

AmplideX® DM1 Dx Kit*

Transforming DM1 Diagnosis with Reliable *DMPK* Repeat Expansion Analysis



Myotonic Dystrophy Type I (DM1), also known as Steinert's Disease, is an inherited form of muscular dystrophy characterised by the presence of 50 or more CTG repeats within the *DMPK* gene. The AmplideX DM1 Dx Kit seeks to transform DM1 diagnosis by reliably analysing these hard-to-amplify CTG trinucleotide expansions and offering an alternative to Southern blot analysis. The PCR-only workflow, designed with labs in mind for implementation on common laboratory equipment, brings accessibility to *DMPK* analysis. By harnessing the power of proprietary repeat-primed PCR (RP-PCR) technology, **this kit** provides a streamlined, sensitive, and efficient method for analysing *DMPK* expansions, with results in a single day.

Reduced Complexity

- Proprietary PCR chemistry for GC-rich amplification and detection
- Offers an alternative to Southern blot
- Resolves zygosity and detects mosaicism

Optimised Workflow

- Direct injection of PCR products on CE instruments
- Fully kitted, end-to-end solution that reduces hands-on-time compared to Southern blot
- Sample-to-result possible within a single shift

Quality Performance

- Accurate sizing of alleles ≤ 200 CTG repeats and detection of any alleles >200 repeats using CE
- Optional agarose gel electrophoresis (AGE) protocol allows sizing of alleles up to 1000 repeats*
- Analysis tool converts raw base pair data into repeat number

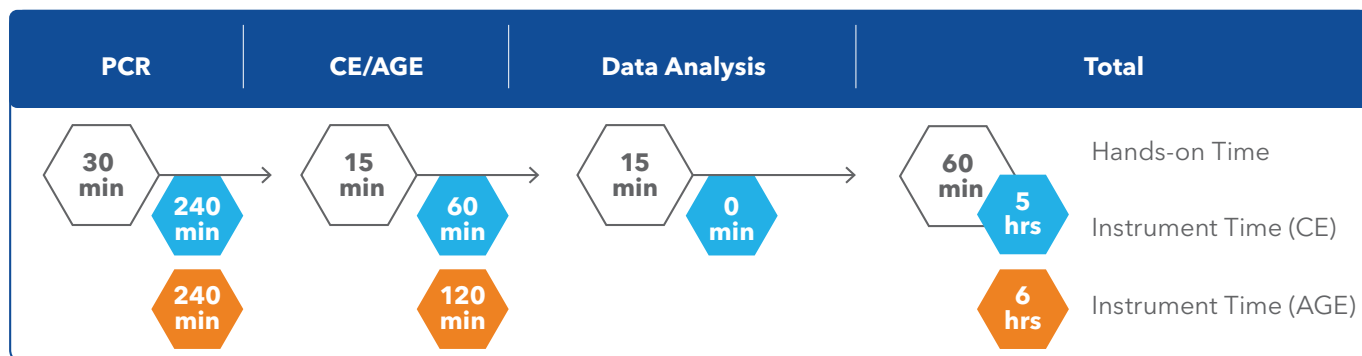


FIGURE 1. AmplideX PCR/CE DM1 Dx Workflow, including optional AGE workflow for larger expansions.

Sample	External Lab	Asuragen	Asuragen
Name	PCR + Southern	PCR/CE	PCR/AGE (allele 2)
S1	10,300-500	11, >200	Smear, High Mosaic 300-500
S2	4, 1200-1350	5, >200	-1100
S3	4, 1100-1500	5, >200	Smear, High Mosaic >500
S4	11, 200-300	12, >200	-260-350
S5	4, 800-900	5, >200	-650-700
S6	25, 1050	26, >200	-1000
S7	12, 100-150	13, 71, 129	70, 130
S8	11, 1900-2220	12, >200	700, 1900
S9	21, 300	22, >200	Mosaic, 261
S10	4, 50-250	5, >200	150, Smear >200

- Increased sensitivity for the detection of high mosaics[‡]
- Further resolution of bigger size repeat expansions
- Detection of polymorphisms

FIGURE 2. Comparison of residual clinical sample results between Asuragen (AmplidX Kit) and partner lab (PCR + Southern blot).

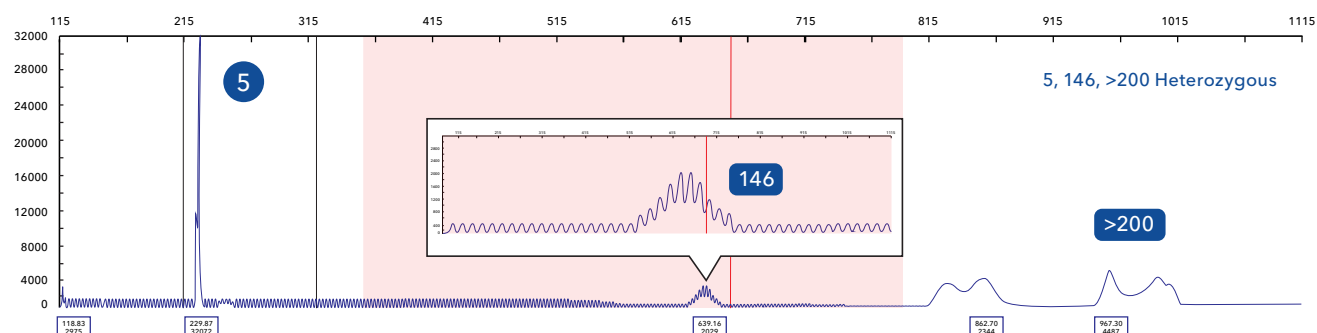


FIGURE 3. Sensitive and accurate sizing of DMPK expansions up to 200 repeats.

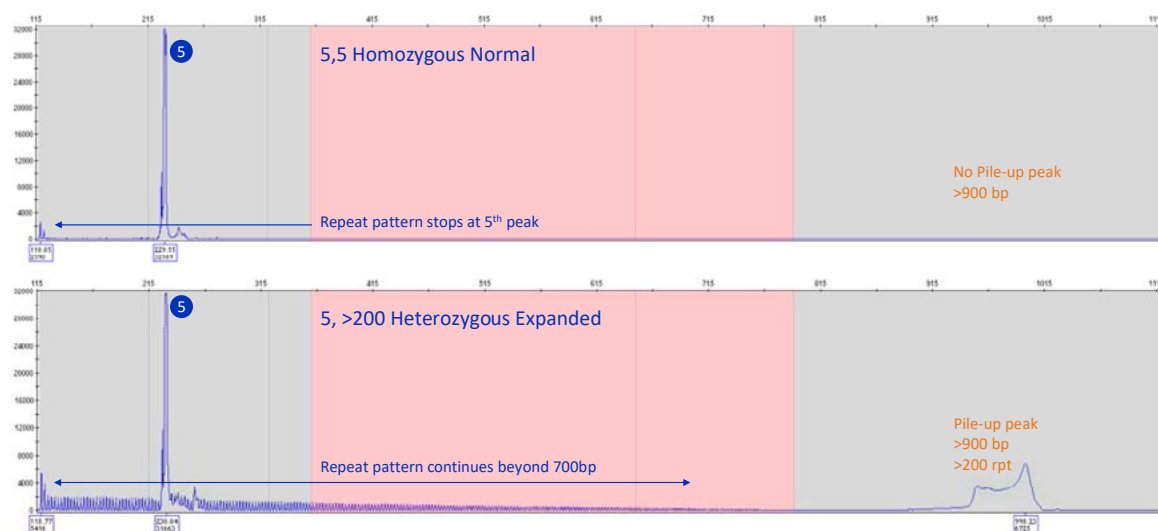


FIGURE 4. TOP FIGURE: Absence of CTG primed amplicons beyond the detected gene-specific peak with 5 CTG repeats, shows lack of a longer, heterozygous peak, demonstrating homozygosity; BOTTOM FIGURE: Both the CTG repeat primed amplicon repeat pattern extends beyond 700bp, and the presence of a pile-up peak >900bp indicates the presence of a longer, heterozygous peak, consistent with >200 CTG repeats.

Kit Ordering Information		
Product Name	Number of Reactions	Catalog Number
AmplidX DM1 Dx Kit*	32	76048

*CE-IVD for US export only. †For research use only
[‡]AmplidX detects mosaicism down to 5%



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