Expanded Neuromuscular Disorders: Deletion/Duplication Panel

Test Code: MD360
Turnaround time: 2 weeks
CPT Codes: 81161 x1, 81404 x1, 81405 x1, 81406 x1

Condition Description

Neuromuscular disorders (NMDs) collectively refer to the many disorders that affect the peripheral nervous system either by impairing the proper development or functioning of muscles, or by damaging the associated nerves or neuromuscular junctions. NMDs comprise over 200 Mendelian disorders, all of which are rare individually, but have an approximate disease prevalence of 1 in 3,000 altogether. Of the inherited NMDs, muscular dystrophies are the most common. Muscular dystrophies are highly heterogeneous muscle disorders that share clinical, genetic, and pathological characteristics; their major clinical characteristics include muscle degeneration and wasting, progressive muscle weakness, hypotonia, and elevated serum creatine kinase levels.

The expanded neuromuscular panel includes 79 genes which demonstrate a wide range of clinical presentation and heterogeneity. They include muscular dystrophies, congenital myopathies, and congenital myasthenic syndrome. Over the past few years a number of genes with overlapping clinical phenotypes have been identified in neuromuscular disorders. The expanded neuromuscular disorders panel is designed to capture the entire mutation spectrum of these genes.

References:


Genes

ACTA1, AMPD1, ANOS1, BAG3, BIN1, BSCL2, CAPN3, CAV3, CAVIN1, CFL2, CHAT, CHRNA1, CHRNB1, CHRNA4, CHRNA5, CHRNA6, CHRNA7, CHRNA8, CHRNA9, CHRNA10, CHRNA11, CHRNA12, CHRNA13, CHRNA14, CHRNA15, CHRNA16, CHRNA17, CHRNA18, CHRNA19, CHRNA20, CHRNA21, CHRNA22, CHRNA23, CHRNA24, CHRNA25, CHRNA26, CHRNA27, CHRNA28, CHRNA29, CHRNA30, CHRNA31, CHRNA32, CHRNA33, CHRNA34, CHRNA35, CHRNA36, CHRNA37, CHRNA38, CHRNA39, CHRNA40, CHRNA41, CHRNA42, CHRNA43, CHRNA44, CHRNA45, CHRNA46, CHRNA47, CHRNA48, CHRNA49, CHRNA50, CHRNA51, CHRNA52, CHRNA53, CHRNA54, CHRNA55, CHRNA56, CHRNA57, CHRNA58, CHRNA59, CHRNA60, CHRNA61, CHRNA62, CHRNA63, CHRNA64, CHRNA65, CHRNA66, CHRNA67, CHRNA68, CHRNA69, CHRNA70, CHRNA71, CHRNA72, CHRNA73, CHRNA74, CHRNA75, CHRNA76, CHRNA77, CHRNA78, CHRNA79, CHRNA80, CHRNA81, CHRNA82, CHRNA83, CHRNA84, CHRNA85, CHRNA86, CHRNA87, CHRNA88, CHRNA89, CHRNA90, CHRNA91, CHRNA92, CHRNA93, CHRNA94, CHRNA95, CHRNA96, CHRNA97, CHRNA98, CHRNA99, CHRNA100, CHRNA101, CHRNA102, CHRNA103, CHRNA104, CHRNA105, CHRNA106, CHRNA107, CHRNA108, CHRNA109, CHRNA110, CHRNA111, CHRNA112, CHRNA113, CHRNA114, CHRNA115, CHRNA116, CHRNA117, CHRNA118, CHRNA119, CHRNA120, CHRNA121, CHRNA122, CHRNA123, CHRNA124, CHRNA125, CHRNA126, CHRNA127, CHRNA128, CHRNA129, CHRNA130, CHRNA131, CHRNA132, CHRNA133, CHRNA134, CHRNA135, CHRNA136, CHRNA137, CHRNA138, CHRNA139, CHRNA140, CHRNA141, CHRNA142, CHRNA143, CHRNA144, CHRNA145, CHRNA146, CHRNA147, CHRNA148, CHRNA149, CHRNA150, CHRNA151, CHRNA152, CHRNA153, CHRNA154, CHRNA155, CHRNA156, CHRNA157, CHRNA158, CHRNA159, CHRNA160, CHRNA161, CHRNA162, CHRNA163, CHRNA164, CHRNA165, CHRNA166, CHRNA167, CHRNA168, CHRNA169, CHRNA170, CHRNA171, CHRNA172, CHRNA173, CHRNA174, CHRNA175, CHRNA176, CHRNA177, CHRNA178, CHRNA179, CHRNA180, CHRNA181, CHRNA182, CHRNA183, CHRNA184, CHRNA185, CHRNA186, CHRNA187, CHRNA188, CHRNA189, CHRNA190, CHRNA191, CHRNA192, CHRNA193, CHRNA194, CHRNA195, CHRNA196, CHRNA197, CHRNA198, CHRNA199, CHRNA200, CHRNA201, CHRNA202, CHRNA203, CHRNA204, CHRNA205, CHRNA206, CHRNA207, CHRNA208, CHRNA209, CHRNA210, CHRNA211, CHRNA212, CHRNA213, CHRNA214, CHRNA215, CHRNA216, CHRNA217, CHRNA218, CHRNA219, CHRNA220, CHRNA221, CHRNA222, CHRNA223, CHRNA224, CHRNA225, CHRNA226, CHRNA227, CHRNA228, CHRNA229, CHRNA230, CHRNA231, CHRN
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Neuromuscular Disorders Panel.
- Limb-girdle Muscular Dystrophy Panel.
- Congenital Muscular Dystrophy Panel.
- Expanded Neuromuscular Disorders: Sequencing Panel.