

## PMP22 Gene, Large Deletion/Duplication Analysis, Varies

**Test ID:** PMPDD

**Explanation:** On the effective date, the Specimen Required, Stability, and Days Performed will be updated. The whole blood specimen type will no longer accept any anticoagulant and additional acceptable specimen types will be added.

Current Specimen Required	New Specimen Required
<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.</p> <p><b>Specimen Type: Whole blood</b> <b>Container/Tube:</b> <b>Preferred:</b> Lavender top (EDTA) or yellow top (ACD) <b>Acceptable:</b> Any anticoagulant <b>Specimen Volume:</b> 3 mL <b>Collection Instructions:</b> 1. Invert several times to mix blood. 2. Send specimen in original tube. <b>Additional Information:</b> To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.</p>	<p><b>Patient Preparation:</b> A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.</p> <p><b>Submit only 1 of the following specimens:</b></p> <p><b>Specimen Type: Whole blood</b> <b>Container/Tube:</b> <b>Preferred:</b> Lavender top (EDTA) or yellow top (ACD) <b>Acceptable:</b> None <b>Specimen Volume:</b> 3 mL <b>Collection Instructions:</b> 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. <b>Do not aliquot.</b> <b>Specimen Stability Information:</b> Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days <b>Additional Information:</b> 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.</p> <p><b>Specimen Type: Cord blood</b> <b>Container/Tube:</b> <b>Preferred:</b> Lavender top (EDTA) or yellow top (ACD) <b>Acceptable:</b> None <b>Specimen Volume:</b> 3 mL <b>Collection Instructions:</b> 1. Invert several times to mix blood. 2. Send specimen in original tube. <b>Do not aliquot.</b> <b>Specimen Stability Information:</b> Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days <b>Additional Information:</b></p>



1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.

2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

3. While a properly collected cord blood sample may not be at risk for maternal cell contamination, unanticipated complications may occur during collection. Therefore, maternal cell contamination studies are recommended to ensure the test results reflect that of the patient tested and are available at an additional charge.

4. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

**Specimen Type: Extracted DNA**  
**Container/Tube:**  
**Preferred:** Screw Cap Micro Tube, 2mL with skirted conical base  
**Acceptable:** Matrix tube, 1mL  
**Collection Instructions:**

1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.

**Specimen Stability Information:** Frozen (preferred) 1 year/  
Ambient/Refrigerated

**Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Current Specimen Stability			
Specimen	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerate		

New Specimen Stability			
Specimen	Temperature	Time	Special Container
Varies	Varies		

Current Days Performed
Batched 1 time per week

New Days Performed
Varies

**Questions**

Contact Michelle Rath, Laboratory Resource Coordinator at 800-533-1710.