Ordered Items
HBB Deletion/Duplication

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<th>REFERENCE INTERVAL</th>
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No Copy Number Variation Detected in the HBB gene

Interpretation:
No pathogenic variants in the HBB gene were detected by deletion/duplication analysis.

The indication for HBB deletion/duplication analysis was carrier screening or a suspected diagnosis of beta thalassemia.

Recommendation:
Genetic counseling is recommended to discuss the clinical implications of this result. Genetic counselors are available for health care providers to discuss this result further at (800)345-GENE.

Comments:
It cannot be excluded that pathogenic copy number variations in HBB were missed due to limitations inherent to the Deletion/Duplication analysis method used here. Any interpretation given here should be clinically correlated with available information about presentation and relevant family history of the patient.

Methods/Limitations:
The multiple-ligation-probe amplification assay (MLPA) was performed to detect copy number variations (deletions and/or duplications) in the HBB gene. Rare sequence variations may affect the performance of this assay. Precise breakpoints for deletions and duplications are not determined. The presence of point mutations in the HBB gene cannot be excluded as this assay does not detect these changes.

References:
2) Galanello R and Origa R. Beta Thalassemia. Orphanet J
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Disclaimer:
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Director Review  Comment:  Melissa Hayden,Ph.D.,FACMG

For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087
**Test:**  HBB Deletion/Duplication

**Result:**  No Copy Number Variation Detected in the HBB gene

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Client/Sending Facility:
LabCorp Test Master
Test Account
5450 Millstream Road
MCLEANVILLE, NC  27301
Ph: (336)436-8645
POE-00

LCLS Specimen Number: 026-225-9628-0
Patient Name: NORMAL, 252240
Date of Birth: 01/11/1990
Gender: F
Patient ID:

Account Number: 90000999
Ordering Physician:
Specimen Type: BLOOD
Date Collected: 01/26/2019
Date Received: 02/04/2019

Melissa Hayden, Ph.D., FACMG

Arundhati Chatterjee, MD
Medical Director

Testing performed by Laboratory Corporation of America Holdings,
1912 TW Alexander Drive , RTP , NC , 27709-0000   (800) 735-4087
**Patient Details**

- **DOB:** 01/11/1990
- **Age (y/m/d):** 029/00/15
- **Gender:** F
- **SSN:**
- **Patient ID:**

**Specimen Details**

- **Date collected:** 01/26/2019 0000 Local
- **Date received:** 01/26/2019
- **Date entered:** 01/26/2019
- **Date reported:** 02/09/2019 0000 ET

**Physician Details**

- **Ordering:**
- **Referring:**
- **ID:**
- **NPI:**

**Ordered Items**

- **HBB Deletion/Duplication**

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**Interpretation:**

Homozygous for pathogenic deletion of the entire HBB gene

**Method/Pathology:**

- **Homozygous deletion of the entire HBB gene was detected by deletion/duplication analysis.** While the exact breakpoints of this deletion cannot be determined by this assay, the deletion encompasses the entire HBB gene with one breakpoint lying outside the beta globin gene cluster and the other breakpoint located between the HBB and HBD gene.

**Similar sized deletions have been previously reported in association with Beta-Thalassemia; thus this homozygous deletion has been classified as pathogenic.** This result supports a diagnosis of Beta-Thalassemia or indicates a predisposition for Beta-Thalassemia.

**Recommendation:**

Genetic counseling is recommended to discuss the clinical implications of this result.

**Comments:**

- It cannot be excluded that pathogenic copy number variations in HBB were missed due to limitations inherent to the Deletion/Duplication analysis method used here. Any interpretation given here should be clinically correlated with available information about presentation and relevant family history of the patient.

**Methods/Limitations:**

- The multiple-ligation-probe amplification assay (MLPA) was performed to detect copy number variations (deletions and/or duplications) in the HBB gene. Rare sequence variations may affect the performance of this assay. Precise breakpoints for deletions and duplications are not determined. The presence of point mutations in the HBB gene cannot be excluded as this assay does not detect these changes.

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Director Review

Comment: 01

Melissa Hayden, Ph.D., FACMG

PDF

01 TG LabCorp RTP 1912 TW Alexander Drive, RTP, NC 27709-0150

Dir: Arundhati Chatterjee, MD

For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087
LCLS Specimen Number: 026-225-9629-0
Patient Name: ABNORMAL, 252240
Date of Birth: 01/11/1990
Gender: F
Patient ID: 
Indications: 

Test: HBB Deletion/Duplication

Result: Homozygous for pathogenic deletion of the entire HBB gene

Interpretation:
A homozygous deletion of the entire HBB gene was detected by deletion/duplication analysis. While the exact breakpoints of this deletion cannot be determined by this assay, the deletion encompasses the entire HBB gene with one breakpoint lying outside the beta globin gene cluster and the other breakpoint located between the HBB and HBD gene. Similar sized deletions have been previously reported in association with Beta-Thalassemia; thus this homozygous deletion has been classified as pathogenic. This result supports a diagnosis of Beta-Thalassemia or indicates a predisposition for Beta-Thalassemia.

Recommendation:
Genetic counseling is recommended to discuss the clinical implications of this result.

Comments:
It cannot be excluded that pathogenic copy number variations in HBB were missed due to limitations inherent to the Deletion/Duplication analysis method used here. Any interpretation given here should be clinically correlated with available information about presentation and relevant family history of the patient.

Methods/Limitations:
The multiple-ligation-probe amplification assay (MLPA) was performed to detect copy number variations (deletions and/or duplications) in the HBB gene. Rare sequence variations may affect the performance of this assay. Precise breakpoints for deletions and duplications are not determined. The presence of point mutations in the HBB gene cannot be excluded as this assay does not detect these changes.

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Date of Birth: 01/11/1990
Gender: F
Patient ID:

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Ordering Physician:
Specimen Type: BLOOD
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