

HOTLINE: Effective August 19, 2019

0051368 RhD Gene (RHD) Copy Number

RHD

## **Interpretive Data:**

Background Information for RhD Gene (RHD) Copy Number:

Characteristics: Fetal or neonatal erythroblastosis and hydrops.

Incidence of RHD-negative genotype: 15 percent Caucasians, 5 percent African Americans, less than 1 percent Asians

Inheritance: Autosomal recessive

Cause: Maternal-fetal Rh D antigen incompatibility

**Methods:** Determine the presence of the *RHD* exons 5, 7, and a 37 base pair insertion in the intron 3/exon 4 boundary by PCR and fluorescence monitoring. Allelic height ratios are used to determine the number of copies of *RHD* as compared to *RHCE*.

**Limitations:** Bloody amniotic fluid specimens may give false-negative results because of maternal cell contamination; specificity may be compromised by mutations in primer sites or those outside the *RHD* exons examined; fetuses predicted to be unaffected should continue to be monitored by noninvasive means. Diagnostic errors can occur due to rare sequence variations.

Analytic Sensitivity and Specificity: Greater than 99 percent

Clinical Sensitivity: Greater than 98 percent

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

See Compliance Statement C: www.aruplab.com/CS

## HOTLINE NOTE: There is a clinically significant charting name change associated with this test.

Change the charting name for component 0051369, RhD Antigen (RhD) Specimen to RhD Gene (RHD) Copy Number Specimen. Change the charting name for component 0050422 RhD, Antigen (RhD) Genotyping to RhD Gene (RHD) Copy Number.