STAT Prenatal CNGnome

Test Code	D0900E
Test Summary	STAT prenatal CNV analysis utilizing low pass genome sequencing Assay (8x).
Turn-Around-Time (TAT)*	10 - 12 days
Acceptable Sample Types	Cultured Amniocytes Cultured Chorionic Villi DNA, Isolated Products of Conception
Acceptable Billing Types	Self (patient) Payment Institutional Billing Commercial Insurance

Indications for Testing

Abnormal NIPT result
Abnormal maternal serum screen
Advanced maternal age

Test Description

This STAT prenatal test detects large copy number changes (CNV) greater than or equal to 25kb throughout the genome and reliably detects chromosome uniparental disomy. Testing is performed utilizing low pass genome sequencing (8x).

Test Methods and Limitations

Direct sequencing of genomic DNA was performed using 2X150bp reads on Illumina next generation sequencing (NGS) systems at a mean coverage of 8X in the target region. Alignment to the human reference genome (hg19) was performed and copy number variant (CNV) calls made using the NxClinical software v5.1 (BioDiscovery, Inc., El Segundo, CA). CNVs meeting internal quality assessment guidelines are confirmed by real time quantitative PCR (qPCR) for records after results are reported. Some CNVs are confirmed by qPCR before reporting at a director's discretion. This assay cannot detect CNVs in regions of the genome that are not amenable to NGS and does not interrogate CNVs in mitochondrial DNA. This assay will not detect tandem repeats, balanced alterations (reciprocal translocations, Robertsonian translocations, inversions, and balanced insertions), point mutations, methylation abnormalities, genomic imbalances in segmentally duplicated regions and mosaicism; possible cases of mosaicism may be investigated at the discretion of the laboratory director. Small pathogenic CNVs within the exon, some small intragenic deletions or duplications, as well as complex rearrangements may not be detected. This assay has been validated to detect copy number variants >25 Kb and also has the ability to detect copy number changes such as homozygous deletions. For targeted CNV testing, smaller CNVs may be interrogated, analyzed, and reported per director discretion. This assay may not be able to discern between CNVs that are high copy number gains such as, duplication >=4X. CNVs involving genes with pseudogenes and pseudoexons may not be reliable detected or reported. Due to high similarity of certain regions on chromosome X and chromosome Y, CNVs in the following regions may not be detected for male patients (chrX: 60000-2699520; chrX:154930289-155260560; chrY:10000-2649520; chrY: :59033286-59363566).

Detailed Sample Requirements

Cultured Amniocytes

SPECIAL INSTRUCTIONS: Please contact a PKIG Laboratory genetic counselor for these requests.

Cultured Chorionic Villi

Collection Container(s):

Two T-25 flasks

Collection:

All prenatal specimens will be tested for maternal cell contamination (MCC). Send maternal blood (EDTA tube) for comparison. If blood is unavailable, we will accept genomic DNA and Saliva sample types.

Sample Condition: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at ?80% confluence.
Shipping: Cultures topped off with sterile medium and shipped immediately at ambient temperature by overnight express with arrival Monday-Friday only.

SPECIAL INSTRUCTIONS: For prenatal samples, PerkinElmer will provide a shipping label to use for shipping the sample to our lab. Please call 1 (866) 354-2910 to arrange this. At this time, you will also be connected to a laboratory genetic counselor to answer any questions about the testing.

DNA, Isolated

Collection:

Required DNA Quantity by Test Type*:

- **Next Generation Sequencing (NGS):** Send >1000 ng total gDNA @ >15 ng/?L. Please ship samples in 10mM Tris. Do not use EDTA.
- **Sanger Sequencing:** Send >500 ng total gDNA @ >15 ng/?L (varies by the size of the gene and the variants requested).
- **Non-Sanger Sequencing Tests:** Send >500 ng total gDNA @ >15 ng/?L.

Sample Condition: * Required DNA Quality: High molecular weight DNA (>12kb). A260/A280 reading should be ? 1.8. A260/230 a ratio range of 1.8 to 2.2. Contact the laboratory for specific amounts if total ng cannot be met.
Shipping: Ship overnight at ambient temperature.

SPECIAL INSTRUCTIONS:

- **Research Laboratories:** DNA extracted in research laboratories is not acceptable. Only under exceptional circumstances (e.g., proband not available) will DNA extracted in a research laboratory be accepted for clinical testing. Additional testing (e.g., of other family members) may be required to confirm results.
- **Laboratories outside the United States:** Non-US laboratories are not subject to CLIA regulations and will be reviewed on a case-by-case basis. Please call to speak with a laboratory genetic counselor prior to submitting a DNA sample from any non-CLIA certified laboratory.
- **Special Notes**: If extracted DNA is submitted, information regarding the method used for extraction should be sent along with the sample.

Products of Conception