CombiSNP Array Analysis

INVITAE | CombiMatrix

PATIENT

Patient Name:	Doe, Jane
Date of Birth:	1/2/03
Accession #:	M14-00123456
Patient Gender:	Female
Medical Record:	1234567
Reason For Referral:	Developmental Delay

SPECIMEN

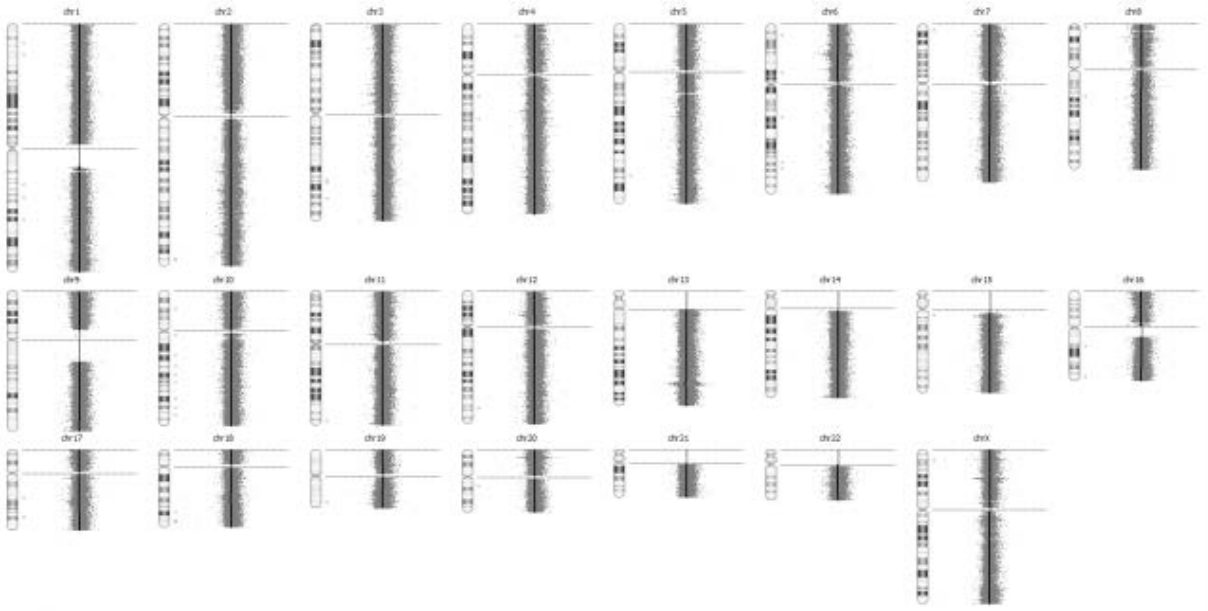
Collection Date:	1/2/2014
Received Date/Time:	1/3/2014 11:50:00 AM
Report Date/Time:	1/09/2014 8:35:17 AM
Specimen ID:	123-45-678
Specimen Type:	Whole Blood

FACILITY

Ordering Facility:	Anywhere Pediatrics
Ordering Physician:	Dr. John White
Address:	1234 Anyplace Ave. Suite 123
City/State/Zip:	Anywhere, CA 92000
Referring Physician:	Dr. Jane Black

Clinical History: ventricular septal defect

CombiSNP™ Array for Pediatric Analysis Results	
Results	Normal female genomic profile
ISCN	arr(1-22,X)x2



***INTERPRETATION:** An array profile consistent with a female sex chromosome complement was identified. No diagnostic copy number changes were observed.

Karine Hovanes

Karine Hovanes, Ph.D., FACMG
Laboratory Director
Electronic Signature

Richard Hockett Jr., MD, FACP, Medical Director. CLIA #05D1052995, CAP #7193645

METHOD: CombiSNP™ Array analysis was performed using a custom-designed Illumina single nucleotide polymorphism (SNP) array. This array contains >845,000 SNP markers covering both coding and non-coding human genome sequences. The median spatial resolution between probes is 1 Kb within gene rich regions and 5 Kb outside of gene-rich regions. Extracted DNA was evaluated for copy number changes involving ≥16 probes, and for regions of homozygosity of ≥5 Mb. Genomic imbalances are reported using UCSC Human Genome Build 19 (NCBI build 37, Feb 2009). Mosaicism for partial or whole chromosome aneuploidy is reported when present at or above the detection threshold of 15%. The CombiSNP™ Array lot number 9298189005 from Illumina was used. The technical component of this test was performed by CombiMatrix Diagnostics (Irvine, CA; CLIA #05D1052995).

DISCLAIMER: The CombiSNP™ Array is designed to identify copy number changes genome-wide, covering regions of known clinical significance (recognized microduplication/microdeletion syndromes), pericentromeric and subtelomeric regions, and the genomic backbone. As with any microarray testing, the CombiSNP™ Array does not detect: point mutations; small intragenic deletions or duplications; balanced chromosomal aberrations such as Robertsonian or reciprocal translocations, inversions, and balanced insertions; or imbalances in genomic regions that are not represented on the microarray. Carrier status for recessive disorders due to a deletion/duplication of a single gene is typically not reported. Copy number gains and losses in the regions that contain no known or suspected clinical associations are not reported. The possibility of consanguinity will be reported if the regions of homozygosity (ROH) comprise ≥10% of the entire genome Uniparental disomy testing will be recommended for cases in which ROH on an imprinted chromosome is detected in the absence of other large ROH on other chromosomes. Although this test can detect uniparental isodisomy, it cannot detect uniparental heterodisomy. Thus, for ROH ≥20 Mb on imprinted chromosomes, UPD testing will be recommended. If an X-linked or autosomal recessive disorder is clinically suspected, please contact us for a list of detected ROH. A list of the genes according to inheritance pattern can be obtained by visiting the University of Miami's Online SNP Evaluation Tool at: http://www.ccs.miami.edu/cgibin/ROH/ROH_analysis_tool.cgi. Clinical assessment is required to determine if any of these genes may be implicated.

The clinical implications of some of the reported findings may be unknown at this time. Normal microarray results do not rule out the possibility of a genetic disorder or syndrome that is due to a genetic alteration not detected or evaluated by this test. Consultation with a genetics professional is recommended for results interpretation. As a participant in the International Collaboration for Clinical Genomics (ICCG), this clinical cytogenetics laboratory contributes submitted clinical information and test results to a HIPAA compliant, de-identified public database as part of the NIH's effort to improve understanding of the relationship between genetic changes and clinical symptoms. Confidentiality is maintained. Patients may request to opt out of this scientific effort by calling the laboratory at 800.710.0624 and asking to speak with a laboratory genetic counselor.

CombiSNP™ Array was developed and its performance characteristics determined by CombiMatrix Diagnostics. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is indicated for clinical purposes. It should not be regarded as investigational or for research use only. CombiMatrix Diagnostics laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88) as qualified to perform high complexity clinical laboratory testing.