## Chromosome Microarray

**Tests Ordered**

<table>
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<th>Test</th>
<th>Result</th>
<th>Flag</th>
<th>Units</th>
<th>Reference Interval</th>
<th>Lab</th>
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<td>arr(1−22,X)x2</td>
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The whole genome chromosome SNP microarray (Reveal) analysis was normal. No significant DNA copy number changes or copy neutral regions within the 2.695 million region specific SNP and structural targets were detected under the present reporting criteria indicated below. Archival records can be re-examined on request as new clinically significant genes are identified.

Methodology:

SNP microarray analysis was performed using the Affymetrix Cytoscan HD platform which uses over 743,000 SNP probes and 1,953,000 NPCN probes with a median spacing of 0.88 kb. 250ng of total genomic DNA extracted from lymphocytes was digested with NspI and then ligated to NspI adaptors, respectively, and amplified using Titanium Taq with a GeneAmp PCR System 9700. PCR products were purified using AMPure beads and quantified using NanoDrop 8000. Purified DNA was fragmented and biotin labeled and hybridized to the Affymetrix Cytoscan HD GeneChip. Data was analyzed using Chromosome Analysis Suite. The analysis is based on the GRCh37/hg19 assembly.
Positive evaluation criteria include:
* DNA copy gain/loss within a known clinically significant gene region of 50 Kb or greater.
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* UPD testing is recommended for patient results demonstrating a long contiguous region of homozygosity in a single chromosome of >20 Mb interstitially or >10 Mb telomerically (15 and 8 Mb, respectively, for imprinted chromosomes).
* Contiguous homozygosity of >8 Mb within multiple chromosomes suggests common descent. These regions of potential recessive allele risk are designated.
* A high level of allele homozygosity due to numerous contiguous short runs (associated with a geographically or socially limited gene pool) is reported at the 99th percentile.

Truly balanced chromosome alterations will not be detected by this analysis. The threshold for mosaicism is variable, depending on the size of segment. Empiric studies have detected whole chromosome 22 mosaicism below 10.0%. CNVs cited in the Database of Genomic Variants are not reported.

Director Review:  Comment:  01
M. Katharine Rudd, PhD, FACMG
Preauthorization  Will Follow  01

01  YU  LabCorp RTP  Dir: Arundhati Chatterjee, MD
1904 TW Alexander Drive Suite C, RTP, NC 27709-0153
For inquiries, the physician may contact Branch: 800-222-7566  Lab: 800-735-4087
**MICROARRAY RESULT:** NORMAL FEMALE

**INTERPRETATION:**

\[\text{arr}(1\text{-}22, X)\times 2\]

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LCLS Specimen Number: 238-225-9003-0
Patient Name: SAMPLE REPORT, 510002
Date of Birth: 03/16/1963
Gender: F
Patient ID: 
Lab Number: YU16-67587 G

Account Number: 90000999
Ordering Physician: 
Specimen Type: BLOOD
Client Reference: 
Date Collected: 08/25/2016
Date Received: 08/26/2016

M. Katharine Rudd, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings, 1904 TW Alexander Drive, RTP, NC, 27709-0153 (800) 345-4363
Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD. Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

This document contains private and confidential health information protected by state and federal law.
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<tr>
<td>Interpretation</td>
<td>7.36 MB TERMINAL DELETION OF 2Q37.1-&gt;2QTER</td>
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<td>arr 2q37.1q37.3(235,381,633-242,738,117)x1</td>
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The whole genome SNP microarray (Reveal) analysis detected a terminal deletion of the chromosomal segment listed above. This deletion includes numerous OMIM genes [start:ARL4C to end:GAL3ST2] which are likely to contribute to the resultant abnormal phenotype.

The 2q37 microdeletion syndrome, also called Albright Hereditary Osteodystrophy-Like Syndrome, Brachydactyly-Mental Retardation Syndrome is characterized by Albright hereditary osteodystrophy (AHO)-like phenotype (developmental delay/intellectual disability, brachymetaphalangy of digits 3-5, short stature, obesity), typical facial features, autism spectrum disorder, joint hypermobility/dislocation, and scoliosis. The HDAC4 gene is considered to be critical to most of the syndromic features of the 2q37 microdeletion syndrome. Most deletions have occurred de novo (see reference).

No other DNA copy number changes or copy neutral ROH were detected within the present reporting criteria.

Parental chromosome analysis OR FISH utilizing a 2q subtelomere probe is recommended to confirm a de novo origin.
and rule out a balanced rearrangement with high recurrence risk. Genetic counseling is recommended.

The follow-up parental blood (green top sodium heparin tube) may be submitted under the test code 052215 (chromosomes)/511760 (FISH). Charges will apply. Please reference the proband name, date of birth, and specimen number when submitting parental or familial samples. Billing policy details are available for view on www.labcorp.com.

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Director Review: M. Katharine Rudd, PhD, FACMG

Comment: Will Follow

Preauthorization

01
MICROARRAY RESULT: 7.36 MB TERMINAL DELETION OF 2Q37.1->2QTER

INTERPRETATION: 2Q37 DELETION SYNDROME

arr 2q37.1q37.3(235,381,633-242,738,738,117)x1

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Doherty ES, Lacbawan F. 2q37 Microdeletion Syndrome. 2007 May 3 [Updated 2013 Jan 31]. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington,
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