EMERY-DREIFUSS MUSCULAR DYSTROPHY, X-LINKED
EMD GENE SEQUENCING

Emery-Dreifuss muscular dystrophy, X-linked (EDMD; OMIM 310300) is due to mutations in the EMD gene (OMIM 300384). The X-linked form of Emery-Dreifuss muscular dystrophy is characterized by joint contractures, progressive muscle weakness, and cardiac conduction defects and arrhythmias in males. Female carriers are at increased risk for cardiac disease.

**Testing:** Testing is performed by sequence analysis of the EMD gene. The entire coding region, surrounding intronic regions, and parts of the 5’ and 3’ untranslated regions are included. This assay will detect point mutations, small deletions, and small insertions. For females, the assay will not detect a partial or whole gene deletion. For males, a partial or whole gene deletion may appear as failure to amplify a region of the gene.

This assay will detect mutations in EMD in greater than 99% of symptomatic individuals with a clear pattern of X-linked inheritance and/or with no emerin detected by immunodetection methods.

A negative test result does not rule out a diagnosis of Emery-Dreifuss muscular dystrophy (EDMD). A similar X-linked disorder, sometimes referred to as EDMD type 6, is caused by mutations in FHL1. Autosomal dominant and autosomal recessive forms of EDMD can be caused by mutations in LMNA.

A negative test does not rule out a genetic cause of muscular dystrophy. There are many other genes associated with different types of muscular dystrophy, some of which have features in common with EDMD.

**Turnaround time:** 10 -14 days

**CPT codes and cost:**

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
<th>Cost</th>
</tr>
</thead>
<tbody>
<tr>
<td>83891</td>
<td>x1 Proband</td>
<td>$450</td>
</tr>
<tr>
<td>83898</td>
<td>x4 Known mutation</td>
<td>$240</td>
</tr>
<tr>
<td>83904</td>
<td>x10</td>
<td></td>
</tr>
<tr>
<td>83912</td>
<td>x1</td>
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</tr>
</tbody>
</table>

**BILLING:** *We do not bill third party payers (insurance companies) for samples received from external sources.* The person or institution (Clinical Lab; Send-out Lab; Physician Office) sending the sample is responsible for full payment of the invoices within 30 days of receipt of the invoice. If the patient is on Medical assistance, please contact the lab prior to sample submission. Direct patient billing will be accepted only when a valid credit card form is received with the patient sample.

**Online resources:**
Muscular Dystrophy Association, USA – [http://www.mda.org](http://www.mda.org)
<table>
<thead>
<tr>
<th>Credit Card Billing Information</th>
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</table>
| **Patient Name:**              | **Credit Card:**  
|                                 | MasterCard        |
|                                 | American Express  |
|                                 | Visa              |
|                                 | Discover          |
|                                 | Other: _________  |
| **Name of Card holder:**       | **Credit Card Number:** |
| **Card holder address:**        | **Expiration Date:** |
| **Card holder phone:** Home:    | **Security Code:**  
| Work:                           | (on back of card) |
| **Card holder signature:**      | **Authorized payment amount:** |

**For Direct Patient Billing**

Prepayment for the testing services is required prior to beginning our testing. Please complete this form and include this paperwork with the shipment of the patient sample.

Billing questions can be addressed to:  
Denise Axsmith  
Senior Budget/Financial Analyst  
Nemours/A.I. duPont Hospital for Children  
daxsmith@nemours.org  
Phone: 302.651.6802  
Fax: 302.651.6881
Facsimile Verification Form

Name of Facility receiving Fax: __________________________________________________________

Name of Physician/Lab receiving Fax: ______________________________________________________

Street Address: _________________________________________________________________________

City_________________________ State: _____

Fax Number: ________________________________________
(to which lab results and/or patient information may be sent)

Phone Number: ______________________________

By signing this Facsimile Verification Form, I validate the accuracy of the above information and assume responsibility for assuring that the Fax machine is in a location which will maintain confidentiality of all reports transmitted by the Molecular Diagnostics Laboratory of the Alfred I. duPont Hospital for Children, to the above fax number.

Authorized Contact Person: ____________________________________________________________

Signature: ________________________________ Date: ______________

Title: __________________________________________________

In our continuing efforts to maintain patient confidentiality, the Molecular Diagnostics Laboratory of the Alfred I. duPont Hospital for Children requests you to verify the fax number only once (for this patient sample and for future patient reports) from your medical practice or institution and to assure that all faxes regarding patient information are received in a secure location in accordance with HIPAA regulations.

Please complete this Facsimile Verification Form and fax back to 302.651.6795.
If you have any questions regarding this form please contact Susan Kirwin, Assistant Director of The Molecular Diagnostics Laboratory, at 302.651.6777.

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