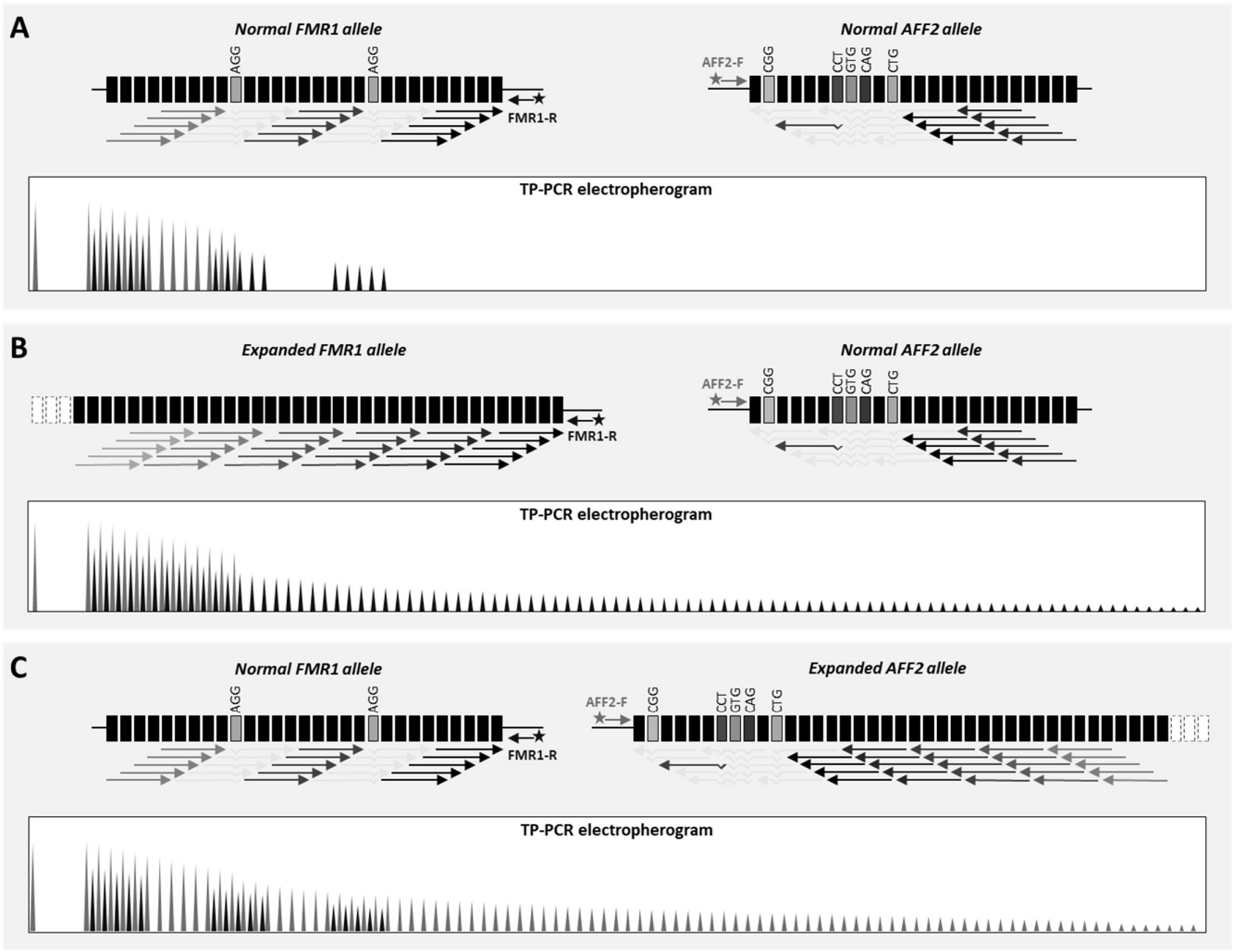

A
Normal FMR1 allele
AGG
AGG
FMR1-R
Normal AFF2 allele
AFF2-F
CGG
CCT
GTG
CAG
CTG
TP-PCR electropherogram
B
Expanded FMR1 allele
FMR1-R
Normal AFF2 allele
AFF2-F
CGG
CCT
GTG
CAG
CTG
TP-PCR electropherogram
C
Normal FMR1 allele
AGG
AGG
FMR1-R
Expanded AFF2 allele
AFF2-F
CGG
CCT
GTG
CAG
CTG
TP-PCR electropherogram

**Figure 1**

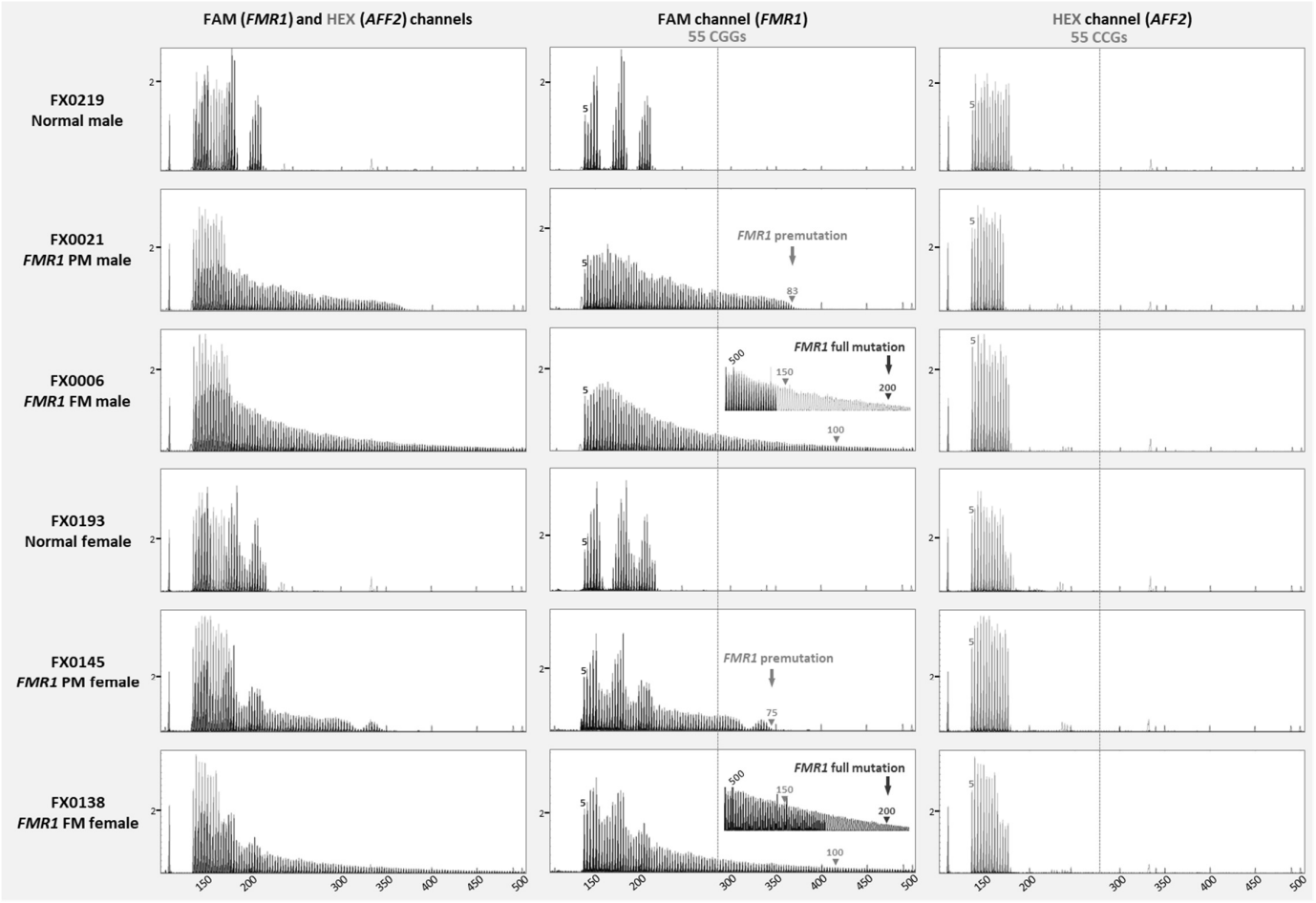

FAM (FMR1) and HEX (AFF2) channels
FAM channel (FMR1)
55 CGGs
HEX channel (AFF2)
55 CCGs
FX0219
Normal male
FX0021
FMR1 PM male
FMR1 premutation
83
FX0006
FMR1 FM male
FMR1 full mutation
500
150
200
100
FX0193
Normal female
FX0145
FMR1 PM female
FMR1 premutation
75
FX0138
FMR1 FM female
FMR1 full mutation
500
150
200
100
150
200
300
350
400
450
500

**Figure 2**

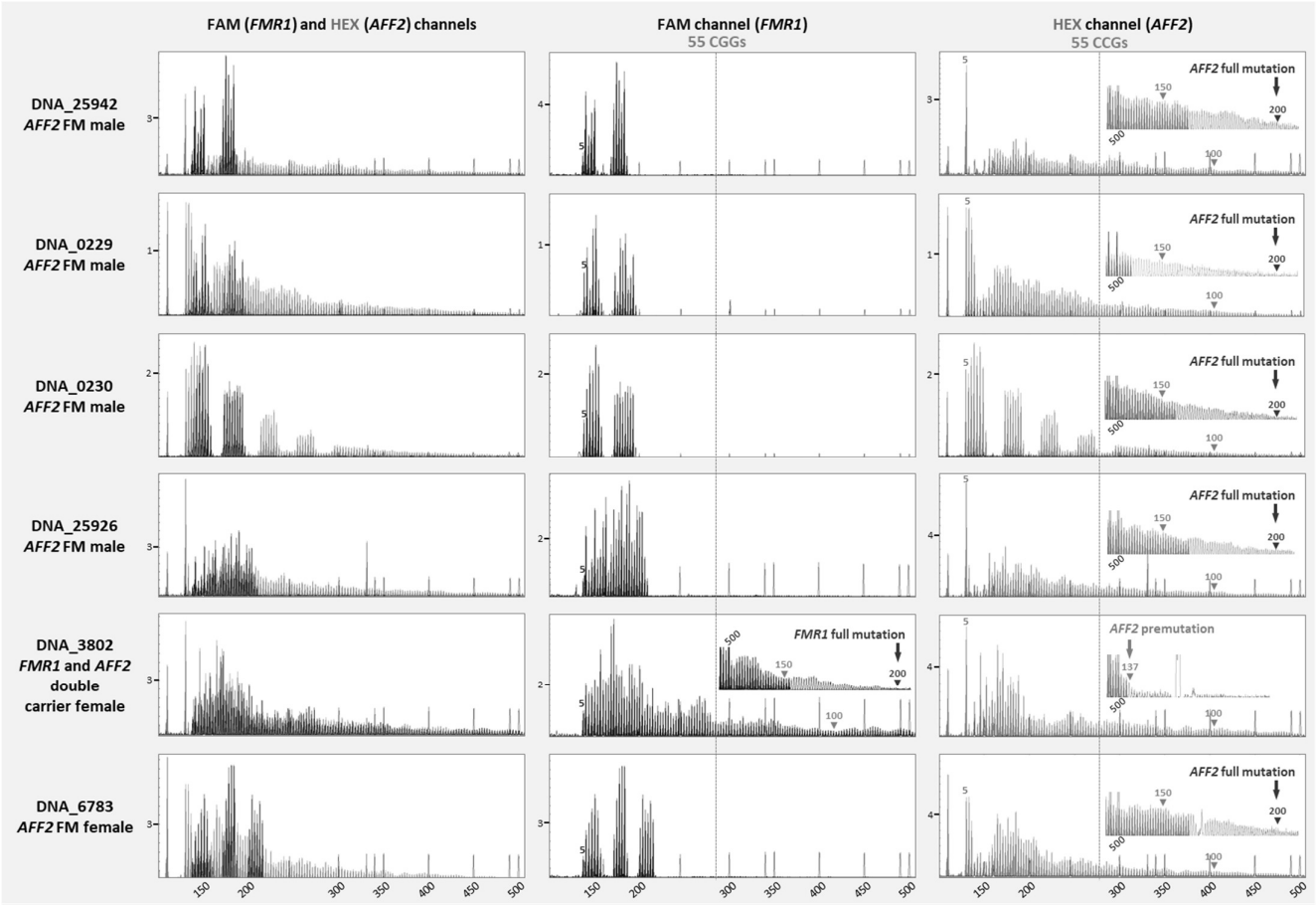

FAM (FMR1) and HEX (AFF2) channels
FAM channel (FMR1)
55 CGGs
HEX channel (AFF2)
55 CCGs
DNA_25942
AFF2 FM male
DNA_0229
AFF2 FM male
DNA_0230
AFF2 FM male
DNA_25926
AFF2 FM male
DNA_3802
FMR1 and AFF2
double
carrier female
DNA_6783
AFF2 FM female
AFF2 full mutation
FMR1 full mutation
AFF2 premutation
150
200
300
350
400
450
500
100
137

**Figure 3**

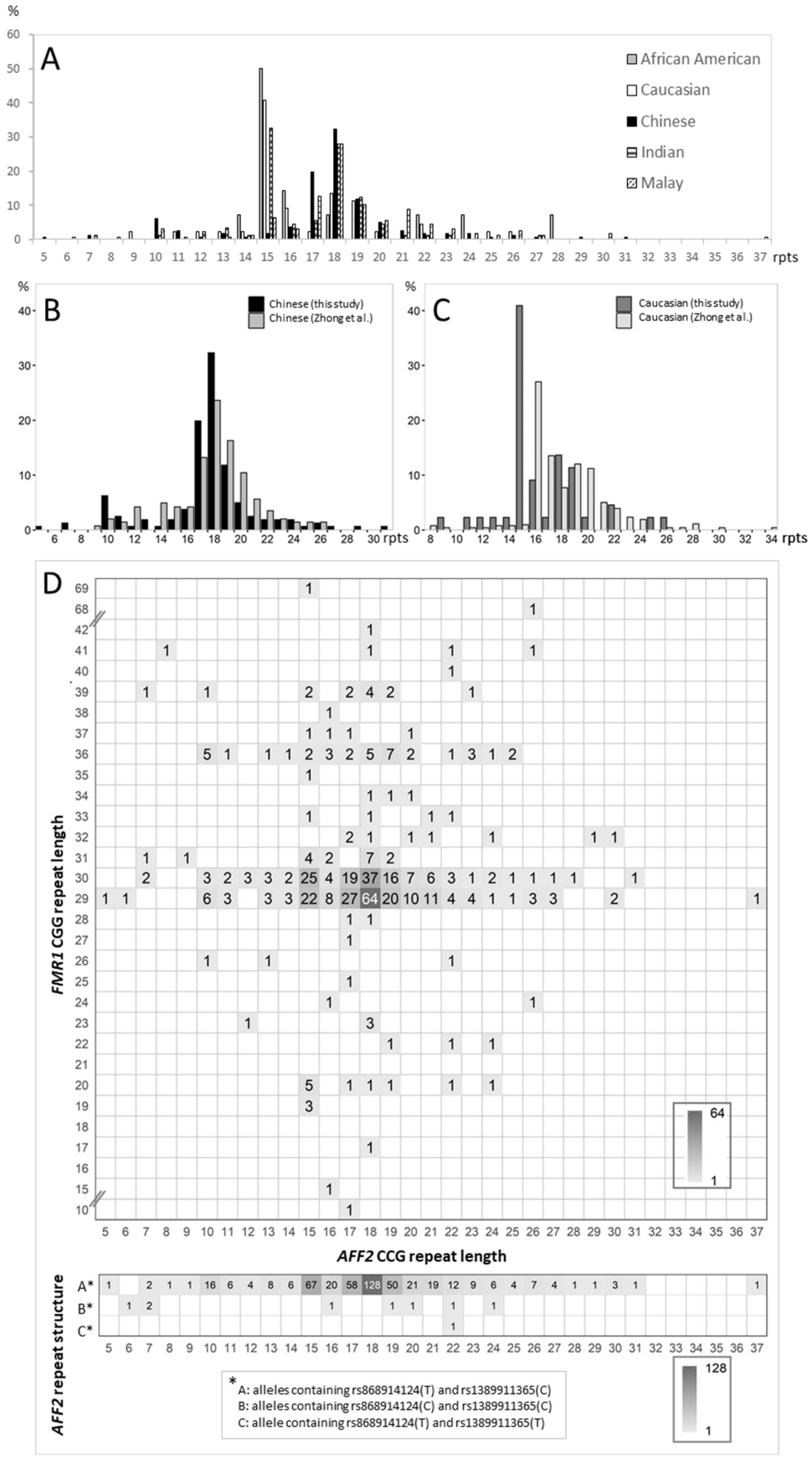

A
African American
Caucasian
Chinese
Indian
Malay
%
rpts
B
Chinese (this study)
Chinese (Zhong et al.)
C
Caucasian (this study)
Caucasian (Zhong et al.)
D
FMR1 CGG repeat length
AFF2 CCG repeat length
AFF2 repeat structure
A*
B*
C*
*A: alleles containing rs868914124(T) and rs1389911365(C)
B: alleles containing rs868914124(C) and rs1389911365(C)
C: allele containing rs868914124(T) and rs1389911365(T)

**Figure 4**

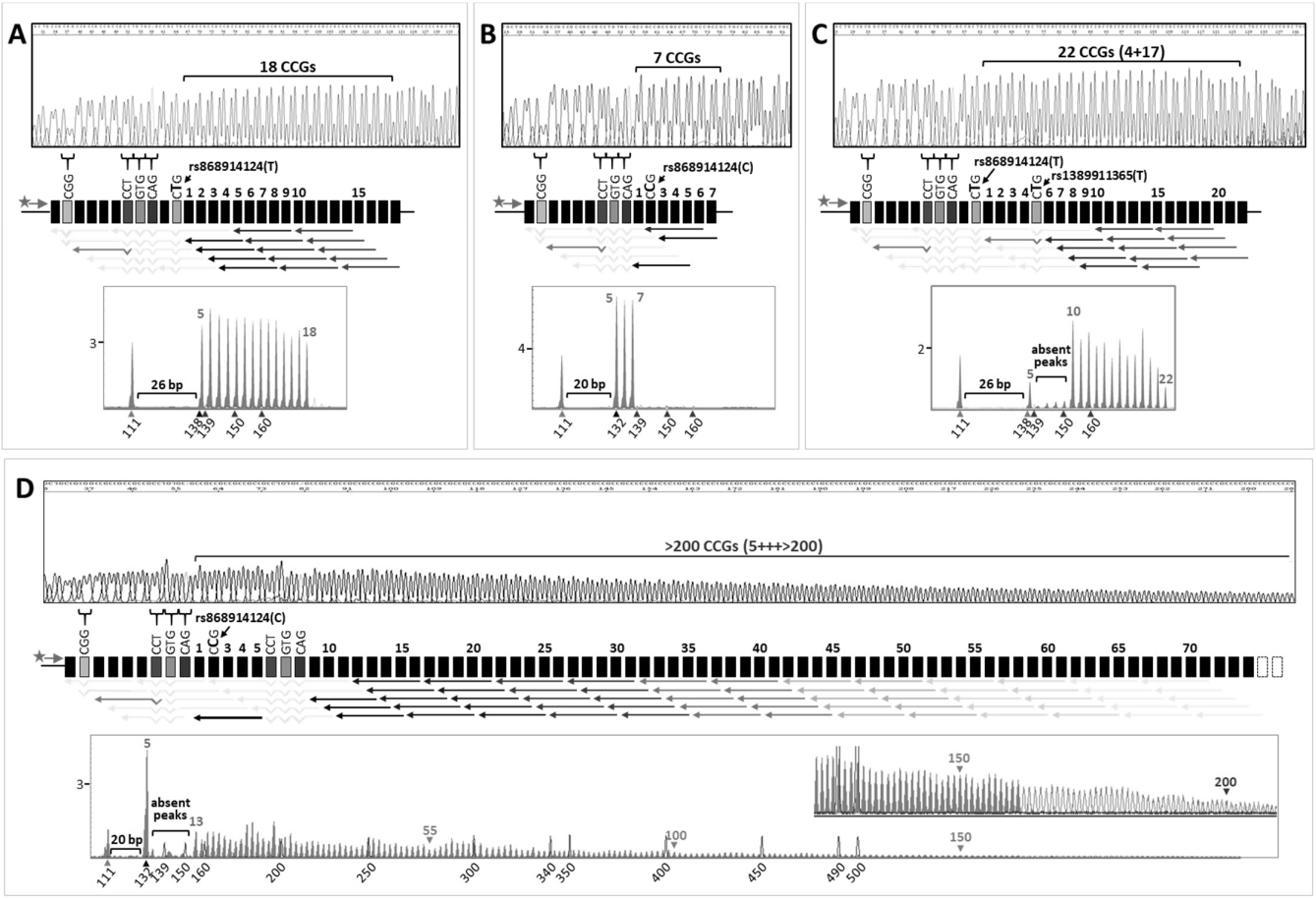

A
18 CCGs
rs868914124(T)
26 bp
B
7 CCGs
rs868914124(C)
20 bp
C
22 CCGs (4+17)
rs868914124(T)
rs1389911365(T)
absent peaks
26 bp
D
>200 CCGs (5+++>200)
rs868914124(C)
absent peaks
20 bp

**Figure 5**

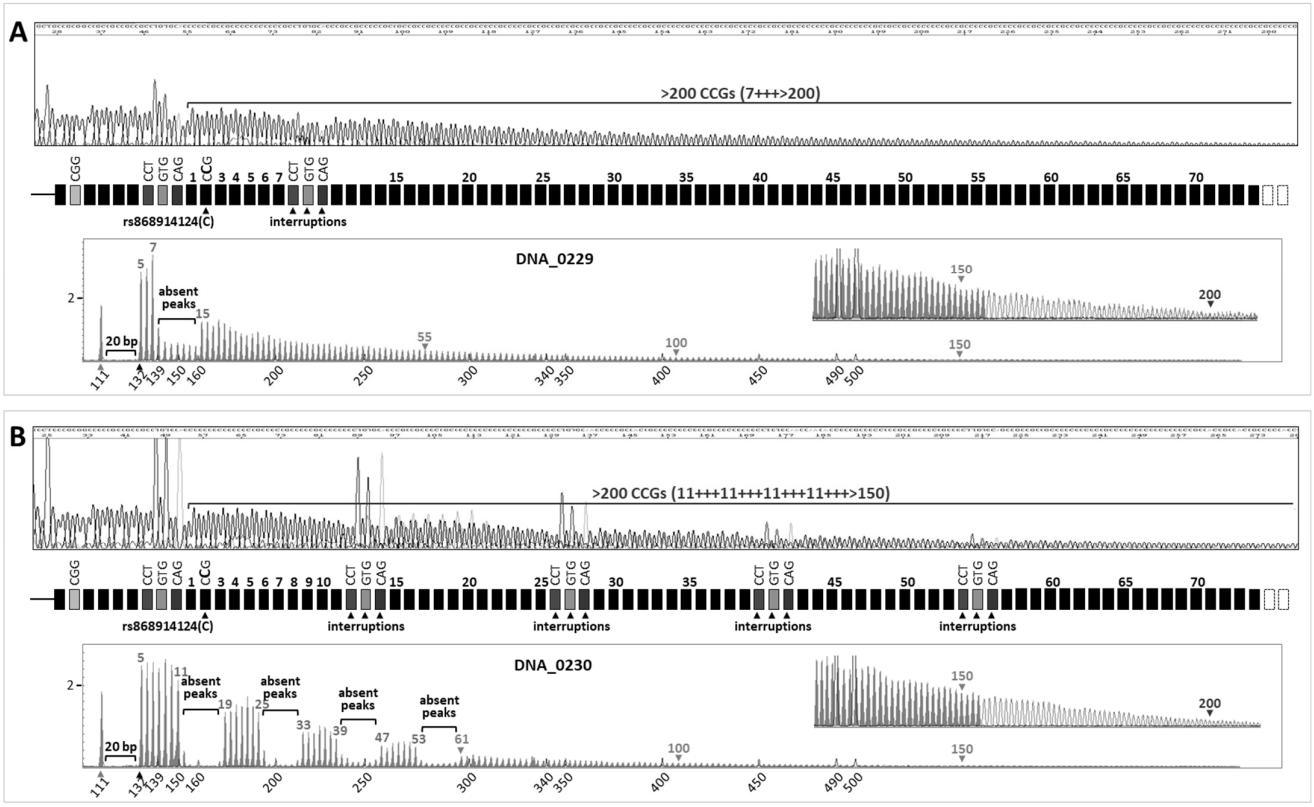

A
>200 CCGs (7+++>200)
CGG
CCT GTG CAG
CCG
CCT GTG CAG
rs868914124(C)
interruptions
DNA_0229
absent peaks
20 bp
B
>200 CCGs (11+++11+++11+++11+++>150)
rs868914124(C)
interruptions
interruptions
interruptions
interruptions
DNA_0230
absent peaks
absent peaks
absent peaks
absent peaks
20 bp

**Figure 6**

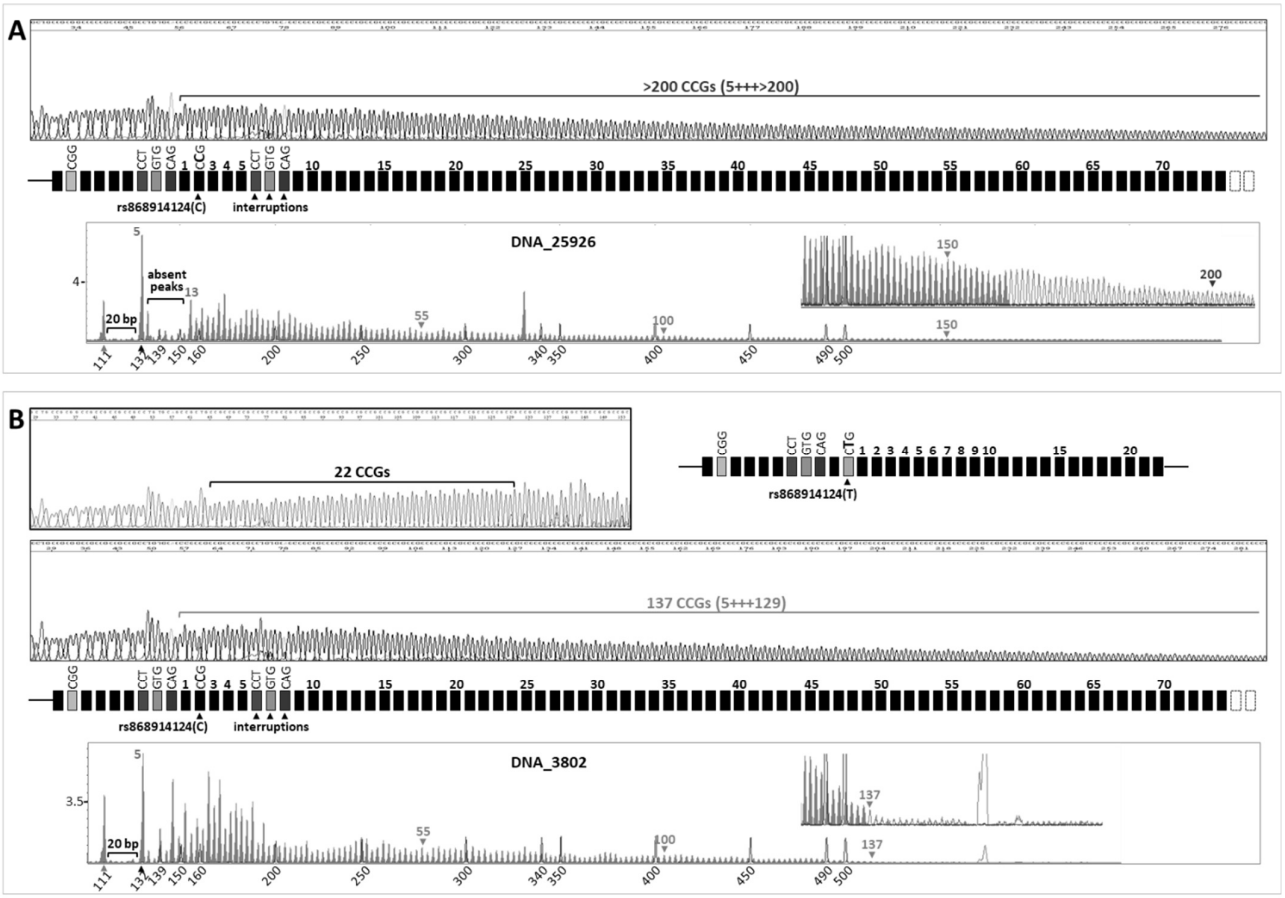

A
>200 CCGs (5+++>200)
CGG
CCT
GTG
CAG
CCG
CCT
GTG
CAG
rs868914124(C)
interruptions
DNA_25926
absent peaks
20 bp
B
22 CCGs
CTG
rs868914124(T)
137 CCGs (5+++129)
DNA_3802

**Figure 7**

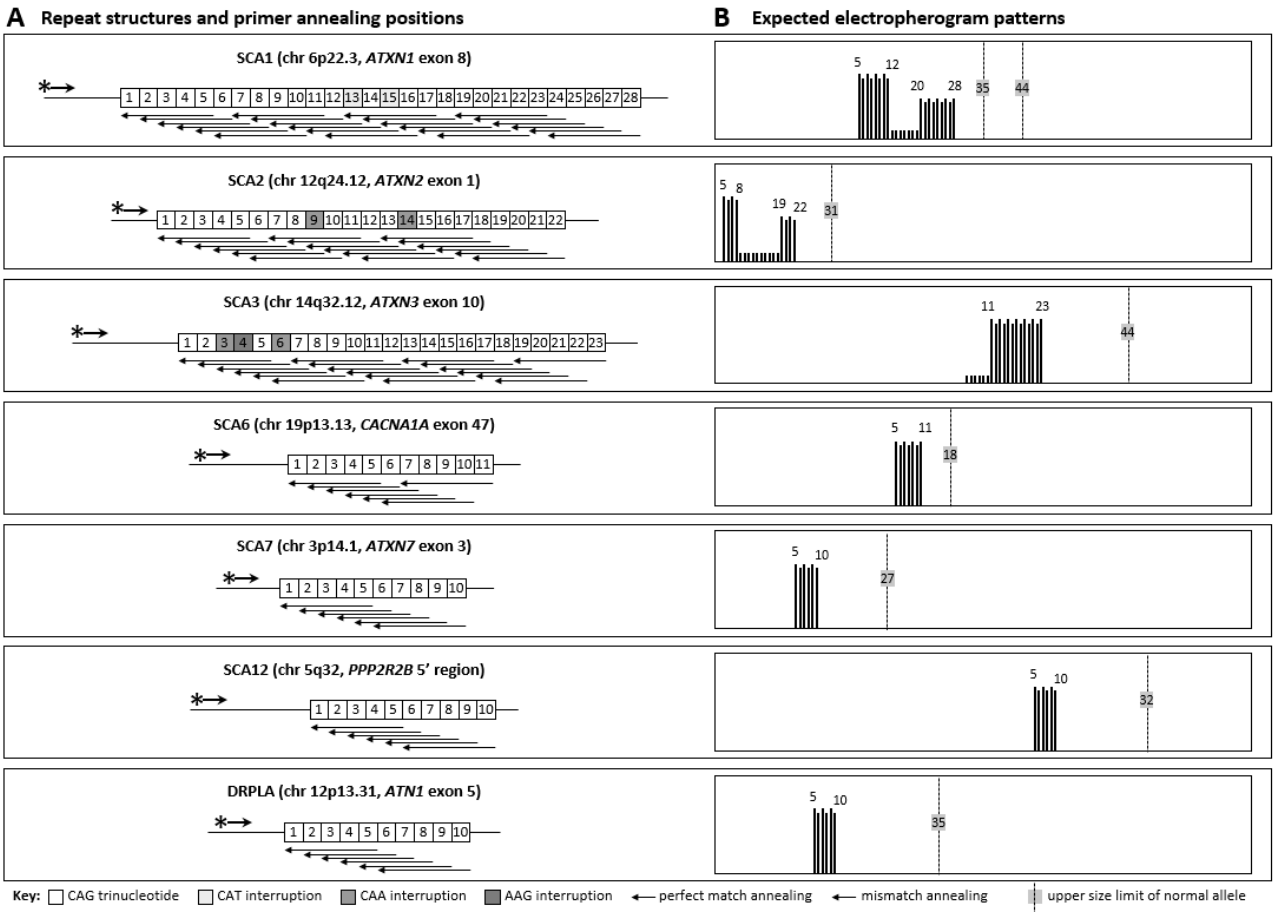

A Repeat structures and primer annealing positions
B Expected electropherogram patterns
SCA1 (chr 6p22.3, ATXN1 exon 8)
SCA2 (chr 12q24.12, ATXN2 exon 1)
SCA3 (chr 14q32.12, ATXN3 exon 10)
SCA6 (chr 19p13.13, CACNA1A exon 47)
SCA7 (chr 3p14.1, ATXN7 exon 3)
SCA12 (chr 5q32, PPP2R2B 5' region)
DRPLA (chr 12p13.31, ATN1 exon 5)
Key: CAG trinucleotide
CAT interruption
CAA interruption
AAG interruption
perfect match annealing
mismatch annealing
upper size limit of normal allele

**Figure 8**

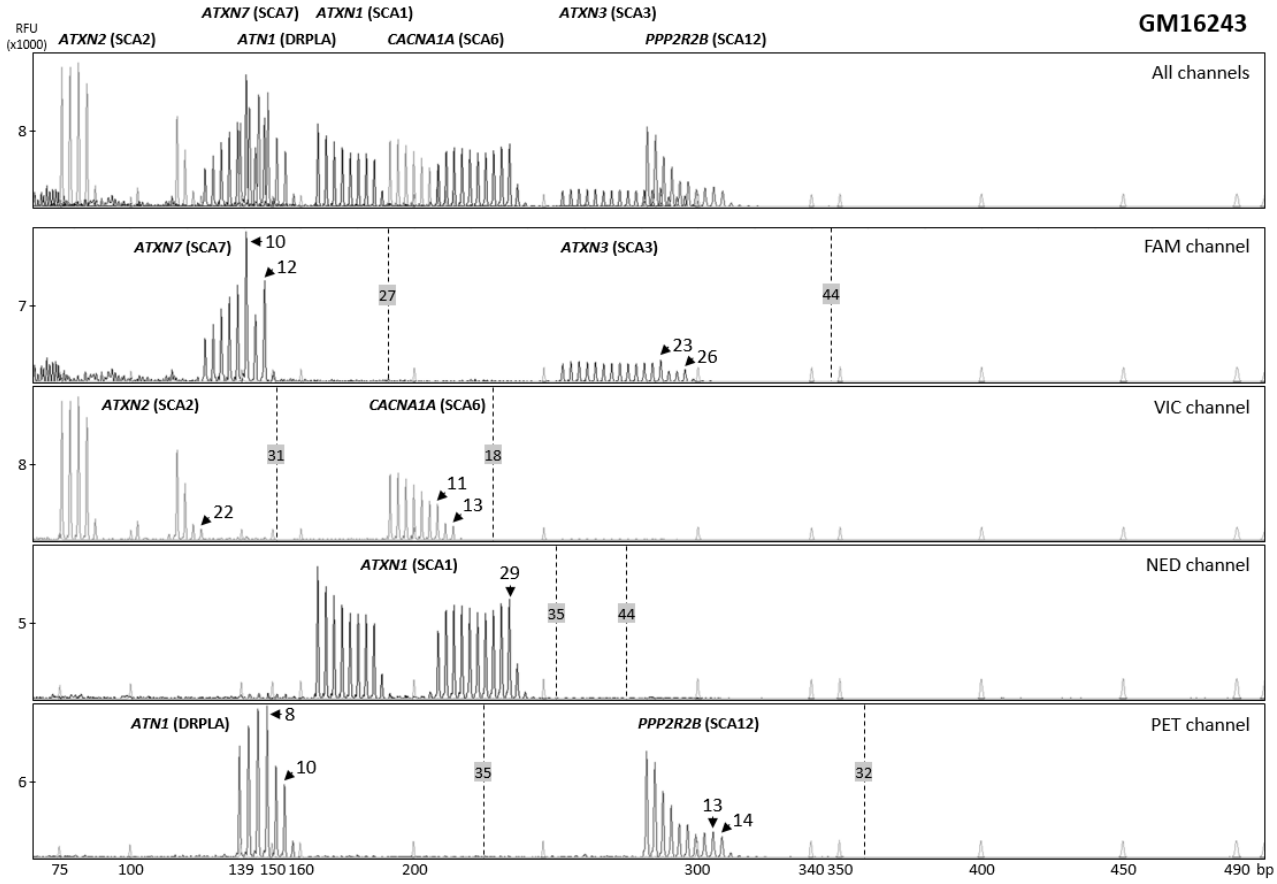

GM16243
RFU (x1000)
ATXN2 (SCA2)
ATXN7 (SCA7)
ATN1 (DRPLA)
ATXN1 (SCA1)
CACNA1A (SCA6)
ATXN3 (SCA3)
PPP2R2B (SCA12)
All channels
FAM channel
VIC channel
NED channel
PET channel
10
12
27
44
23
26
31
18
22
11
13
29
35
8
32
14
75
100
139
150
160
200
300
340
350
400
450
490 bp

**Figure 9**

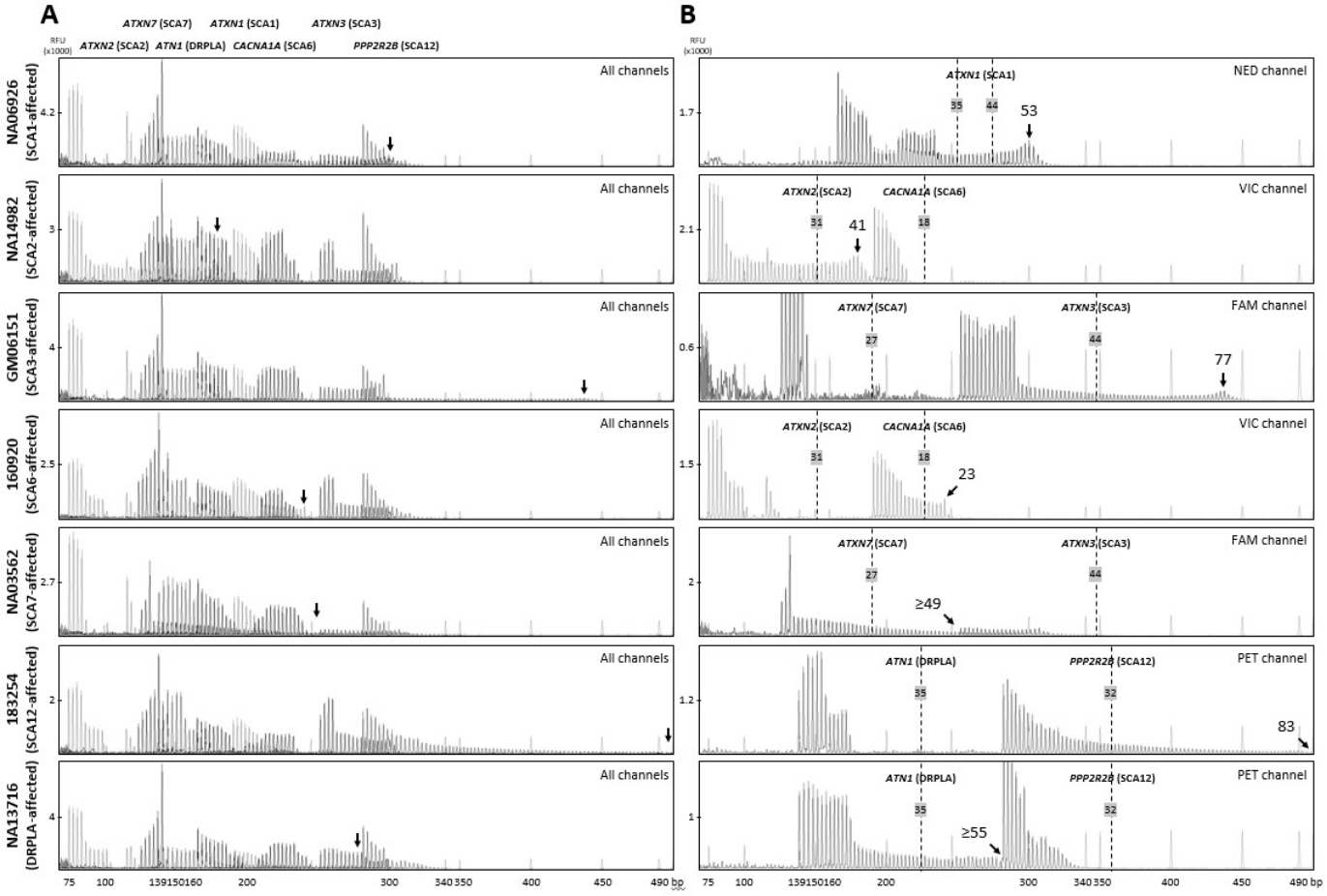

A
B
ATXN7 (SCA7)
ATXN1 (SCA1)
ATXN3 (SCA3)
ATXN2 (SCA2)
ATN1 (DRPLA)
CACNA1A (SCA6)
PPP2R2B (SCA12)
RFU (x1000)
NA06926 (SCA1-affected)
NA14982 (SCA2-affected)
GM06151 (SCA3-affected)
160920 (SCA6-affected)
NA03562 (SCA7-affected)
183254 (SCA12-affected)
NA13716 (DRPLA-affected)
All channels
NED channel
VIC channel
FAM channel
PET channel
53
41
77
23
≥49
83
≥55
75 100 139150160 200 300 340350 400 450 490 bp

**Figure 10**