

## HOTLINE: Effective February 16, 2021

New Test 3003144 Deletion/Duplication Analysis by MLPA DELDUP

Methodology: Multiplex Ligation-dependent Probe Amplification

**Performed:** Sun- Sat **Reported:** Within 14 days

Specimen Required: Collect: Contact ARUP's genetic counselor at (800) 242-2787 extension 2141 prior to test submission. Disease-specific patient

history forms are available at www.aruplab.com/Testing-Information/consentforms-patienthistory.jsp

Remarks: Submission of a completed patient history form is required. If testing is ordered to assess for a large deletion/duplication previously identified in a family member, submission of the family member's laboratory report is required. Testing will begin once all

required documentation is received.

## **Interpretive Data:**

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

## Note:

Deletion/duplication analysis by MLPA is offered for the following genes: ABCD1, ACADVL, ACVRL1, APC, ATP7A, BMPR1A, BRCA1, BRCA2, CFTR, COL4A5, ENG, F8, F9, FBN1, HBB, MECP2, MEN1, MLH1/MSH2, MSH6, NF1, OTC, PKD1, PKD2, PLOD1, PMS2, PRSS1, PTEN, RASA1, SDHB, SDHC, SDHD, SLC22A5, SHOX, SMAD4, SPINK1, SPRED1, STK11, TP53, VHL.

Suspected deletions or duplications in exons 12-15 of *PMS2*, require additional sequencing to exclude pseudogene copy number variants. Additional charges apply.

CPT Code(s): CPT codes vary based on gene.

New York DOH approval pending. Call for status update.

**HOTLINE NOTE:** Refer to the Test Mix Addendum for interface build information.