ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]1994
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Huntington Disease (HD) CAG Repeat Expansion, Fetal

ARUP test code 3019937

Maternal Contamination Study Fetal Spec	Fetal Cells Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood
Huntington Disease Fetal Specimen	Cultured Amnio
Huntington Disease Fetal Allele 1	42
Huntington Disease Fetal Allele 2	17
Huntington Disease Fetal Interpretation	See Note Indication for Testing: Prenatal diagnosis for Huntington disease (HD). Interpretation: According to information provided to ARUP Laboratories, the mother of this fetus is reported to harbor an HTT allele with 42 CAG repeats. One HTT allele in the full penetrance affected range and one HTT allele in the normal range were identified in this prenatal sample. This result is consistent with a diagnosis of Huntington disease (HD) in this fetus. Recommendations: Genetic consultation is recommended. This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Huntington Disease (HD) CAG Repeat Expansion, Fetal
CHARACTERISTICS: Neurodegenerative disorder causing progressive cognitive, motor, and psychiatric disturbances typically beginning at 35-44 years of age. An estimated 5 percent of individuals with HD are symptomatic as juveniles and 25 percent of individuals after age 50.
INCIDENCE: 1 in 15,000.
INHERITANCE: Autosomal dominant.
CAUSE: Expanded number of CAG repeats in the HTT gene. HD allele with reduced penetrance 36-39 CAG repeats; HD allele with full penetrance 40 or more CAG repeats.
CLINICAL SENSITIVITY AND SPECIFICITY: 99 percent.
METHODOLOGY: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis. Repeat sizing precision is +/- 2 for alleles less than or equal to 50 repeats, +/- 3 for alleles with 51 to 75 repeats, and +/- 4 for alleles greater than 75 repeats.
ANALYTIC SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Other neurodegenerative disorders will not be detected. Diagnostic errors can occur due to rare sequence variations. Prenatal specimens with maternal cell contamination may give false-negative results.

Phenotype	Number of CAG Repeats
Normal allele	less than or equal to 26
Mutable normal (intermediate) allele	27-35
HD allele with reduced penetrance	36-39
HD allele with full penetrance	greater than or equal to 40

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Fetal Specimen	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Fetal Allele 1	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Fetal Allele 2	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Huntington Disease Fetal Interpretation	25-295-114068	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical