TEST: Genome-wide oligonucleotide array CGH (105K)

DESCRIPTION: Whole-GENOME array-based comparative genomic hybridization (aCGH) is performed using the BlueGnome CytoChip oligo 2X105K microarray v1.1. This array contains ~105,000 oligonucleotide probes spaced at an average distance of 35 kb based on the NCBI build (36) of the human genome. This array can detect copy number changes >50 kb in 138 targeted regions (microdeletion/duplication loci) and >150 kb in the remainder of the genome. Copy number changes that have been published or identified in clinically normal individuals or that are of unknown clinical significance may not be reported. Results are confirmed by fluorescence in situ hybridization (FISH) or polymerase chain reaction (PCR) on a case by case basis depending upon the abnormal findings.

INDICATIONS:
- congenital anomalies
- developmental delay / mental retardation

SPECIMEN REQUIREMENTS: 5-8 cc (2 cc for infants and small children) in an EDTA tube. Send to CompGene at room temperature with a Requisition for Chromosome Analysis form as soon as possible. Microarray analysis requires high quality DNA so the blood sample should be as fresh as possible. May be refrigerated overnight. **Specimen must not be frozen.** Label tube with patient's name and medical record number.

REFERENCE VALUE: arr(1-22,X)x2 normal female or arr(1-22)x2,(XY)x1 normal male

TURN AROUND TIME: 2-3 weeks (may be faster depending upon the clinical situation)

CPT CODES: 88386x4 - array-based evaluation of multiple molecular probes (251-500 probes)