Test Result

Positive Trisomy 7

Lab Director’s Comments
This specimen showed an increased representation of chromosome 7, suggestive of trisomy 7. Genetic counseling and clinical correlation are recommended. Trisomy 7 mosaicism is commonly reported. Confined placental mosaicism (CPM) is likely. Chromosome 7 is known to be imprinted. Therefore, subsequent trisomy rescue in fetal tissue carries residual risk for UPD. Confirmatory testing is required if fetal confirmation and clinical interpretation of the suspected event are desired. Please refer to the “Performance” and “Limitations of the Test” sections of this laboratory report for additional information.

Fetal Fraction: 7%

An approximate 159.00Mb gain of chromosome 7 material was observed, suggestive of trisomy 7.
### Result Table

<table>
<thead>
<tr>
<th>Content</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>AUTOSOMAL ANEUPLOIDIES</strong></td>
<td></td>
</tr>
<tr>
<td>Trisomy 21 (Down syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>Trisomy 18 (Edwards syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>Trisomy 13 (Patau syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td><strong>Other autosomal aneuploidies</strong></td>
<td>Positive</td>
</tr>
<tr>
<td><strong>SEX CHROMOSOME ANEUPLOIDIES</strong></td>
<td></td>
</tr>
<tr>
<td>Fetal sex</td>
<td>Consistent with female</td>
</tr>
<tr>
<td>Monosomy X (Turner syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>XYY (Jacobs syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>XXY (Klinefelter syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>XXX (Triple X syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td><strong>GENOME-WIDE COPY NUMBER VARIANTS ≥7 Mb</strong></td>
<td></td>
</tr>
<tr>
<td>Gains/Losses ≥7 Mb</td>
<td>Negative</td>
</tr>
<tr>
<td><strong>SELECT MICRODELETIONS</strong></td>
<td></td>
</tr>
<tr>
<td>22q11 deletion (associated with DiGeorge syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>15q11 deletion (associated with Prader-Willi / Angelman syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>11q23 deletion (associated with Jacobsen syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>8q24 deletion (associated with Langer-Giedion syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>5p15 deletion (associated with Cri-du-chat syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>4p16 deletion (associated with Wolf-Hirschhorn syndrome)</td>
<td>Negative</td>
</tr>
<tr>
<td>1p36 deletion syndrome</td>
<td>Negative</td>
</tr>
</tbody>
</table>