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Array Comparative Genomic Hybridization (array CGH) Analysis for Constitutional Abnormalities

*See link for *Array CGH Clinical Testing Handout using the Agilent 4x180k aCGH+SNP array*

Description	Turn Around	CPT codes
This test utilizes the Agilent 4x180k aCGH+SNP array, which is based on the ISCA (International Standards for Cytogenomic Arrays) Consortium design. This array contains ~110,000 oligo probes for the detection of copy number variations (CNVs), and ~60,000 SNP probes for the detection of uniparental disomy (UPD) and absence of heterozygosity (AOH). It contains high-density coverage for ~500 targeted regions with an average oligo probe spacing of ~5 kb or at least 20 probes per gene. These targeted regions include clinically relevant haploinsufficient genes, X-linked intellectual disability genes, all recurrent microdeletion/microduplication syndrome regions, and all unique subtelomeric and pericentromeric regions. In addition, it contains genome-wide backbone coverage with an average probe spacing of ~25 kb. This array is designed to detect CNVs with a minimum size of ~200-300 kb across the genome or smaller in the targeted genes/regions. It will also detect uniparental isodisomies and AOH.	~2 weeks	81229x1
Specimen requirements: One EDTA tube (purple top) tube and one sodium heparin (green top) tube, 4-5 cc per tube		