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**PATIENT REPORT**

Patient Age/Sex: 32 years Female

**Specimen Collected: 13-Aug-25 15:40**

**Huntington Disease CAG Repeat, Fetal** | **Received: 13-Aug-25 15:40** **Report/Verified: 15-Aug-25 11:42**

| Procedure | Result | Units | Reference Interval |
|---|---|---|---|
| Maternal Contamination Study Fetal Spec | Fetal Cells [f1] | | |
| Maternal Contam Study,Maternal Spec | Whole Blood | | |
| Huntington Disease Fetal Specimen | Cultured Amnio | | |
| Huntington Disease Fetal Allele 1 | 45 | | |
| Huntington Disease Fetal Allele 2 | 19 | | |
| Huntington Disease Fetal Interpretation | See Note [f2 i1] | | |

## Result Footnote

f1: Maternal Contamination Study Fetal Spec

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

f2: Huntington Disease Fetal Interpretation

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 45 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.

Please send any postnatal outcomes concerning this prenatal result to ARUP Genetic Counseling fax: 801-584-5236.

This result has been reviewed and approved by [redacted]

## Test Information

i1: Huntington Disease Fetal Interpretation

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

## Test Information

i1: Huntington Disease Fetal Interpretation

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.