



# Myotonic Dystrophy

## Non-radioactive Myotonic Dystrophy CTG repeat genotyping

Catalog No. 40-2026-10 10 nmole

**For research use only. Not for use in diagnostic procedures for clinical purposes****Myotonic Dystrophy Genemer™ Primer** pair for amplification of CTG triple repeat spanning region.

The quantity supplied is sufficient for 400 regular 50 µl PCR reactions.

### Background

Myotonic dystrophy (DM) is the most common form of adult onset muscular dystrophy. It is an autosomal dominant disorder with a prevalence of about 1 in 8000. Clinical expression is highly variable and is related to age of onset. Onset of this disorder commonly occurs during young adulthood. However, it can occur at any age and is extremely variable in degree of severity. Myotonic dystrophy affects skeletal muscle and smooth muscle, as well as the eye, heart, endocrine system, and central nervous system.

The underlying mutations of DM are expansions of the CTG repeats located in the 3' untranslated region (UTR) of the myotonic dystrophy protein kinase (*DMPK*) gene on chromosome 19q. Severity of the disease is correlated with the length of the repeat expansion. Normal individuals have from 5 to 30 repeat copies; mildly affected persons have at least 50 repeats, while more severely affected patients have expansion of the repeat-containing segment up to several kilobase pairs.

Expansion is frequently observed in parent-to-child transmission, but extreme expansions are not transmitted through the male line. This explains: 1.) the occurrence of the severe congenital form is almost exclusively in the offspring of affected women; 2.) anticipation is commonly observed in affected families, that is, the disease demonstrates earlier onset and greater severity in each successive generation. The overall risk of having a congenitally affected child for any carrier woman is about 10%. If the woman has clinical signs of the condition, the risk of congenital myotonic dystrophy in offspring is 40% and this rises to 50% in subsequent pregnancies if an affected child has previously been born.

### Genotyping

Molecular diagnosis of myotonic dystrophy involves a combination of direct PCR analysis and Southern blotting tests to determine the CTG-repeat number within the *DMPK* gene. PCR can identify CTG expansions between 5-200 CTG repeats.

With larger expansions, Southern blot analysis of restriction fragments can be used for an accurate measure of the repeat size. Genomic DNA is restriction enzyme digested with Bam HI or Pst I. The DNA blot is then hybridized with a DM CTG repeat specific DNA probe. For more information, refer to **GLDM GeneProber™**.

### Material supplied

One tube containing 10 nmole lyophilized primers. The quantity supplied is sufficient for 400 regular 50 µl PCR reactions.

### Reconstitution

- Stock Primer solution:** Add 50 µl sterile water to the tube containing the primers. The 10 nmole of primer when dissolved in 50 µl water will give a solution of 200 µM, i.e. 200 pmole/µl.
- Primer Mix:** Prepare a 10 pmole/µl Primer Mix solution. Example: Transfer 10µl of stock primer solution to a new tube. Add 190 µl sterile water to this tube. Label this tube as **Primer Mix 10 pmole/µl**.

PCR\* reaction

### PCR profile

Denaturation	94°C	30 sec.
Annealing	63°C	30 sec.
Elongation	72°C	1 min.

**30 cycles, followed by 7 min at 72°C, and hold at 4°C.**

### Electrophoresis

Load samples in a 1.5% agarose gel. Run gel with appropriate voltage and time according to the electrophoresis device in your lab.

### Results and interpretation

Normal individuals have from 5 to 30 CTG repeats. For an individual with 10 CTG repeats, a 144-bp PCR product would be expected from the PCR reaction. A general formula,  $114 + 3n$  bp, can be applied to calculate other sizes of CTG repeats, where  $n$  represents the number of CTG repeats.

### References:

- The International Myotonic Dystrophy Consortium (2000) *Neurology* 54: 1218-1221.
- Steinbach, P. et al. (1998) *Am. J. Hum. Genet.* 62: 278-285.



## Myotonic Dystrophy Product Ordering Information

Product	Size	Catalog No.	Price, \$
Myotonic Dystrophy Genemer™ Primer pair for amplification of CTG triple repeat spanning region. The quantity supplied is sufficient for 400 regular 50 µl PCR reactions.	10 nmole	40-2026-10	100.00
Myotonic Dystrophy unlabeled GeneProber™ GLDM1 Probe unlabeled. Myotonic dystrophy CTG triple repeat spanning region unlabeled probe for radioactive labeling and Southern blot detection of <b>Bam HI</b> digested DNA. Suitable for random primer labeling.	500 ng	40-2026-40	350.00
Myotonic Dystrophy unlabeled GeneProber™ GLDM2 Probe unlabeled. Myotonic dystrophy CTG triple repeat spanning region unlabeled probe for radioactive labeling and Southern blot detection of <b>Pst I</b> digested DNA. Suitable for random primer labeling.	500 ng	40-2026-39	350.00
Myotonic Dystrophy unlabeled GeneProber™ GLDM3 Probe unlabeled. Myotonic dystrophy CTG triple repeat spanning region unlabeled probe for radioactive labeling and Southern blot detection. Suitable for random primer labeling.	500 ng	40-2026-38	350.00
Myotonic Dystrophy Digoxigenin labeled GeneProber™ GLDMDig2 Probe Digoxigenin labeled. Myotonic dystrophy CTG triple repeat spanning region digoxigenin labeled probe for Southern blot non-radioactive detection of <b>Pst I</b> digested DNA.	110 µl	40-2026-41	400.00
Myotonic Dystrophy PCRProber™ AP labeled probe Alkaline phosphatase labeled probe for PCR amplification based detection.	12 µl	40-2026-31	400.00
Myotonic Dystrophy PCRProber™ Kit. Kit for performing non-radioactive PCR amplification based detection. 5 blots (50 rxns)	5 blots	40-2026-32	650.00

**Genemer™ control DNA** Cloned fragment of the mutation region of a particular gene. These control DNA's are ideal genotyping templates for optimizing and performing control amplification with unknown DNA. The size of the triple repeats has been determined by sequencing and gel electrophoresis. The stability of size repeats upon cloning and amplification has NOT been determined. Thus, the size should be considered approximate and there is no claim for each fragment to contain the exact number of triple repeats. These control DNA's are sold with the express condition that these NOT be used for exact triple repeat size determination of DNA of unknown genotype. The control DNA should be used for determining the performance of specific Genemer™ and PCRProber™ Gene Link products.

GLDM 12 ~CTG repeat Genemer™ Control DNA	500 ng	40-2026-01	175.00
GLDM 45 ~CTG repeat Genemer™ Control DNA	500 ng	40-2026-02	175.00
GLDM 93 ~CTG repeat Genemer™ Control DNA	500 ng	40-2026-03	175.00
GLDM 129 ~CTG repeat Genemer™ Control DNA	500 ng	40-2026-04	175.00
GLDM 194 ~CTG repeat Genemer™ Control DNA	500 ng	40-2026-05	175.00

Please visit [www.genelink.com](http://www.genelink.com) for other Genemer™ control DNA not listed here

**Genemer™ Control DNA (Selected List)** Control DNA for use with gene or mutation specific Genemer™

Product	Size	Catalog No.	Price, \$
Fragile X, various CGG triple repeat region control DNA	500 ng	40-2004-XX	175.00
Huntington Disease various CAG triple repeat region control DNA	500 ng	40-2025-XX	175.00
Myotonic Dystrophy various CTG triple repeat region control DNA	500 ng	40-2026-XX	175.00
Friedreich's Ataxia, various GAA triple repeat region control DNA	500 ng	40-2027-XX	175.00

\*Please visit [www.genelink.com](http://www.genelink.com) for other Genemer™ not listed here

**Genemer™ (Selected List)** Primer pair for gene or mutation specific amplification. Special optimized conditions may be required for certain amplifications

Product	Size	Catalog No.	Price, \$
Fragile X (spanning CGG triple repeat region)	10 nmole	40-2004-10	100.00
Huntington Disease (spanning CAG triple repeat region)	10 nmole	40-2025-10	100.00
Myotonic Dystrophy (spanning CTG triple repeat region)	10 nmole	40-2026-10	100.00
Friedreich's Ataxia (spanning GAA triple repeat region)	10 nmole	40-2027-10	100.00
Factor V	10 nmole	40-2035-10	100.00
Factor VIII (Hemophilia)	10 nmole	40-2036-10	100.00
STS (Steroid Sulfatase)	10 nmole	40-2023-10	100.00
HGH (Human Growth Hormone)	10 nmole	40-2024-10	100.00
Sickle Cell	10 nmole	40-2001-10	100.00
RhD (Rh D gene exon 10 specific)	10 nmole	40-2002-10	100.00
Rh EeCc (Rh Ee and Cc exon 7 specific)	10 nmole	40-2003-10	100.00
Gaucher (various mutations)	10 nmole	40-2047-10	100.00
Cystic Fibrosis (various mutations)	10 nmole	40-2029-10	100.00
SRY (sex determining region on Y)	10 nmole	40-2020-10	100.00
X alphoid repeat	10 nmole	40-2021-10	100.00
Y alphoid repeat	10 nmole	40-2022-10	100.00

\*Please visit [www.genelink.com](http://www.genelink.com) for other Genemer™ not listed here

\*\*The polymerase chain reaction (PCR) process is covered by patents owned by Hoffmann-La Roche. A license to perform is automatically granted by the use of authorized reagents.

Prices subject to change without notice

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