### Chromo Leuk/Lymph Rfx CMA

**Specimen Type**
- Comment: BONE MARROW

**Cells Counted**
- 20

**Cells Analyzed**
- 20

**Cells Karyotyped**
- 2

**GTG Band Resolution Achieved**
- 400

**Cytogenetic Result**
- Comment: 46,XX[20]

**Interpretation**
- Comment: NORMAL FEMALE KARYOTYPE

Cytogenetic analysis of unstimulated cultures revealed a female karyotype with an apparently normal GTG banding pattern in all cells analyzed.

A normal karyotype does not rule out clonal molecular alterations below the resolution of light microscopy. In some cases additional molecular or FISH testing may be warranted, eg. BCR/ABL, PML/RARA, T and B cell rearrangements, etc. In addition, indolent clones may have mitotic rates below the level of cytogenetic detection within the standard 20 cells. A FISH panel (test #510830) may be effectively used in low mitotic multiple myeloma, for example, to detect high incidence, prognosis related alterations. *A test option for a whole genome SNP microarray is available (test #510146) that can resolve genomic imbalance at a level of sensitivity over 200 times cytogenetic resolution. A cancer gene set is analyzed at an exon level resolution. The array can also detect acquired copy neutral-LOH based clonal evolution associated with gene mutations. The array is especially useful in diagnostically uncertain MDS/MPN/LPD and, as opposed to chromosome analysis, does not require mitotic activity, offering clonal detection from peripheral blood.*
This testing can be performed from the current remaining sample, if available. Call (800)533-0567 x 4060.

**Director Review:**

Inder K. Gadi, PhD, FACMG

**Reflex**

Microarray analysis is indicated for this specimen. Final report will follow under separate cover.

**Specimen Type**

BONE MARROW

**# of Genotyping Targets**

2695000

**Diagnosis**

NORMAL FEMALE

Results for this test are for research purposes only by the assay's manufacturer. The performance characteristics of this product have not been established. Results should not be used as a diagnostic procedure without confirmation of the diagnosis by another medically established diagnostic product or procedure.

**Interpretation**

arr(1-22,X)x2

The whole genome chromosome SNP microarray (Reveal) analysis was normal. No clinically significant copy number or copy neutral alterations were detected within the reporting criteria indicated below.

**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, within genes. 250ng of total genomic DNA extracted was digested with NspI and then ligated to NspI adaptors, 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 or including a known clinically significant cancer related gene (530 in database) of 50 Kb or greater.
**TESTS** | **RESULT** | **FLAG** | **UNITS** | **REFERENCE INTERVAL** | **LAB**
---|---|---|---|---|---
* DNA copy number loss of >1 Mb or gain >2 Mb outside known clinical oncology significant regions with at least one OMIM annotated gene of possible clinical significance.

* Contiguous allele homozygosity >8Mb through the telomere of a single chromosome is consistent with copy-neutral loss of heterozygosity (CN-LOH). These regions designate clonal evolution associated with the acquisition of homozygosity for a gene mutation within the homozygotic stretch. Candidate gene(s) will be indicated.

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: Inder K. Gadi, PhD, FACMG

**01**

For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087
LCLS Specimen Number: 285-225-9103-0  
Patient Name: SAMPLE REPORT, 511040  
Date of Birth: 06/12/1985  
Gender: F  
Patient ID:  
Lab Number: YU16-81505 B  
Indications: NORMAL FEMALE  
Account Number: 90000999  
Ordering Physician:  
Specimen Type: BONE MARROW  
Client Reference:  
Date Collected: 10/11/2016  
Date Received: 10/14/2016  
Date Reported: 10/14/2016  
Test: CHROMO LEUK/LYMPH RFX CMA  
Cells Counted: 20  
Cells Analyzed: 20  
Cells Karyotyped: 2  
Band Resolution: 400  

CYTOGENETIC RESULT: 46,XX[20]  
INTERPRETATION: NORMAL FEMALE KARYOTYPE  

Cytogenetic analysis of unstimulated cultures revealed a female karyotype with an apparently normal GTG banding pattern in all cells analyzed.  
A normal karyotype does not rule out clonal molecular alterations below the resolution of light microscopy. In some cases additional molecular or FISH testing may be warranted, eg. BCR/ABL, PML/RARA,T and B cell rearrangements, etc. In addition, indolent clones may have mitotic rates below the level of cytogenetic detection within the standard 20 cells. A FISH panel (test #510830) may be effectively used in low mitotic multiple myeloma, for example, to detect high incidence, prognosis related alterations.

*A test option for a whole genome SNP microarray is available (test #510146) that can resolve genomic imbalance at a level of sensitivity over 200 times cytogenetic resolution. A cancer gene set is analyzed at an exon level resolution. The array can also detect acquired copy neutral-LOH based clonal evolution associated with gene mutations. The array is especially useful in diagnostically uncertain MDS/MPN/LPD and, as opposed to chromosome analysis, does not require mitotic activity, offering clonal detection from peripheral blood.

This testing can be performed from the current remaining sample, if available. Call (800)533-0567 x 4060.
LCLS Specimen Number: 285-225-9103-0
Patient Name: SAMPLE REPORT, 511040
Date of Birth: 06/12/1985
Gender: F
Patient ID: 
Lab Number: YU16-81505 B

Account Number: 90000999
Ordering Physician: 
Specimen Type: BONE MARROW
Client Reference: 
Date Collected: 10/11/2016
Date Received: 10/14/2016

Inder K. Gadi, PhD, FACMG
Board Certified Cytogeneticist

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

Integrated Oncology 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.
Chromo Leuk/Lymph Rfx CMA; Chromosomes Leuk/Lymph; Analyze 20–25 cells

<table>
<thead>
<tr>
<th>Tests Ordered</th>
<th>RESULT</th>
<th>FLAG</th>
<th>UNITS</th>
<th>REFERENCE INTERVAL</th>
<th>LAB</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chromo Leuk/Lymph Rfx CMA</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Specimen Type</td>
<td>Comment:</td>
<td>01</td>
<td>BONE MARROW</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cells Counted</td>
<td></td>
<td>20</td>
<td>01</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cells Analyzed</td>
<td></td>
<td>20</td>
<td>01</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cells Karyotyped</td>
<td></td>
<td>2</td>
<td>01</td>
<td></td>
<td></td>
</tr>
<tr>
<td>GTG Band Resolution Achieved</td>
<td></td>
<td>400</td>
<td>01</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cytogenetic Result</td>
<td>Comment:</td>
<td>01</td>
<td>46,XX,t(9;22)(q34;q11.2)[20]</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Interpretation</td>
<td>Comment:</td>
<td>01</td>
<td>PHILADELPHIA TRANSLOCATION KARYOTYPE (CML/ALL)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Cytogenetic analysis of GTG banded metaphases has revealed the presence of the Philadelphia translocation in all cells analyzed. This may be observed in CML or ALL (the latter with an adverse prognosis). Rare cases in AML have also been reported. No other karyotype changes indicative of an accelerated or blastic phase of CML were evident.

Monitoring of response to therapy may be facilitated by fluorescence in situ hybridization (FISH) or real time PCR, if not already ordered. Adjunctive dual fusion FISH has very low background (<1%) in the typical 200 interphase cell analysis offered, quantitative PCR has the highest sensitivity (call the number below).

Director Review: Comment: 01
Inder K. Gadi, PhD, FACMG
Reflex 01
Microarray analysis is not indicated on this specimen.
<table>
<thead>
<tr>
<th>Account Number</th>
<th>Patient ID</th>
<th>Control Number</th>
<th>Date and Time Collected</th>
<th>Date Reported</th>
<th>Sex</th>
<th>Age(Y/M/D)</th>
<th>Date of Birth</th>
</tr>
</thead>
<tbody>
<tr>
<td>90000999</td>
<td>285-225-9104-0</td>
<td>01488475</td>
<td>10/11/16 00:00</td>
<td></td>
<td>F</td>
<td>31/03/29</td>
<td>06/12/85</td>
</tr>
</tbody>
</table>

**Patient Name**: Arundhati Chatterjee, MD

**Address**: 1904 TW Alexander Drive Suite C, RTP, NC 27709-0153

**Contact Information**: For inquiries, the physician may contact Branch: 800-222-7566 Lab: 800-735-4087
LCLS Specimen Number: 285-225-9104-0
Account Number: 90000999

Patient Name: SAMPLE REPORT, 511040
Ordering Physician: BONE MARROW

Date of Birth: 06/12/1985
Client Reference: Date Collected: 10/11/2016
Gender: F
Date Received: 10/14/2016
Patient ID: Lab Number: YU16-81511 B

Indications: SAMPLE REPORT FOR ABNORMAL FEMALE
Date Reported: 10/14/2016

Test: CHROMO LEUK/LYMPH RFX CMA

Cells Counted: 20
Cells Karyotyped: 2
Cells Analyzed: 20
Band Resolution: 400

CYTOGENETIC RESULT: 46,XX,t(9;22)(q34;q11.2)[20]

INTERPRETATION: PHILADELPHIA TRANSLOCATION KARYOTYPE (CML/ALL)

Cytogenetic analysis of GTG banded metaphases has revealed the presence of the Philadelphia translocation in all cells analyzed. This may be observed in CML or ALL (the latter with an adverse prognosis). Rare cases in AML have also been reported. No other karyotype changes indicative of an accelerated or blastic phase of CML were evident.

Monitoring of response to therapy may be facilitated by fluorescence in situ hybridization (FISH) or real time PCR, if not already ordered. Adjunctive dual fusion FISH has very low background (<1%) in the typical 200 interphase cell analysis offered, quantitative PCR has the highest sensitivity (call the number below).

Inder K. Gadi, 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 Oncology 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.