CombiSNP™ Array for Pregnancy Loss Results

**Results**
- Female with a variant of uncertain significance

**ISCN**
- arr 1q21.1q21.2(146,295,308-147,826,789)x1

**INTERPRETATION:**

The region on 1q21.1-q21.2 is known to show copy number variation in normal individuals. However, reciprocal microdeletions and microduplications involving this region have also been associated with a range of developmental disorders including: intellectual disability, autism spectrum disorders, abnormal head size and other variable features. Deletions of this region also appear to be enriched in patients with congenital heart defects. This deletion may be inherited from a parent or be a de novo event. A parent with this microdeletion may have a normal phenotype or an abnormal phenotype that is similar to, but often less severe than that of their child.

There is no evidence to suggest that this microdeletion contributed to the miscarriage. However, for the purposes of genetic counseling, the parents may wish to undergo testing to determine whether this deletion is de novo or is also present in one of them. Parental studies should be offered only in conjunction with genetic counseling, as there is the potential to diagnose an asymptomatic parent. If parental studies are desired following genetic counseling, chromosomal microarray analysis can be performed for an additional charge. For parental studies, please collect 4 mL of blood in an EDTA (purple top) tube and send along with a completed CombiMatrix Test Requisition Form. For assistance, please contact Client Services at (800) 710-0624.

**References:**
RECOMMENDATION: Genetic counseling is recommended to discuss the implications of these results and the option of parental karyotyping and/or microarray analysis for assistance in planning future pregnancies. If you wish to identify a genetic counselor in your local area, you may wish to consult the National Society of Genetic Counselors' website at www.nsgc.org.

GENES: Genes with loss of a copy number on 1q21.1-q21.2(146,295,308-147,826,789): HYDIN2, LOC728989, PRKAB2, Pdia3p, FMO5, CHD1L, LINC00624, BCL9, ACP6, GJA5, GJA8, GPR89B, GPR89C, PDZK1P1, NBPF11, NBPF24, MIR5087.