Product Sheet

# Myotonic Dystrophy Genemer™ Control DNA*

***Specific control DNA for use with Gene Link Genemer™ & GeneProber™ product lines.**

Catalog No. 40-2026-0X

**For research use only. Not for use in diagnostic procedures for clinical purposes**

| Product | | Catalog Number | Unit Size |
|---|---|---|---|
| ❑ | GLDM 12 ~CTG repeat Genemer Control DNA | 40-2026-01 | **500 ng** |
| ❑ | GLDM 45 ~CTG repeat Genemer Control DNA | 40-2026-02 | **500 ng** |
| ❑ | GLDM 93 ~CTG repeat Genemer Control DNA | 40-2026-03 | **500 ng** |
| ❑ | GLDM 129 ~CTG repeat Genemer Control DNA | 40-2026-04 | **500 ng** |
| ❑ | GLDM 182 ~CTG repeat Genemer Control DNA | 40-2026-05 | **500 ng** |

**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.

**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 PstI digested or double enzyme digested with BamHI and HindIII. The DNA blot is then hybridized with a DM CTG repeat specific DNA probe. For more information, refer to **GLDM GeneProber**.

**Material Supplied**

A tube containing 500 ng of lyophilized control DNA segment.. The above control DNA is an ideal genotyping template for optimizing and performing control amplification with unknown DNA.

The quantity supplied is sufficient for 1000 regular 50µl PCR** reaction.

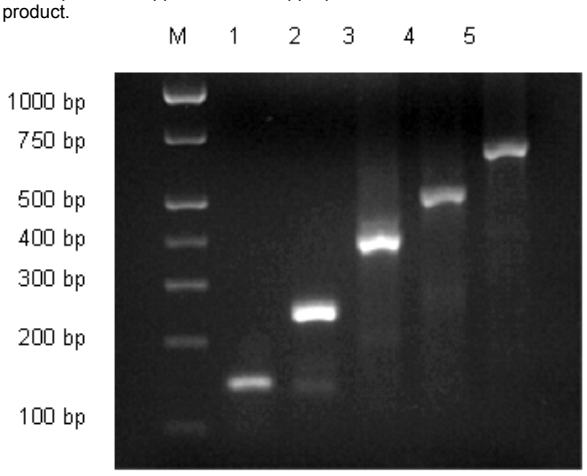
**Reconstitution**

1. **Stock Solution**: Add 100µl sterile water to the tube containing the lyophilized DNA to yield a solution of 5 ng/µl.
2. **Working Solution**: Dilute 1:10 an aliquot of the stock solution.

**Usage:** Initially use 1µl each of the stock and working template solution for amplification and optimization of the reaction. Based on the results, use 1µl of template at the lowest concentration.

**Protocol for PCR Analysis of Triple Repeat Size**

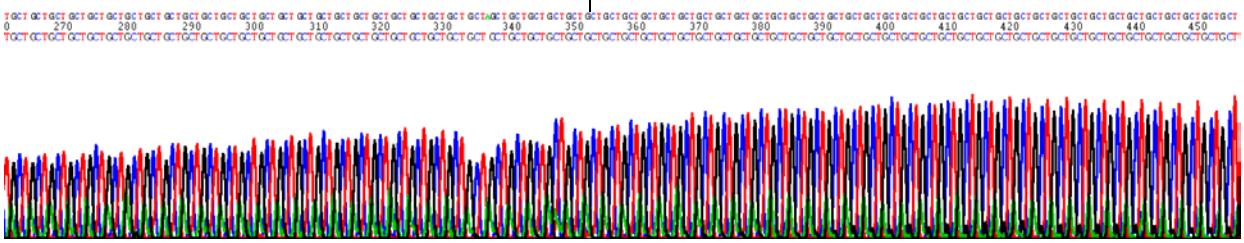
Follow protocol supplied with the appropriate Genemer™ or GeneProber™ product.




Lane M is molecular weight markers. Lanes 1-5 represents PCR products from DM genomic clones that contain 12, 45, 93, 129 and 182 CTG repeats respectively.

**References:**


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


Gene Link™

140 Old Saw Mill River Road Hawthorne, NY 10532
Tel: 914.769.1192 www.genelink.com Fax: 914.769.1193

# Myotonic Dystrophy Product Ordering Information

| Product | Size | Catalog No. | Price, $ |
|---|---|---|---|
| Myotonic Dystrophy Genemer™<br>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.<br>Myotonic dystrophy CTG triple repeat spanning region unlabeled probe for radioactive labeling and Southern blot detection of **Bam HI** digested DNA. Suitable for random primer labeling. | 500 ng | 40-2026-40 | 350.00 |
| Myotonic Dystrophy unlabeled GeneProber™ GLDM2 Probe unlabeled.<br>Myotonic dystrophy CTG triple repeat spanning region unlabeled probe for radioactive labeling and Southern blot detection of **Pst I** digested DNA. Suitable for random primer labeling. | 500 ng | 40-2026-39 | 350.00 |
| Myotonic Dystrophy unlabeled GeneProber™ GLDM3 Probe unlabeled.<br>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.<br>Myotonic dystrophy CTG triple repeat spanning region digoxigenin labeled probe for Southern blot non-radioactive detection of **Pst I** digested DNA. | 110 µl | 40-2026-41 | 400.00 |
| Myotonic Dystrophy PCRProber ™ AP labeled probe<br>Alkaline phosphatase labeled probe for PCR amplification based detection. | 12 µl | 40-2026-31 | 400.00 |
| Myotonic Dystrophy PCRProber ™ Kit.<br>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 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 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 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* All Gene Link products are for research use only

Gene Link ™
140 Old Saw Mill River Road Hawthorne, NY 10532
Tel: 914.769.1192 www.genelink.com Fax: 914.769.1193