TEST ID: PTEM

PLATELET TRANSMISSION ELECTRON MICROSCOPIC STUDY

USEFUL FOR

Diagnosing platelet disorders

GENETICS TEST INFORMATION

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

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 transmission electron microscopy (PTEM) 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-Trousseau-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

REFERENCE VALUES

Mean dense granules/platelet
≥ 1.2

ANALYTIC TIME

10 days
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

SPECIMEN REQUIRED

Specimen Type
Whole Blood ACD

Container/Tube
Preferred: Yellow top (ACD, solution B)
Accepted: Yellow top (ACD, solution A)

Specimen Volume
6 mL

INTERPRETATION

Ultrastructural abnormalities identified by platelet transmission electron microscopy 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 transmission electron microscopy.
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.

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