A new powerful PCR-based assay for the molecular diagnosis of myotonic dystrophy type I

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Introduction

Myotonic dystrophy type 1 (DM1) is an autosomal dominant disease caused by an expansion of CTG triplets (> 50) in the 3'UTR region of *DMPK* gene. In affected alleles, triplets expansions range from 50 to 4000 CTG repeats. Currently, the diagnostic approach in our laboratory consists in carrying out a *DMPK* fluorescent PCR (amplification up to 110 CTG) followed by Southern blot (SB) for apparently homozygous patients. This process can be time-consuming and expensive.

We have assessed the sensitivity and specificity of the new prototype reagents AmplideX® PCR/CE *DMPK* Kit.

Materials and Methods

DNA were isolated from blood, chorionic villi (CVS) and amniotic fluid (AF) from 7 healthy donors and 42 DM1-patients. DM1-patients had CTG repeat size ranging from 58 to 1800.

DNA were amplified using prototype AmplideX® PCR/CE *DMPK* kit reagents (Asuragen) with two gene-specific (GS) primers and one repeat primer located in the repeats (Figure 1). FAM-labeled amplicons were migrated on 3500xL Genetic Analyzer Capillary Electrophoresis (CE) instruments (ThermoFisher) (PCR/CE). 10 samples from DM1-patients (88 to 1500 CTG repeats) were also amplified by the two gene-specific primers only and sized by agarose gel electrophoresis (PCR/AGE) using Reliant Mini 12-well precast 1% SeaKem Gold agarose gels (Lonza), Bionic Buffer (Sigma) and Quick-Load 2-Log DNA ladder (NEB) (Figure 3).

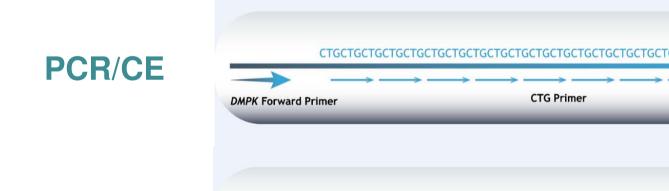
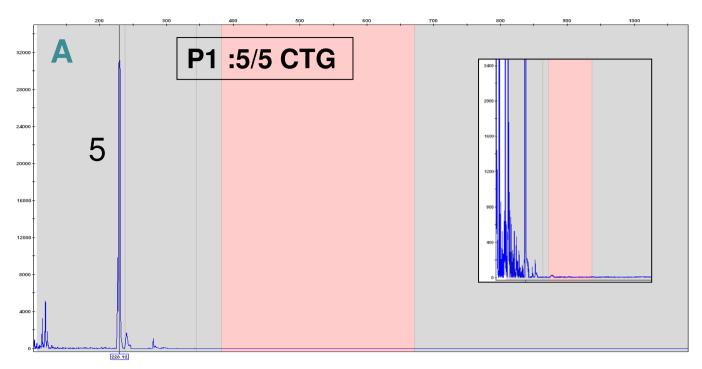


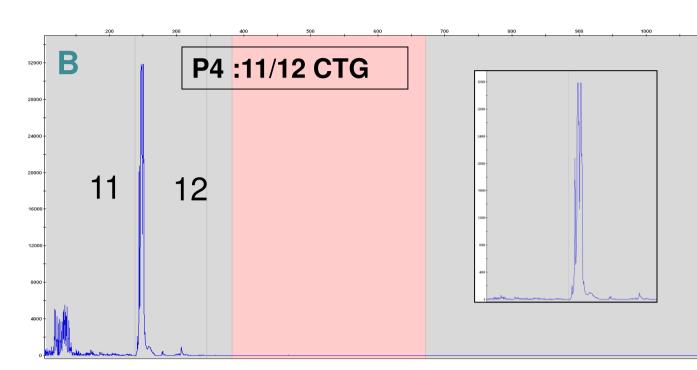
Figure 1 : Principle of the Asuragen's AmplideX® DMPK technology

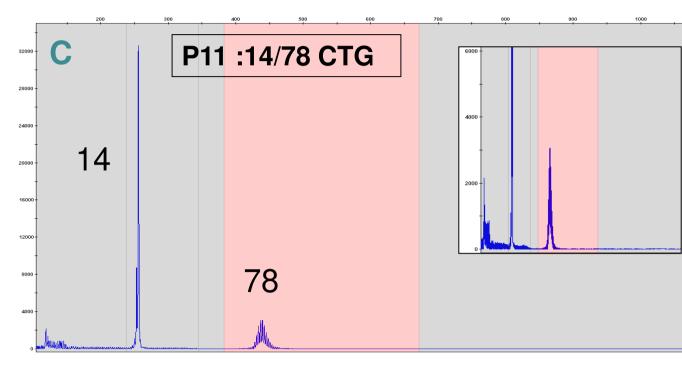
Figure 1: Principle of the Asuragen's AmplideX® DMPK technology. The assay consists of two gene-specific primers (GS) (FAM-labeled reverse primer), and one repeat primer that can hybridize and prime anywhere in the repeat region. The three primers are used for CE migration (PCR/CE) and only GS primers for sizing on agarose gel (PCR/AGE).

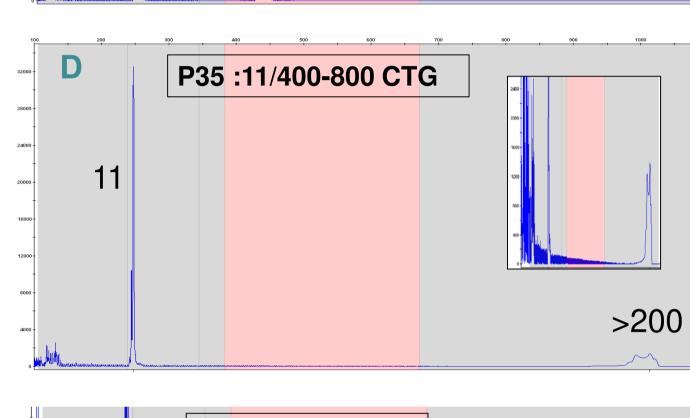
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28 blood 12 220-625 12/>200 YES 29 blood 12 240-700 12/66/>200 YES 30 CVS 5 270-420 5/>200 YES 31 blood 11 300-600 11/>200 YES 32 blood 5 350-650 5/>200 YES 33 blood 5 350-700 5/>>200 YES 34 blood 13 400-600 13/>>200 YES 35 blood 11 400-800 11/>>200 YES 36 CVS 5 500 5/>>200 YES 37 blood 12 500-850 12/>>200 YES 38 CVS 13 800 13/>>200 YES 39 blood 5 730-980 5/>>200 YES 40 blood 5 475-1080 5/>>200 YES 41 blood 5 <td>26</td> <td>blood</td> <td>12</td> <td>230-500</td> <td>12 / >200</td> <td>YES</td>	26	blood	12	230-500	12 / >200	YES
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30 CVS 5 270-420 5/>200 YES 31 blood 11 300-600 11/>200 YES 32 blood 5 350-650 5/>200 YES 33 blood 5 350-700 5/>200 YES 34 blood 13 400-600 13/>200 YES 35 blood 11 400-800 11/>200 YES 36 CVS 5 500 5/>200 YES 37 blood 12 500-850 12/>200 YES 38 CVS 13 800 13/>200 YES 39 blood 5 730-980 5/>200 YES 40 blood 5 475-1080 5/>200 YES 41 blood 5 500-1100 5/>200 YES 42 blood 5 500-1100 5/>200 YES 43 blood 5 <td< td=""><td>28</td><td>blood</td><td>12</td><td>220-625</td><td>12 / >200</td><td>YES</td></td<>	28	blood	12	220-625	12 / >200	YES
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35 blood 11 400-800 11/>200 YES 36 CVS 5 500 5/>200 YES 37 blood 12 500-850 12/>200 YES 38 CVS 13 800 13/>200 YES 39 blood 5 730-980 5/>200 YES 40 blood 5 475-1080 5/>>200 YES 41 blood 13 560-1000 13/>>200 YES 42 blood 5 500-1100 5/>>200 YES 43 blood 5 500-1100 5/>>200 YES 44 blood 5 1000-1300 5/>>200 YES 45 CVS 11 1334 11/>>200 YES 46 CVS 11 1400 11/>>200 YES 47 AF 23 1500 23/>>200 YES 48 blood 11 <	33	blood	5	350-700	5 / >200	YES
36 CVS 5 500 5/>200 YES 37 blood 12 500-850 12/>200 YES 38 CVS 13 800 13/>200 YES 39 blood 5 730-980 5/>200 YES 40 blood 5 475-1080 5/>200 YES 41 blood 13 560-1000 13/>>200 YES 42 blood 5 500-1100 5/>>200 YES 43 blood 11 570-1110 11/>>200 YES 44 blood 5 1000-1300 5/>>200 YES 45 CVS 11 1334 11/>>200 YES 46 CVS 11 1400 11/>>200 YES 47 AF 23 1500 23/>>200 YES 48 blood 11 1500 11/>>200 YES	34	blood	13	400-600	13 / >200	YES
37 blood 12 500-850 12 / >200 YES 38 CVS 13 800 13 / >200 YES 39 blood 5 730-980 5 / >200 YES 40 blood 5 475-1080 5 / >200 YES 41 blood 13 560-1000 13 / >200 YES 42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	35	blood	11	400-800	11 / >200	YES
38 CVS 13 800 13 / >200 YES 39 blood 5 730-980 5 / >200 YES 40 blood 5 475-1080 5 / >200 YES 41 blood 13 560-1000 13 / >200 YES 42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	36	CVS	5	500	5 / >200	YES
39 blood 5 730-980 5 / >200 YES 40 blood 5 475-1080 5 / >200 YES 41 blood 13 560-1000 13 / >200 YES 42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	37	blood	12	500-850	12 / >200	YES
40 blood 5 475-1080 5 / >200 YES 41 blood 13 560-1000 13 / >200 YES 42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	38	CVS	13	800	13 / >200	YES
41 blood 13 560-1000 13 / >200 YES 42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	39	blood	5	730-980	5 / >200	YES
42 blood 5 500-1100 5 / >200 YES 43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	40	blood	5	475-1080	5 / >200	YES
43 blood 11 570-1110 11 / >200 YES 44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	41	blood	13	560-1000	13 / >200	YES
44 blood 5 1000-1300 5 / >200 YES 45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	42	blood	5	500-1100	5 / >200	YES
45 CVS 11 1334 11 / >200 YES 46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	43	blood	11	570-1110	11 / >200	YES
46 CVS 11 1400 11 / >200 YES 47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	44	blood	5	1000-1300	5 / >200	YES
47 AF 23 1500 23 / >200 YES 48 blood 11 1500 11 / >200 YES	45	CVS	11	1334	11 / >200	YES
48 blood 11 1500 11 / >200 YES	46	CVS	11	1400	11 / >200	YES
	47	AF	23	1500	23 / >200	YES
49 blood 13 1100-1800 13 / >200 YES	48	blood	11	1500	11 / >200	YES
	49	blood	13	1100-1800	13 / >200	YES

Table 1: Comparison between *DMPK* fluorescent PCR and AmplideX® PCR/CE results









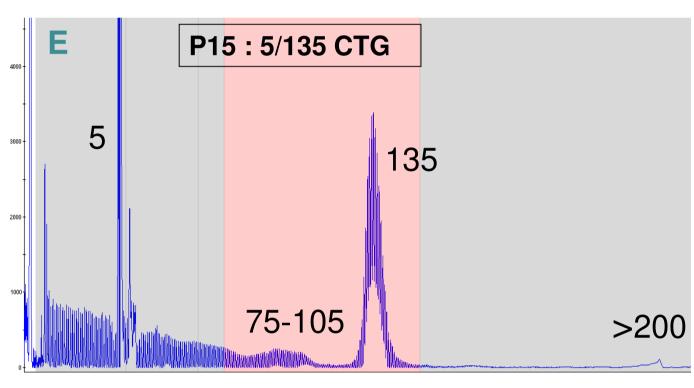


Figure 2: Examples of traces of PCR/CE assays

Expected genotypes are indicated at the top of each trace

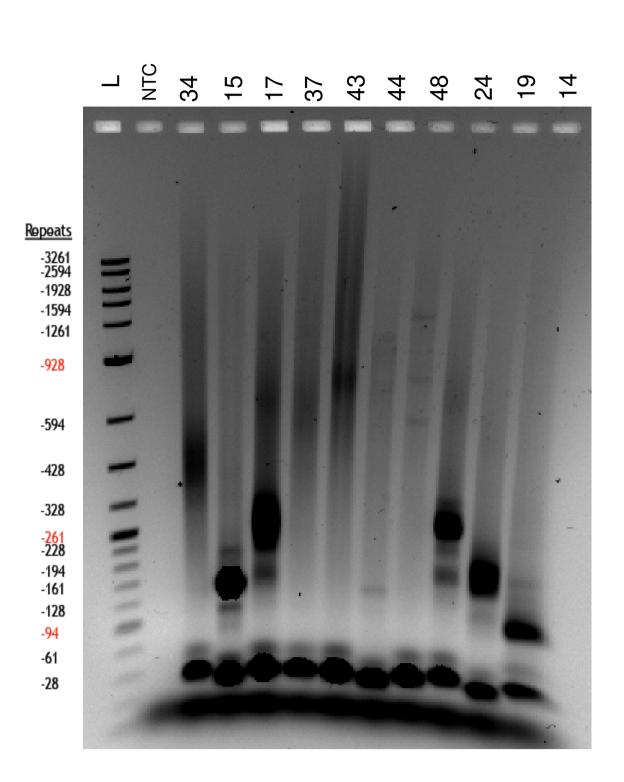


Figure 3: Migration of PCR/AGE: 10 patients carrier of expansion ranging from 88 to 1500 repeats.
L: ladder, NTC: Negative Template Control

Patient	<i>DMPK</i> fluorescent PCR	Southern Blot	AmplideX® PCR/CE	AmplideX® PCR/AGE	Concordant
34	13	400-600	13 / >200	350 - 580	YES
15	5	135	5 / 75-105 / 135 / >200	90 / 120 / 130-180 / 210	YES
17	12	100-230	12 / >200	150-190 / 200-350	YES
37	12	500-850	12 / >200	570-850	YES
43	11	570-1110	11 / >200	650-900	YES
44	5	1000-1300	5 / >200	150	NO
48	11	1500	11 / >200	570 / 800 / 1300	YES
24	16	220-350	16 / >200	150-180 / 260-350	YES
19	5	130-320	5 / 148 / 189	100 / 140-190	YES
14	12/88	-	12 / 90	70-100 / 150	YES

 Table 2 : Comparison between SB and AmplideX® PCR/AGE results

Results

• The AmplideX® PCR/CE assay gave expected results for the 49 samples (100% sensitivity and specificity) (Table 1).

Normal allele size was precisely determined, all the results were concordant with the *DMPK* fluorescent PCR.

Healthy homozygous patients were distinguished from DM1-patients thanks to the negative repeat profile (Figure 2A). Alleles separated by one triplet were easily differentiated (Figure 2B).

Expansions under 200 repeats were well characterized with a positive repeat profile (Figure 2C).

Large expansions over 200 repeats (ranging from 200 to 1800 repeats) were detected and appeared at the end of the profile (Figure 2D).

The repeat profile was positive for all the samples except for 2 CVS that had a negative repeat profile but for which expansions were amplified by the GS primers.

Previously unidentified mosaicism was revealed in 6 patients (Figure 2E).

• The AmplideX® PCR/AGE assay gave concordant results except for one patient (Table 2).

Expansions, ranging from 88 to 1500 repeats, gave various migration profiles with diverse types of mosaicism.

In one sample (patient 44), the expanded allele with 1300 CTG, revealed by Southern blot analysis, was not detected.

Mosaicism was also revealed in 3 patients (14, 15, 48).

Conclusion

The AmplideX® PCR/CE *DMPK* assay is therefore able to efficiently detect expanded alleles, to size alleles up to 200 repeats with a 1 triplet-resolution and to differentiate homozygous genotypes from others.

The AmplideX® PCR/AGE *DMPK* assay is able to size large expanded alleles up to at least 1800 repeats. Its interpretation requires some experience.

AmplideX® PCR *DMPK* technology appears to be simpler and faster than conventional techniques for the molecular DM1 diagnosis.









