CONFIRMATION OF A CLINICAL DIAGNOSIS OF DUCHENNE MUSCULAR DYSTROPHY (DMD) OR BECKER MUSCULAR DYSTROPHY (BMD)

MANAGEMENT OF DMD/MBD IN SOME CASES, BASED ON THE TYPE OF DELETION ACCURSED (ALTERS FOR BETTER PREDICTION OF PROGNOSIS)

DETERMINATION OF CARRIER STATUS IN FAMILY MEMBER AT RISK FOR DMD OR BMD

PREGNATAL DIAGNOSIS OF DMD OR BMD IN AT-RISK PREGNANCIES

CLINICAL INFORMATION

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder characterized initially by proximal muscle weakness beginning before age 5 years. Affected individuals typically have pseudohypertrophy of the calf muscles and exhibit toe-walking, waddling gait, and the Gower sign (climbing up the legs when rising from a seated position on the floor). Not only is skeletal muscle affected in DMD, but also the smooth muscle of the gastrointestinal tract and possibly bladder, as well as cardiac muscle.

Initial symptoms are followed by dramatic progression of weakness leading to loss of ambulation by age 11 or 12. Death is often caused by cardiac failure or by respiratory failure before age 30, unless ventilator support is provided.

The allelic Becker muscular dystrophy (BMD) has a similar presentation, although age of onset is later and the clinical course is much milder. Cardiac involvement can be the only sign and patients are often ambulatory into their thirties.

DMD and BMD are caused by mutations in the DMD gene, which encodes for dystrophin. Approximately 50% to 65% of patients have intragenic deletions and approximately 5% to 10% have intragenic duplications. Less frequently, DMD and BMD result from nondeletion and nonduplication mutations, which are not detected by this assay.

Approximately one-third of sporadic cases of DMD/BMD occur due to new mutations. In sporadic cases, it is possible for the mother of an affected individual to have germline mosaicism. This means that the germ cells may contain a mutation even if the mutation is not detected in peripheral blood. In cases of germline mosaicism, which occurs with a frequency of up to 15%, further offspring are at risk for inheriting a dystrophin mutation.
INTERPRETATION

An interpretive report will be provided.

REFLEX TESTS

This test can only be performed if a mutation has previously been identified in a family member of this individual.
Submit only 1 of the following specimens:

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<th>TEST ID</th>
<th>REPORTING NAME</th>
<th>AVAILABLE SEPARATELY</th>
<th>ALWAYS PERFORMED</th>
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<tr>
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<td>Fibroblast Culture for Genetic Test</td>
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<td>No</td>
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<tr>
<td>CULAF</td>
<td>Amniotic Fluid Culture/Genetic Test</td>
<td>Yes</td>
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<td>MATCC</td>
<td>Maternal Cell Contamination, B</td>
<td>Yes</td>
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</tbody>
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SPECIMEN REQUIRED

This test can only be performed if a mutation has previously been identified in a family member of this individual.

CLINICAL REFERENCES