**DUCHENNE AND BECKER MUSCULAR DYSTROPHIES**  
**DMD DELETION TESTING**

Duchenne muscular dystrophy (OMIM 310200) and Becker muscular dystrophy (OMIM 300376) are due to mutations in the *DMD* gene, located at Xp12.2. Duchenne and Becker muscular dystrophies affect males and are characterized by progressive, symmetrical muscle weakness, loss of ambulation, elevated serum creatine phosphokinase concentration, and dilated cardiomyopathy. Males with Duchenne muscular dystrophy (DMD) are wheelchair-bound before 13 years of age. Males with Becker muscular dystrophy (BMD) are typically wheelchair-bound after 16 years of age. Mutations in *DMD* can also cause DMD-associated dilated cardiomyopathy, which affects both males and females, with more severe disease in males.

**Testing:** In the majority of cases, DMD and BMD are due to deletions of one or more exons of the *DMD* gene. These large deletions account for about 65% of the mutations in DMD and about 85% of the mutations in BMD. Testing is performed by multiplex PCR of select exons of the *DMD* gene. These exons are located throughout the gene and concentrated in regions that are known to be frequently deleted. Deletions that include one or more of these exons will be detected by this assay.

This test will not detect large duplications, small deletions or insertions, point mutations, or splicing mutations. Our laboratory does not perform sequencing of the DMD gene. This assay will not detect a heterozygous deletion; therefore our laboratory **does not perform carrier testing** in females for mutations in the *DMD* gene.

**Turn-around time:** 7 – 10 days

**CPT codes and cost:** 83890 (x1)  83900 (x1)  83901 (x2)  83894 (x1)  83912 (x1)  **$ 485**

**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 **only** be accepted when a valid credit card form is received with the patient sample.

**Online resources:**
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 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.

This is a confidential document that is being sent from a fax machine in a secure location. This fax is covered by the Electronic Communications Privacy Act 18 U.S.C. 2510-521. The information contained in this fax is considered privileged, is otherwise confidential and is intended only for the use of the individual or entity named above. Dissemination, distribution, or copying of this communication is strictly prohibited. If you have received this communication in error please notify us immediately by telephone, and return the original message to us at the above address via the U.S. Postal Service.
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