (Y=C or T, K=G or T, S=G or C) due to a single base (T) deletion at the position of nucleotide -480. The vertical arrow indicates the position of a single base (T) deletion. This schizophrenic patient was a -480delA/A heterozygote.

SSCP analysis



1 2

Sequencing



RFLP analysis

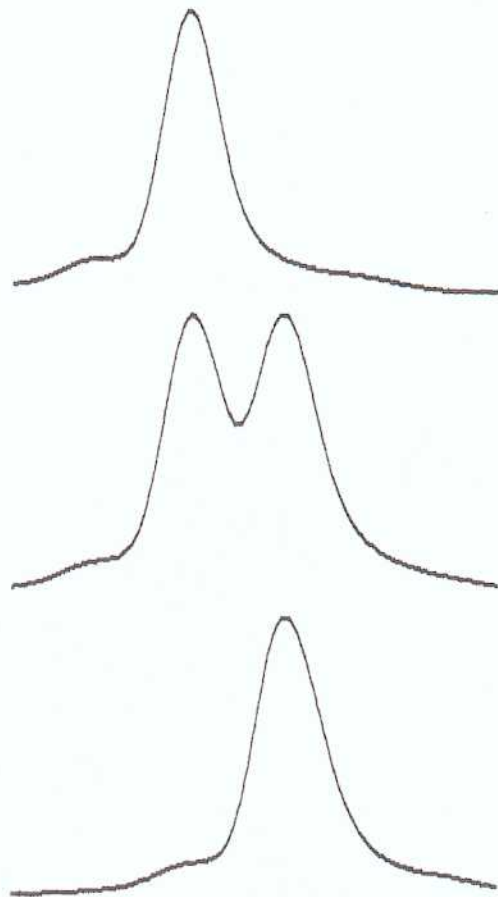


← 306 bp
← 248 bp
← 168 bp
← 80 bp
← 58 bp

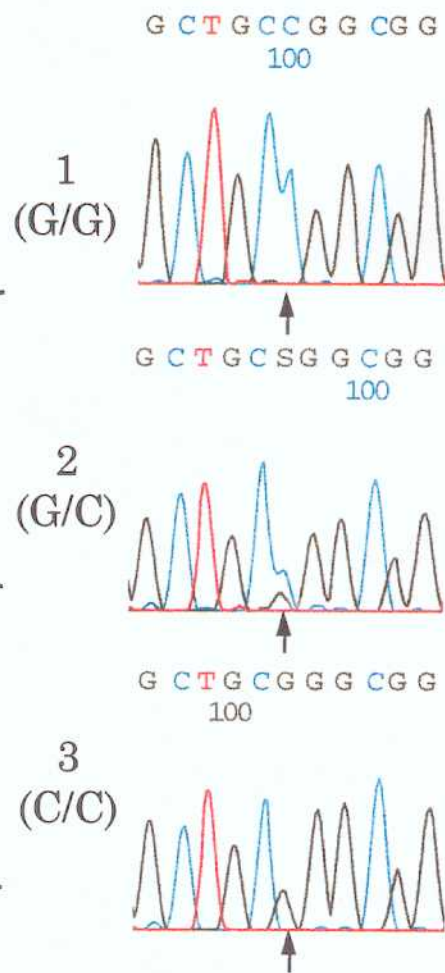
1 2

Fig. 3. SSCP analysis, direct sequencing, and RFLP analysis of PCR products generated with the primer set of 5HT1A-HF and 5HT1A-HR. The vertical arrow of sequencing indicates the position of 815G→A variant. In the presence of the mutated site, the PCR fragment of 248 bp was cut producing fragments of 168 and 80 bp by *Fok* I. The digested products were visualized by electrophoresis in 3 % agarose gel containing ethidium bromide. Sample 1 shows heterozygous genotype of the 815G→A variant, and sample 2 shows homozygous wild-type.

SSCP analysis



Sequencing



RFLP analysis

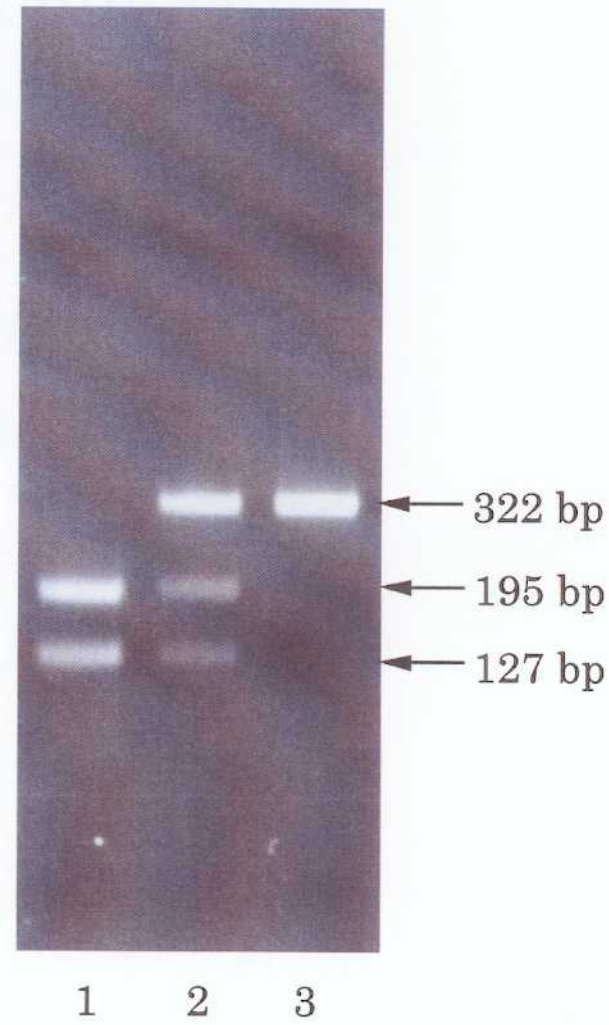
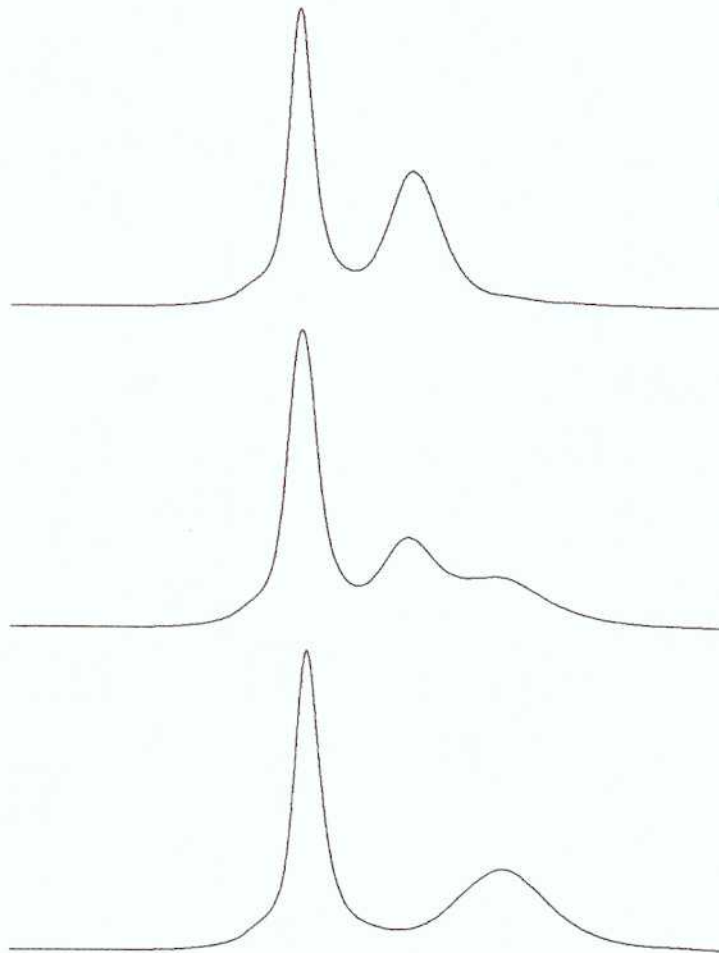


Fig. 4. SSCP analysis, direct sequencing, and RFLP analysis of PCR products generated using the AP2STRPF and AP2STRPR primer set. Shown here is reverse sequencing. A single base pair substitution (G→C) at the nucleotide position -90 was detected by sequencing (S=C or G). The vertical arrows indicate the positions of nucleotide substitution. In absence of the mutated site, the 322 bp PCR product was cut by *Msp* I, producing fragments of 195 and 127 bp. The digested products were visualized by electrophoresis in 2 % agarose gel containing ethidium bromide. Samples 1, 2 and 3 indicate homozygous (G/G), heterozygous (G/C), and homozygous (C/C) genotypes at the -90 locus, respectively.

SSCP analysis



Sequencing

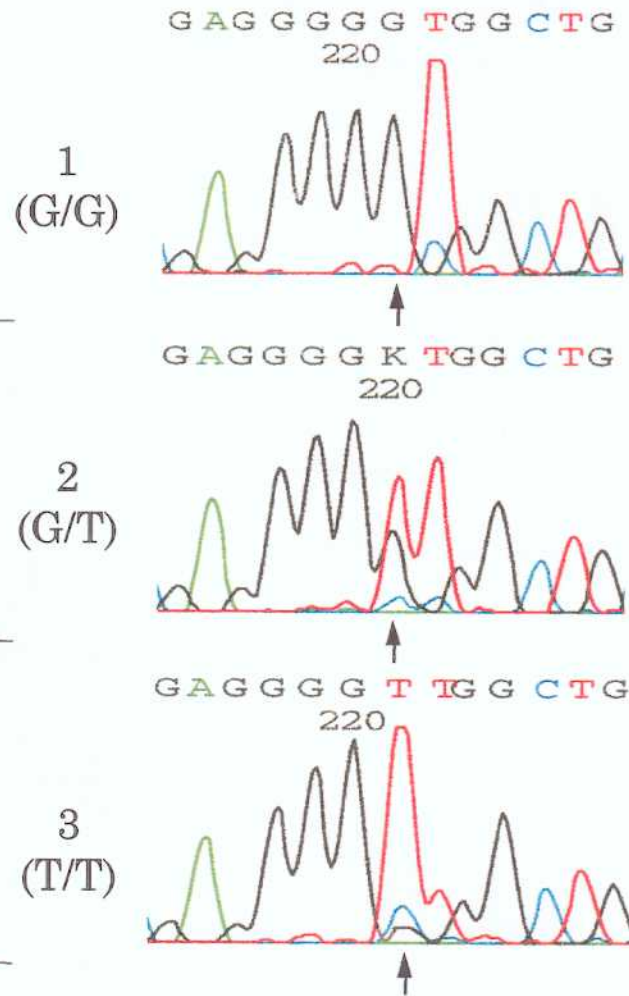
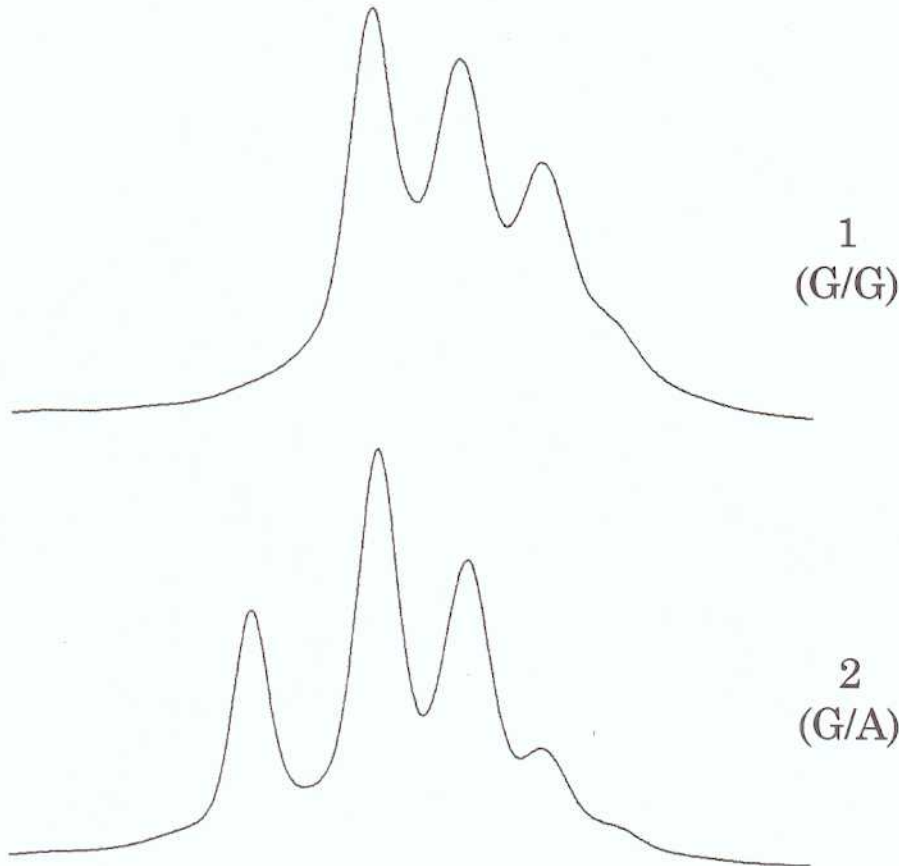


Fig. 5. SSCP analysis and direct sequencing of PCR products generated using the AP2PMF and AP2PMR primer set. A single base pair substitution (G→T) at the nucleotide position -803 was detected by sequencing (K=G or T). The vertical arrows indicate the positions of nucleotide substitution. Samples 1, 2 and 3 indicate homozygous (G/G), heterozygous (G/T), and homozygous (T/T) genotypes at the -803 locus, respectively.

SSCP analysis



Sequencing

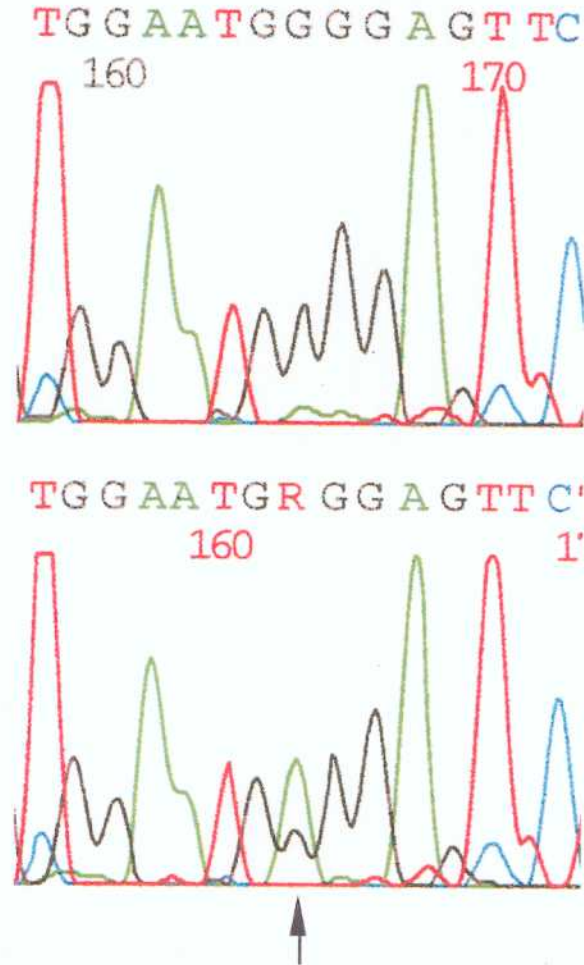
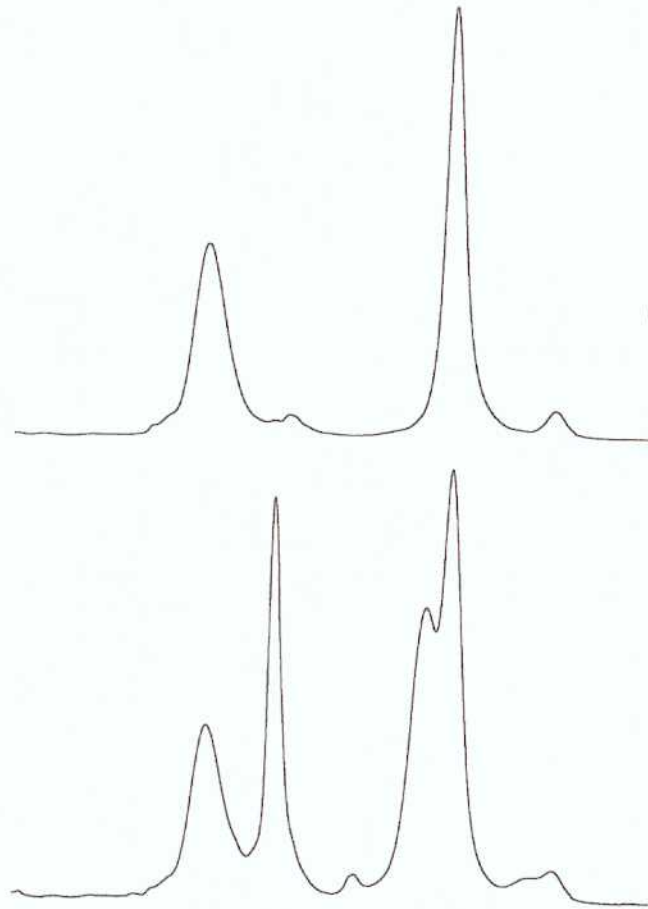


Fig. 6. SSCP analysis and direct sequencing of PCR products generated with the primer set of AP2PUF and AP2PUR. The vertical arrow indicates the position of a single base pair substitution (G→A) at the nucleotide position-1769 (R= G or A). Sample 1 corresponds homozygous wild-type, and sample 2 is heterozygous genotype of the -1769G→A variant.

SSCP analysis



1
(T/T)

2
(T/C)

Sequencing

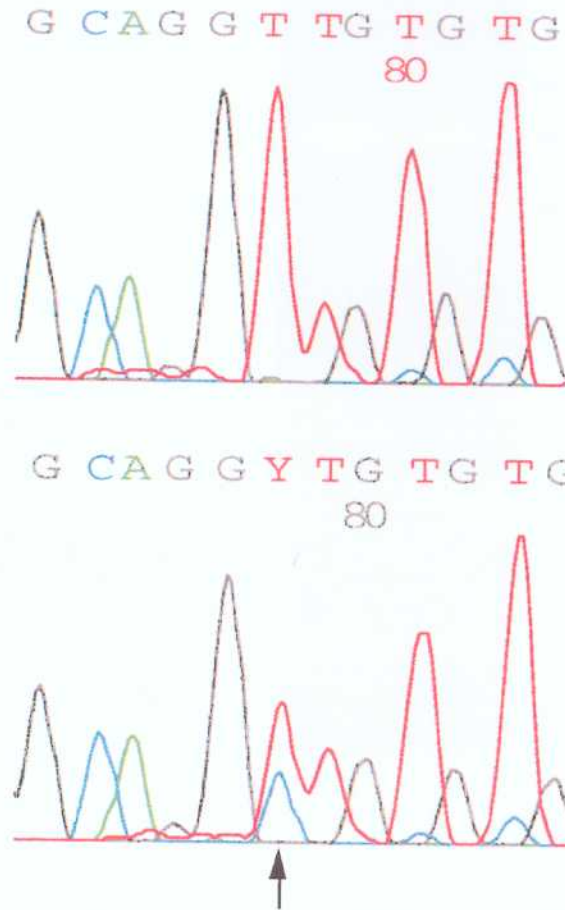
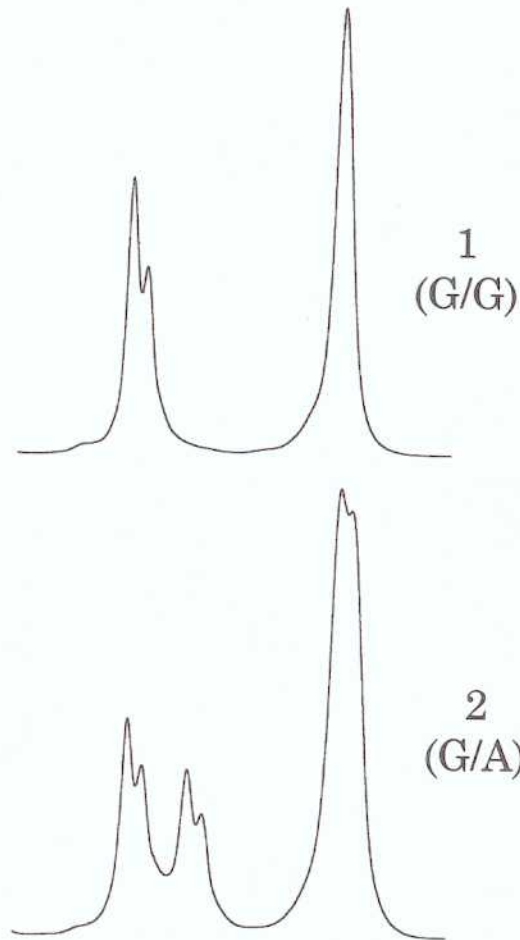
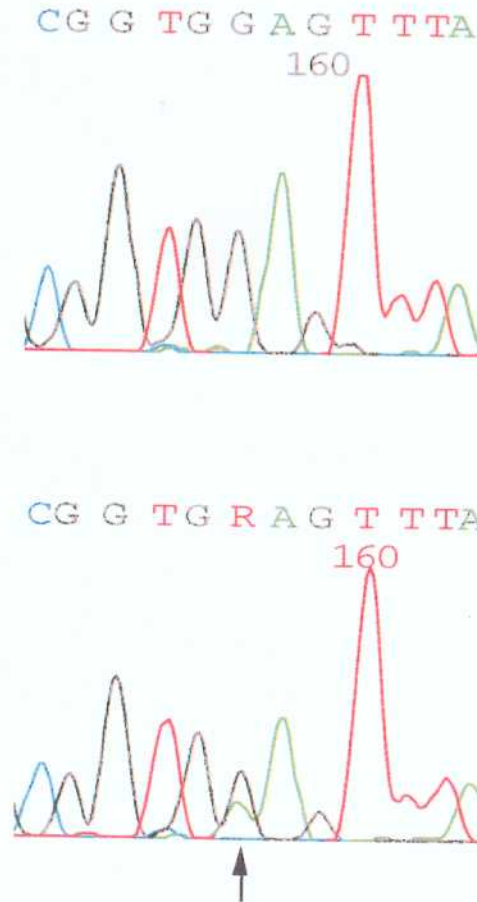


Fig. 7. SSCP analysis and direct sequencing of PCR products generated with the primer set of CREB-UF and CREB-UR. The vertical arrow indicates the position of a single base pair substitution (T→C) at the nucleotide position-933 (Y=C or T). Sample 1 shows the homozygous wild-type, and sample 2 shows the heterozygous genotype of the -933T→C variant.

SSCP analysis



Sequencing



RFLP analysis

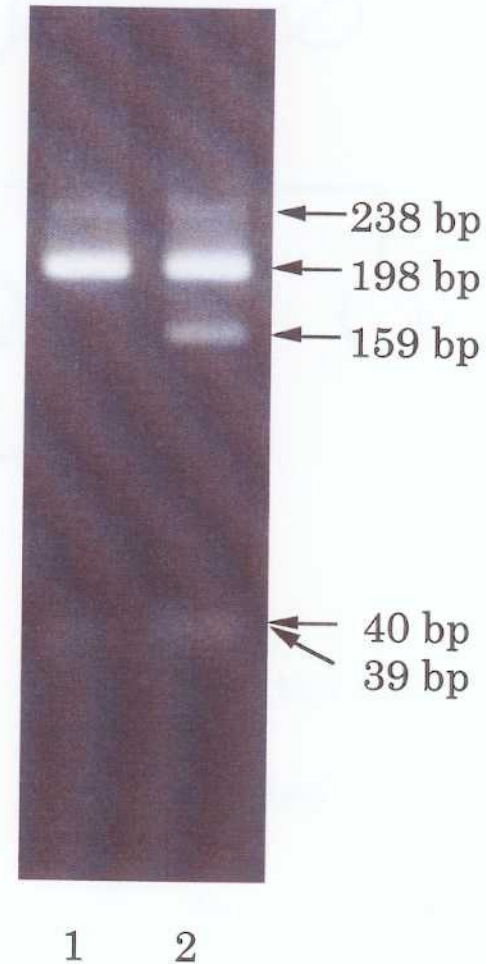


Fig. 8. SSCP analysis, direct sequencing, RFLP analysis of PCR products generated with the primer set of CREB-MF and CREB-MR. The vertical arrow indicates the position of a single base pair substitution (G→A) at the nucleotide position-413 (R=A or G). In the presence of mutated site, the PCR fragment of 198 bp was cut producing fragments of 159 and 39 bp by *Hph* I. The digested products were visualized by electrophoresis in 3 % agarose gel containing ethidium bromide. Sample 1 shows the homozygous wild-type, and sample 2 shows the heterozygous genotype of the -413G→A variant.

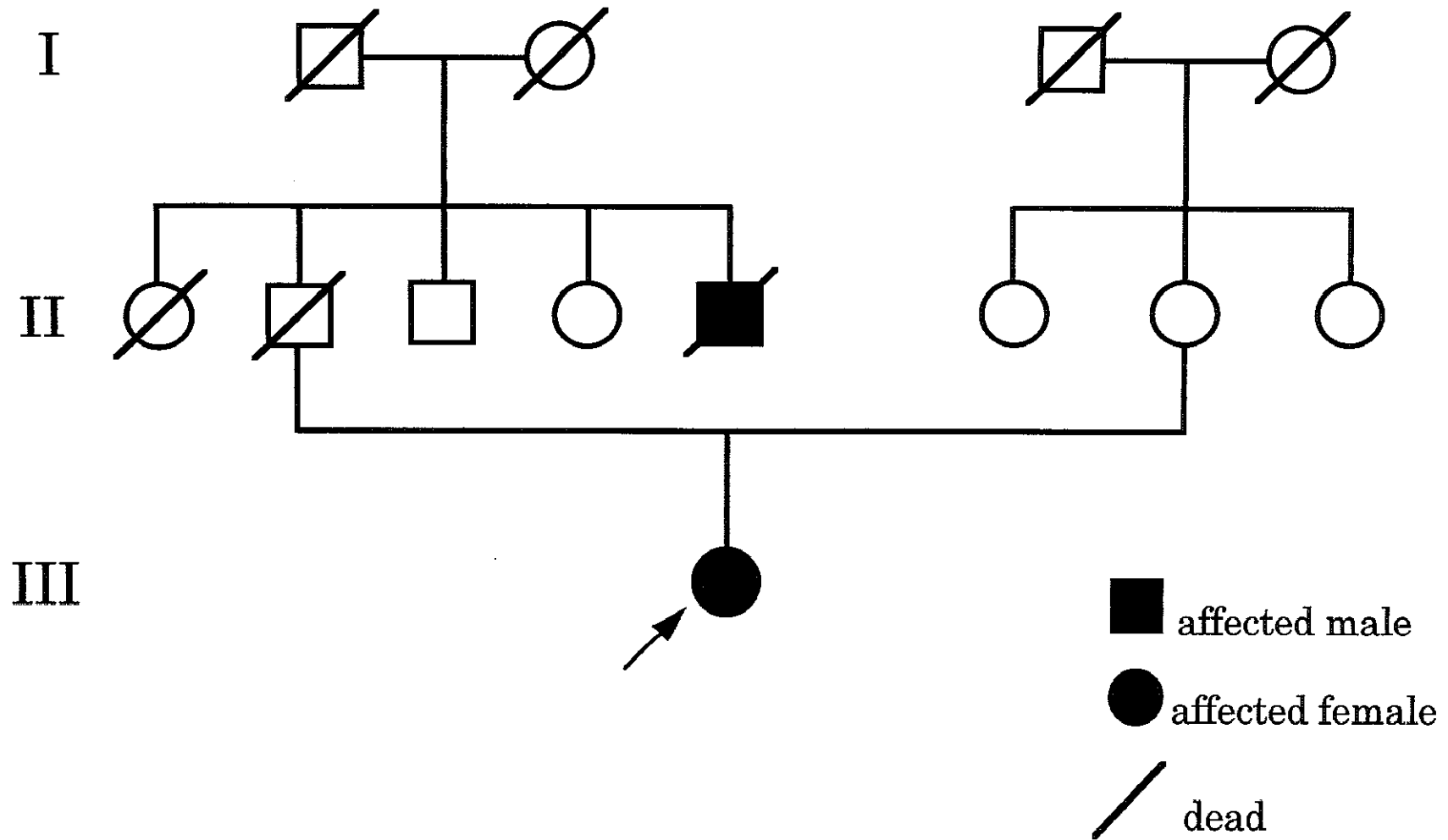
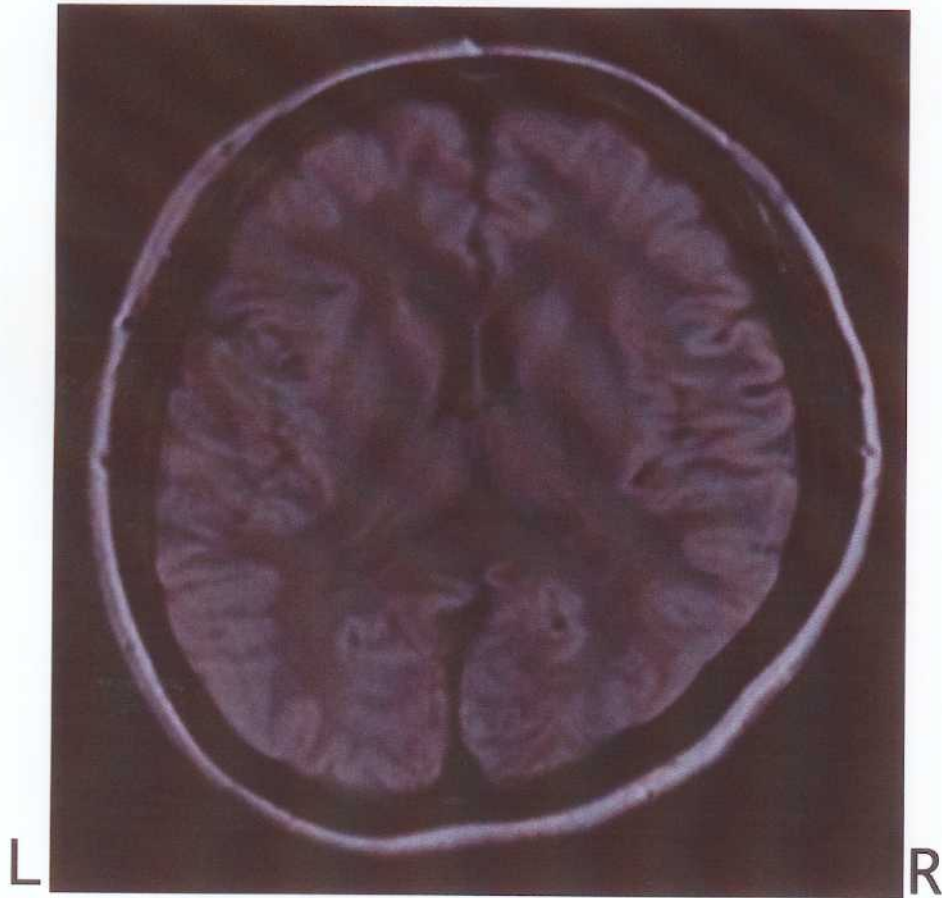


Fig. 9. Figure shows pedigree of the family of the patient with the -933T→C variant. The arrow indicates the female patient with the -933T→C variant. Her uncle was affected with schizophrenia.

MRI



EEG

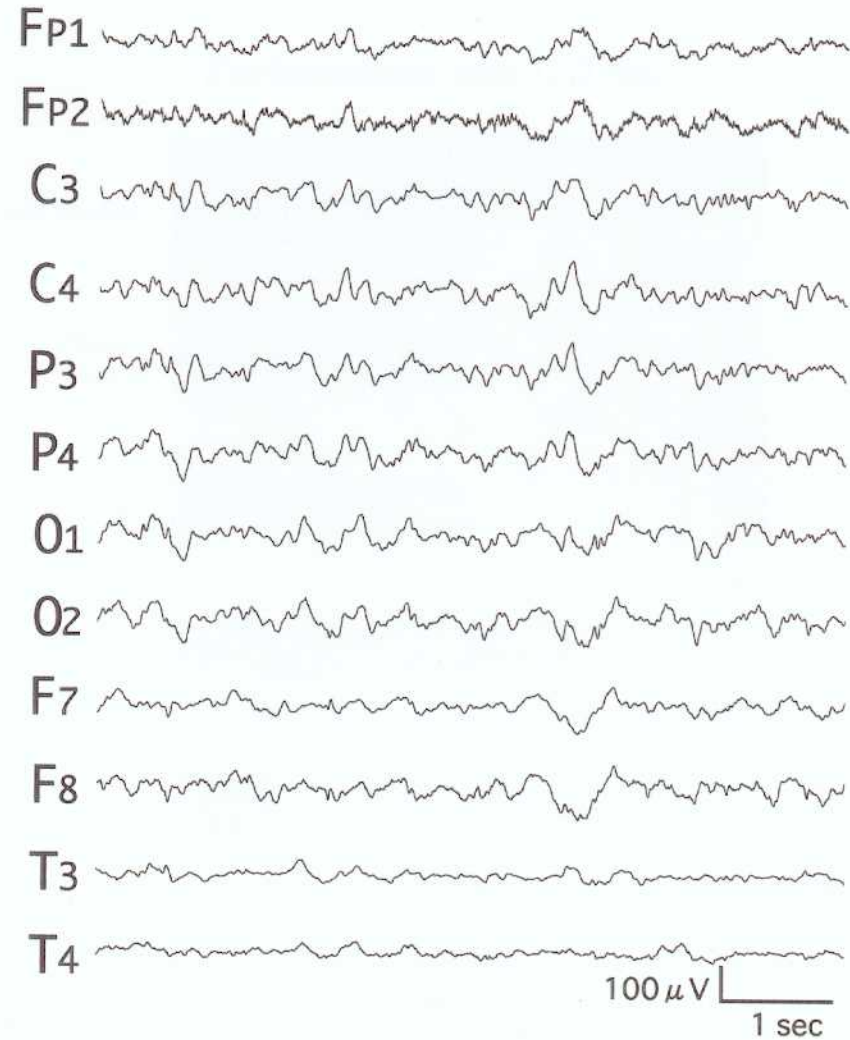


Fig. 10. Figure shows MRI and EEG of the patient with the -933T→C variant. Note that while MRI was within normal limits, EEG revealed abnormalities in the slow-wave; moderate voltage 5~6 per second slow activity with occasional high voltage 2~3 per second slow waves in all areas.

WAIS-R (full-scale IQ 54)

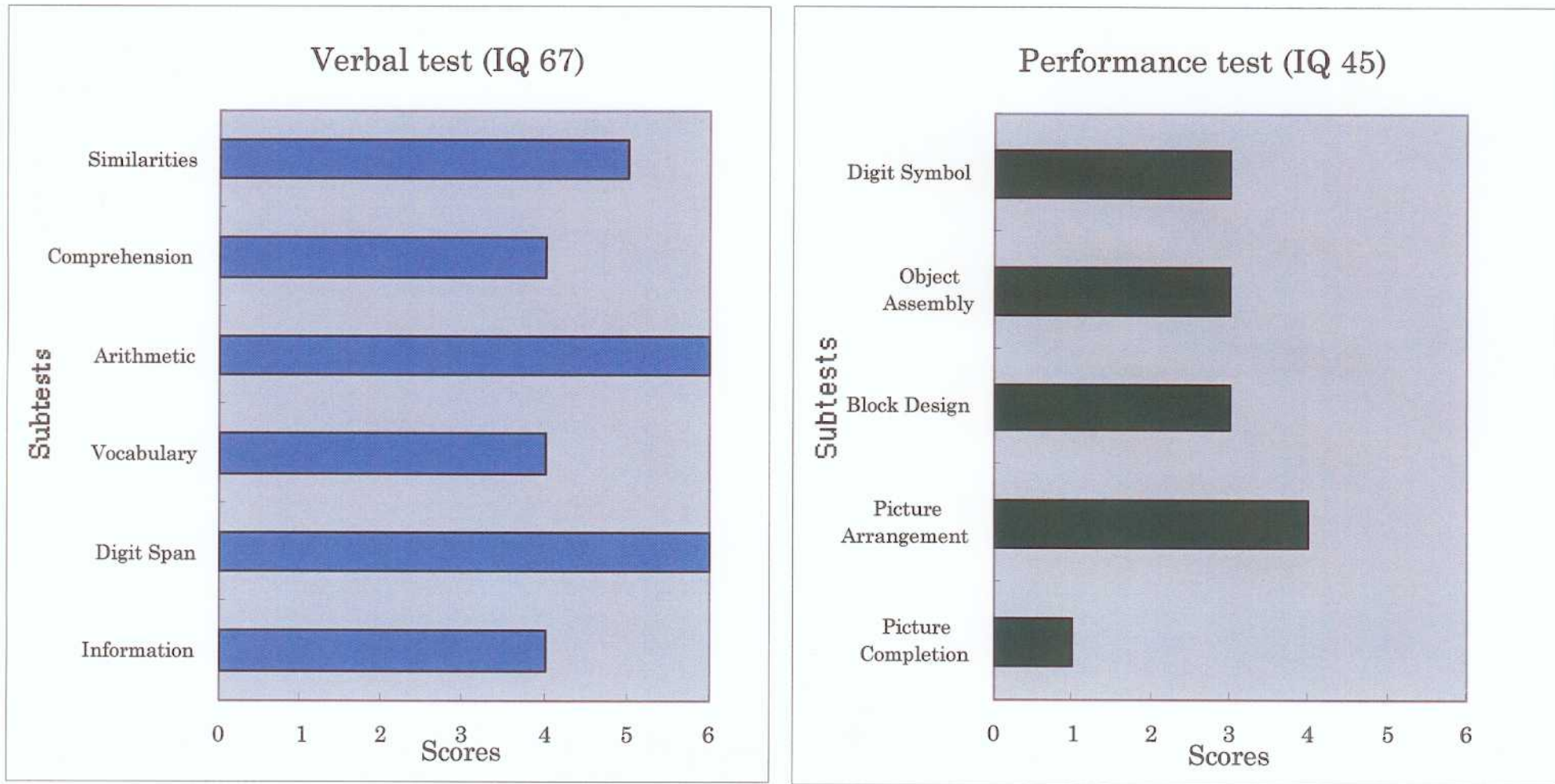


Fig. 11. Figure shows the WAIS-R testing results of the patient with the -933T→C variant. Note that while the test results indicate a lowered intelligence level in general, the deficits in performance in the subtests [Information, Vocabulary, Comprehension, and Picture Completion] are more pronounced than those found in the subtests [Digit Span and Arithmetic]. The former subtests reflect the capacity to accumulate or retain knowledge and concepts in long-term memory, while the latter subtests reflect short-term memory performance.