Overview

Useful For
Diagnosing platelet disorders

Genetics Test Information
This test is indicated for assessing platelet ultra-structural abnormalities in congenital and acquired platelet disorders.

Special Instructions
- Platelet Esoteric Testing Patient Information

Method Name
Platelet Ultra-Structural Examination by Transmission Electron Microscopy

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD

Shipping Instructions
Ship specimen overnight in an Ambient Shipping Box-Critical Specimens Only (T668) following the instructions in the mailer.

Send specimen Monday through Wednesday

Specimen Required

Patient Preparation: Fasting is preferred but not required.

Supplies: Ambient Shipping Box-Critical Specimens Only (T668)

Collection Container/Tube:

Preferred: Yellow top (ACD, solution B)

Acceptable: Yellow top (ACD, solution A)

Specimen Volume: 6 mL

Collection Instructions: Send specimen in original tube. Do not transfer blood to other containers.

Forms
1. Platelet Esoteric Testing Patient Information in Special Instructions

2. If not ordering electronically, complete, print, and send a Coagulation Test Request (T753) with the specimen.
Specimen Minimum Volume
3 mL

Reject Due To

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<td>Gross lipemia</td>
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Specimen Stability Information

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<td>72 hours</td>
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Clinical and Interpretive

Clinical Information

Patients with either hereditary or acquired platelet disorders usually have bleeding diathesis, which can potentially be life threatening. A reliable laboratory diagnosis of a platelet disorder can significantly impact patients’ and, potentially, their family members’ clinical management and outcome.

Platelet (P) transmission electron microscopy (TEM) has been an essential tool for laboratory diagnosis of various hereditary platelet disorders since it was first used to visualize fibrin-platelet clot formation in 1955. PTEM employs 2 main methods to visualize platelet ultrastructure, whole mount (WM) TEM and thin section (TS) TEM.

WM-TEM is considered the gold standard test for diagnosing dense granule deficiencies in Hermansky-Pudlak syndrome, alpha-delta platelet storage pool deficiency, Paris-Trousseus-Jacobsen syndrome, Wiskott-Aldrich syndrome, TAR (thrombocytopenia, absent radii) syndrome, Chediak-Higashi syndrome, and more.

TS-TEM is a preferred method to visualize platelet alpha granules, other organelles and abnormal inclusions.

Platelet disorders that can be detected by PTEM include (but are not limited to):

Delta granules (dense bodies):
- Hermansky Pudlak syndrome
- Wiskott-Aldrich syndrome
- Chediak Higashi syndrome
- Jacobson/Paris Trousseau syndrome
- York platelet syndrome
- Storage pool deficiency, not otherwise specified

Alpha granules:
Gray platelet syndrome

White platelet syndrome

X-linked GATA 1 mutation

Jacobson/Paris Trousseau syndrome

Alpha and delta granules:

-Alpha-delta storage pool deficiency

**Reference Values**

Mean dense granules/platelet \(\geq 1.2\)

**Interpretation**

Ultrastructural abnormalities identified by platelet transmission electron microscopy (TEM) are evaluated by a Mayo hematopathologist.

Platelet size, alpha granules, golgi complex, and abnormal inclusions will be assessed as part of the morphologic examination under TEM.

Distinct and sometimes pathognomonic ultrastructural abnormalities are found in Hermansky Pudlak syndrome, gray platelet syndrome with virtually absent alpha granules, white platelet syndrome, Medich giant platelet disorder, X-linked GATA-1 macrothrombocytopenia, and, recently described, York platelet syndrome.

**Cautions**

Whole blood sample collected in an ACD tube should be stored and transported at ambient temperature within 72 hours of collection. Suboptimal transportation may cause falsely low dense granule counts.

**Supportive Data**

Extensive validation studies with normal donors and known patient samples were performed. A total 111 normal donor platelet samples were assessed to establish the baseline. Of the 10 patient samples, 6 were from patients with Hermansky-Pudlak syndrome, 2 patients had gray platelet syndrome, 1 had \(MYH9\) mutation-associated platelet disorder, and 1 had Paris-Trousseau/Jacobson syndrome.

**Clinical Reference**


**Performance**

**Method Description**

This test identifies dense granule deficiency by semiquantitative counting of dense granules on whole mount grids by technologists and verified by a hematopathologist/hematologist.
Test Definition: PTEM
Platelet TEM, B


PDF Report
No

Day(s) and Time(s) Test Performed
Monday through Friday; 7 a.m.-3 p.m.

Analytic Time
10 days

Maximum Laboratory Time
10 days

Specimen Retention Time
Not retained

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information
85390
88348

LOINC® Information

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