# GeneSeq PLUS, DMD

Order Name: **GeneSeq P/DMD**
Test Number: 5194966
Revision Date: 03/21/2023

| TEST NAME | METHODOLOGY | LOINC CODE |
|---|---|---|
| **GeneSeq PLUS, DMD** | Polymerase Chain Reaction | |

## SPECIMEN REQUIREMENTS

| Specimen | Specimen Volume (min) | Specimen Type | Specimen Container | Transport Environment |
|---|---|---|---|---|
| **Preferred** | **8.5 mL (3 mL)** | **Whole Blood** | **ACD Solution A or B (Yellow Top)** | **Room Temperature** |
| **Alternate 1** | **8.5 mL (3 mL)** | **Whole Blood** | **EDTA (Lavender Top)** | **Room Temperature** |
| **Alternate 2** | **1** | **Saliva** | **Oragene Dx saliva kit** | **Room Temperature** |
| **Alternate 3** | **1** | **Buccal swab** | **PurFlock buccal swab kit** | **Room Temperature** |

**Instructions**

**Specimen Type:** Whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
**Specimen Volume:** 8.5 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
**Mininum Volume:** 3 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
**Collection:** Standard phlebotomy. Follow PurFlock buccal swab kit or Oragene Dx 500 saliva kit collection instructions. Do not eat, drink, smoke, or chew gum 30 min prior to collection.
**Specimen Storage:** Maintain specimen at room temperature or refrigerate at 4C Do not freeze.
**Special Instructions:** Variants of uncertain significance (VUS) will be reported unless VUS opt out is indicated on the requisition. If requesting full gene sequencing for multiple genes, order **GeneSeq PLUS** [**5194962**]. To test fetal specimens, including cordblood, order test code **GeneSeq PLUS, Fetal Analysis** [5194963] Test orders must include an attestation that the provider has the patient's informed consent for genetic testing. See sample physician office consent form: Consent for Genetic Testing.

## GENERAL INFORMATION

| | |
|---|---|
| **Expected TAT** | 14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests. |
| **Clinical Use** | This test is used for carrier and diagnostic testing for Duchenne and Becker muscular dystrophy and x-linked cardiomyopathy. Next-generation sequencing: Identifies genetic variants, including small nucleotide variants (SNVs), insertions, deletions and copy number variants (CNVs). Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples or erroneous representation of family relationships. |
| **Notes** | Labcorp Test Code: 482466 |
| **Service Provided By** | labcorp Oklahoma, Inc. |