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

<table>
<thead>
<tr>
<th>Product</th>
<th>Catalog Number</th>
<th>Unit Size</th>
</tr>
</thead>
<tbody>
<tr>
<td>GLDM 12 ~CTG repeat Genemer Control DNA</td>
<td>40-2026-01</td>
<td>500 ng</td>
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<tr>
<td>GLDM 45 ~CTG repeat Genemer Control DNA</td>
<td>40-2026-02</td>
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<tr>
<td>GLDM 93 ~CTG repeat Genemer Control DNA</td>
<td>40-2026-03</td>
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<td>GLDM 129 ~CTG repeat Genemer Control DNA</td>
<td>40-2026-04</td>
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<td>GLDM 182 ~CTG repeat Genemer Control DNA</td>
<td>40-2026-05</td>
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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.

References:

Genemer™ Kit for performing non-radioactive PCR amplification based detection. 5 blots (50 rxns)

Southern blot non-radioactive detection of Myotonic dystrophy CTG triple repeat spanning region digoxigenin labeled probe for GLDM 12 ~CTG repeat Genemer™ Control DNA use of authorized reagents.

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.

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**Prices subject to change without notice**

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