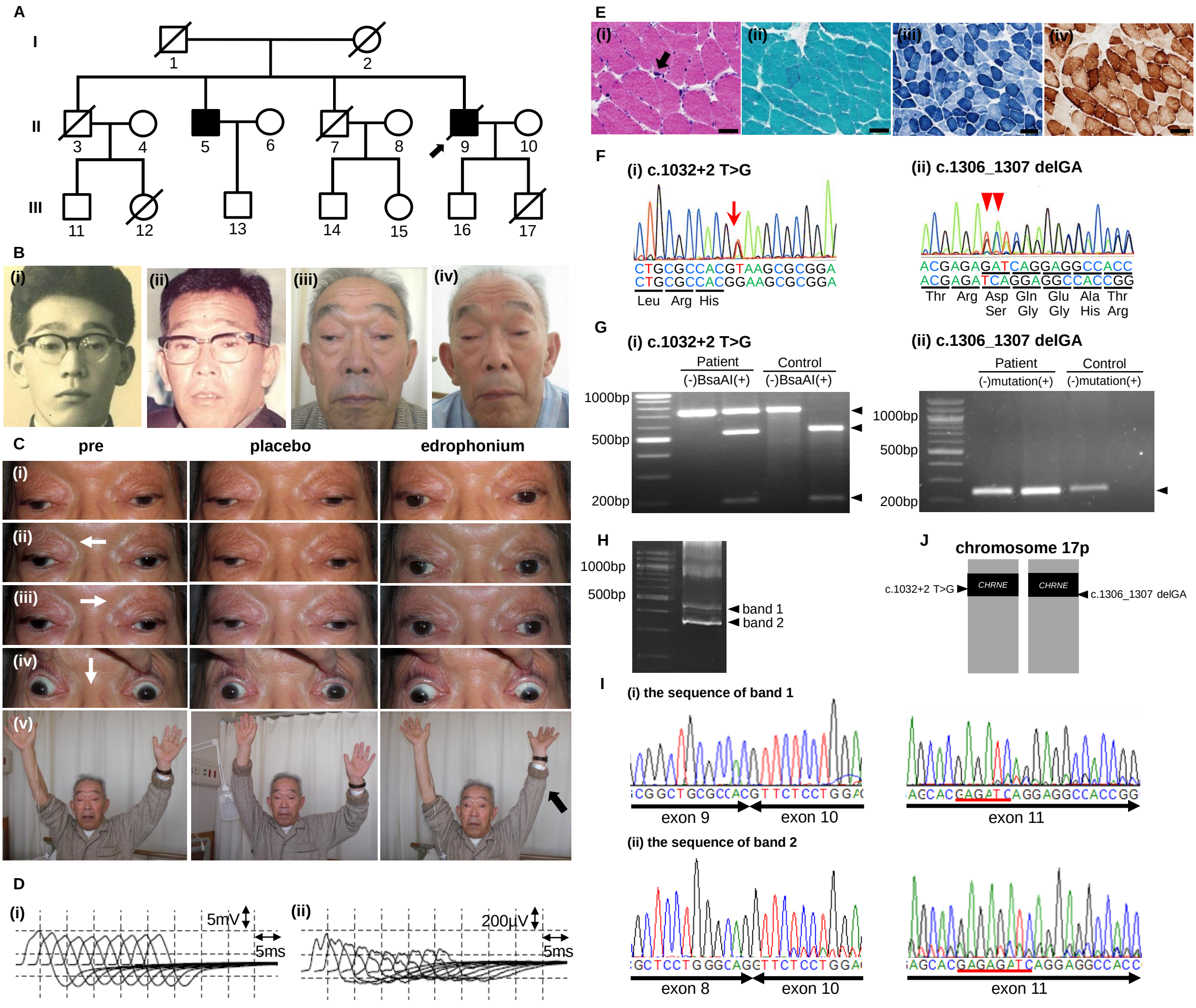
Fig. 1


A
I
II
III
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
B
(i)
(ii)
(iii)
(iv)
C
pre
placebo
edrophonium
(i)
(ii)
(iii)
(iv)
(v)
D
(i)
5mV
5ms
(ii)
200μV
5ms
E
(i)
(ii)
(iii)
(iv)
F
(i) c.1032+2 T>G
CTGCGCCACGTAAGCGCGGA
CTGCGCCACGGAAGCGCGGA
Leu Arg His
(ii) c.1306_1307 delGA
ACGAGAGATCAGGAGGCCACC
ACGAGATCAGGAGGCCACCGG
Thr Arg Asp Gln Glu Ala Thr
Ser Gly Gly His Arg
G
(i) c.1032+2 T>G
Patient
Control
(-)BsaAI(+)
(-)BsaAI(+)
1000bp
500bp
200bp
(ii) c.1306_1307 delGA
Patient
Control
(-)mutation(+)
(-)mutation(+)
1000bp
500bp
200bp
H
1000bp
500bp
band 1
band 2
J
chromosome 17p
c.1032+2 T>G
CHRNE
CHRNE
c.1306_1307 delGA
I
(i) the sequence of band 1
CGGCTGCGCCACGTTCTCCTGGA
exon 9
exon 10
AGCACGAGATCAGGAGGCCACCGG
exon 11
(ii) the sequence of band 2
GCTCCTGGGCAGGTTCTCCTGGA
exon 8
exon 10
AGCACGAGAGATCAGGAGGCCACC
exon 11