Interphase Fluorescence In Situ Hybridization (FISH) Analysis for Molar Pregnancy 
#81006

Clinical

In some abnormal pregnancies, trophoblastic tissue burrows into the uterine wall and 1-3-cm fluid-filled cysts derived from placental chorionic villi are formed. Hence the condition is referred to as a molar pregnancy or a hydatidiform mole. Molar pregnancies can be either complete or partial. It is difficult sometimes to distinguish among early complete molar pregnancy, partial molar pregnancy, and hydropic degeneration, which occurs in 15-40% of nonmolar spontaneous abortions.

Complete molar pregnancies are characterized by:
- Large placenta
- Involvement of almost all villi, which appear as clusters of grapes
- Absence of a fetus
- Haploid 46,XX (85%) and 46,XY (15%) karyotype
- Chromosomes of paternal origin

Partial molar pregnancies are characterized by:
- Slightly enlarged placental volume
- A mixture of normal or hydropic villi
- Fetal tissue in early embryonic stages
- Triploid karyotype (69,XXX; 69,XXY; or 69,XYY)
- One maternal haploid set of chromosomes and 2 paternal haploid sets (diandric triploidy)

Rarely, the partial molar pregnancy can be digynic (2 maternal haploid sets) triploidy, and unlike diandric triploidy, the placenta is smaller. In both forms, malformed fetuses may be present in early embryonic stages.

A molar pregnancy places the mother at risk for choriocarcinoma, although the risk is low in case of partial molar pregnancy. It is estimated that surgical intervention is required in 2.1% of partial molar pregnancies, compared with 10% in complete molar pregnancies.

Useful For
- Confirming a diagnosis of molar pregnancy
- Differentiating between complete and partial hydatidiform moles
- Determining sex chromosome makeup and ploidy level from tissue of suspected molar pregnancies

Interpretation

The sex chromosome makeup, ploidy pattern(s), and the percentage of cells demonstrating each pattern are reported.

The level of accuracy for detection of ploidy level and sex chromosome constitution is estimated at 97% or higher.

Cautions
- The limit of detection of triploidy is 20%; triploidy can be determined only if the proportion of triploid cells exceeds 20%.
- Very rarely triple trisomy involving X, Y, and 18 can occur as a false-positive result.
Supportive Data

The specificity of these probes has been reported to be 100% with no cross hybridization. To establish the analytic sensitivity, 16 blinded specimens were analyzed, of which 7 were normal and 9 were abnormal. Two technologists scored 100 nuclei each for the 3 probes. The combined normal patterns (2 aqua, 2 green or 2 aqua, 1 green, and 1 orange) averaged 91%. The normal cutoff for abnormal signal patterns at 95% confidence interval (CI) was 3%. Nine abnormalities from partial molar pregnancies were correctly identified and the average proportion of triploidy was 82.8%, ranging from 47-100%. The normal cutoff of triploid pattern at 95% CI was 20%. In 2 abnormal specimens, the analysis was not successful. Subsequently, 18 additional specimens of suspected molar pregnancies have been successfully analyzed using this test. (Jalal SM, Law ME, Carlson RO, Dewald GE: Prenatal detection of aneuploidy by directly labeled multicolored probes and interphase fluorescence in situ hybridization. Mayo Clin Proc 1998;73;132-137)

Reference


Method

Five-micron sections of paraffin-embedded tissue are used for interphase FISH analysis. Sections are placed on 3-4 glass slides and the centromere specific probes for X (green), Y (orange), and 18 (aqua) are utilized. The specimens are denatured, hybridized, and scored by 2 technologists (100 nuclei each). When the proportion of 3 green, 3 aqua; or 2 green, 1 red, 3 aqua; or 1 green, 2 red, 3 aqua signal pattern (triploidy) is higher than 20%, it is interpreted as a triploid with XXX, XXY, or XYY chromosomes. When the signal pattern of 2 green, 2 aqua; or 1 green, 1 red, 2 aqua is 97% or higher, it is interpreted as diploid with either XX or XY chromosomes. (Jalal SM, Law ME, Carlson RO, Dewald GE: Prenatal detection of aneuploidy by directly labeled multicolored probes and interphase fluorescence in situ hybridization. Mayo Clin Proc 1998;73;132-137)

Specimen Required: Please provide a reason for referral with each specimen. The laboratory will not delay or reject testing if this information is not provided, but appropriate testing and interpretation may be compromised.

Formalin-fixed, paraffin-embedded tissue block. Include 1 hematoxylin-and-eosin stained slide.

NOTE: Please complete a “Hematopathology/Molecular Oncology/Cytogenetics Request Form” (Supply T241) or a “MayoConnect Additional Test Information Form” (Supply T357) and forward it with the specimen.

Reference Values:

Normal range is <20% for triploidy, <13% for polyploidy, and <3% for individual chromosome trisomy.

Analytic Time: 1-4 days

Days Set Up: Monday through Friday

Fee: $376.60

CPT Code: 88271/x3 DNA probe, each

88275/Interphase in situ hybridization

88313/Goup II, all other, (eg, iron, trichrome) except immunocytochemistry and immunoperoxidase stains, each

88365/Interpretation and report
Lipoprotein Associated Phospholipase A2 (PLAC Test), Serum #81043

Clinical
Lipoprotein-associated phospholipase A2 (Lp-PLA2) is a 50 KD protein associated primarily with low-density lipoprotein (LDL) in human serum. Lp-PLA2 is expressed by macrophages and is upregulated in atherosclerotic lesions. There is increasing evidence that the oxidized phospholipid products of Lp-PLA2 contribute to the development and progression of atherosclerosis by their ability to attract monocytes and contribute to foam cell formation, among other proinflammatory actions. Levels of Lp-PLA2, as measured by a specific immunoassay, were shown to be significantly elevated in patients with angiographically proven coronary artery disease when compared with age matched-controls. In addition, a retrospective case-control study using specimens from hypercholesterolemic men in the West of Scotland Coronary Prevention Study (WOSCOPS) showed a 2-fold greater risk of coronary heart disease for subjects in the upper quintile of Lp-PLA2 levels. Furthermore, Lp-PLA2 was shown to be independent of other markers of inflammation: C-reactive protein, white cell count, and fibrinogen.

Useful For
A potential new risk factor for coronary artery disease

Interpretation
Patients with results in the upper quintile (females: >300 ng/mL; males: >330 ng/mL) show a 2-fold greater risk of coronary heart disease.

Cautions
The utility of this assay is not firmly established and should be used on only a clinical trial or research basis.

Reference

Method
This test utilizes a microtiter enzyme-linked immunosorbent assay (ELISA) using a monoclonal anti-Lp-PLA2 to capture Lp-PLA2 followed by a horseradish peroxidase-linked monoclonal anti-Lp-PLA2. Enzyme substrate is then added and color develops (450 nm) that is proportional to the Lp-PLA2 content. (Caslake MJ, Packard CJ, Suckling KE, et al: Lipoprotein-associated phospholipase A(2), platelet-activating factor acetylhydrolase: a potential new risk factor for coronary artery disease. Atherosclerosis 2000;150:413-419)
Test Title: Lipoprotein Associated Phospholipase A2 (PLAC Test), Serum #81043

Specimen Required: Draw blood in a plain, red-top tube(s). Spin down and send 1.0 mL of serum frozen in plastic vial.

Reference Values: Males: ≥16 years: 134-480 ng/mL  
Females: ≥16 years: 93-472 ng/mL

Analytic Time: 14 days
Days Set Up: Monday through Friday
Fee: $148.70
CPT Code: 83520