



<u>Procedure</u>	<u>Result</u>	<u>Units</u>	<u>Ref Interval</u>	<u>Accession</u>	<u>Collected</u>	<u>Received</u>	<u>Reported/ Verified</u>
RhD Gene (RHD) Copy Number Specimen	Whole Blood			19-168-900027	17-Jun-19 08:55:00	17-Jun-19 08:55:00	17-Jun-19 08:58:34
RhD Gene (RHD) Copy Number	1 copy	f		19-168-900027	17-Jun-19 08:55:00	17-Jun-19 08:55:00	17-Jun-19 08:58:34

17-Jun-19 08:55:00 RhD Gene (RHD) Copy Number:

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Indication for testing: Determine parental RhD zygosity to assess risk for alloimmune hemolytic disease in offspring.

Heterozygous: One copy of the RhD allele was detected in this whole blood sample, predictive of an RhD-positive phenotype in this individual.

This result has been reviewed and approved by Rong Mao, M.D.

17-Jun-19 08:55:00 RhD Gene (RHD) Copy Number:  
 BACKGROUND INFORMATION: 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  
**CLINICAL SENSITIVITY:** Greater than 98 percent.  
**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.  
**ANALYTICAL SENSITIVITY AND SPECIFICITY:** Greater than 99 percent.  
**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.

For quality assurance purposes, ARUP Laboratories will provide confirmation of the above result at no charge for amniotic specimens. Following delivery, please collect a cord blood sample from the infant in a lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A) tube. Please specify on the test request form that this is a confirmatory study to be performed at no charge. Please provide the mother's name for specimen identification purposes.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

\* Abnormal, # = Corrected, C = Critical, f = Footnote, H = High, L = Low, t = Interpretive Text, @ = Reference Lab