Interpretive Report

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CLIA # 07D2046796
CT State License # CL-0687

Patient Name:	[redacted]	*Lab Number:*	[redacted]
Birth Date:	[redacted]	*Sex:*	**M**
Ordered By:	**Conner, William**	*Date Collected:*	**08/15/2022**
Specimen Type:	**Buccal Swab**	*Date Received:*	**08/18/2022**
Client Order #:			
Patient ID:	[redacted]		
Indication:	**Abnormal hematolog finding on antenatal screening**		
Tests Ordered:	**MicroArray**		

Result

arr(X)x2,(Y)x1

Interpretation

Chromosomal Microarray Analysis (CMA) identified 2 copies of the X chromosome and a single copy of the Y chromosome. This genomic constitution is equivalent to a 47,XXY karyotype which is associated with clinical features of Klinefelter syndrome. The clinical presentation of Klinefelter syndrome is highly variable, ranging from ânear normalâ to significantly affected. Varying degrees of cognitive, social, behavioral, and learning difficulties are often reported in 47,XXY individuals. Physical appearance is typically normal until puberty. The phenotype in adults is characterized by small testes, hypergonadotropic hypogonadism, tall stature, and eunuchoid body proportions. Almost all patients are infertile. Given the great variability in the phenotype, it is estimated that up to 75% of patients remain undiagnosed with the majority of diagnoses occurring following a fertility workup in adulthood. Genetic counseling is recommended for this family to discuss the significance of this result.

Limitations:
The chromosomal microarray array used at Genesys Diagnostics (Illumina Infinium CytoSNP-850K) is used in this test for the sole purpose of identifying genomic chromosomal abnormalities. This microarray will detect aneuploidy, copy number gains, copy number losses, and regions of copy number neutral absence or loss of heterozygosity (cnAOH or cnLOH) for the loci represented on the microarray. Analysis is limited to detection of copy number changes that include at least 10 probes and regions of cnAOH that include at least 500 probes. Abnormalities at resolutions below this level are possible. Deletions of at least 500 Kb and duplications of at least 1000 kb are reported. LOH regions at least 5 Mb large are reported. Benign copy number variants are not reported. This test will not detect copy number variation in regions not covered on the microarray. This test will not detect balanced alterations (E.g. reciprocal translocations, Robertsonian translocations, inversions, balanced inversions), methylation anomalies and other epigenetic events, or point mutations.

Methodology:
Genomic DNA is extracted from the patient sample. Whole genome amplification is performed. Amplified gDNA is fragmented and hybridized to the Illumina Infinium CytoSNP-850K BeadChip. This microarray contains 850,000 probes for SNP markers.

This report **electronically signed by Jun Liao, PhD, FACMG at 10/10/2022 04:16:58 PM**

Genesys Diagnostics
8 Enterprise Lane
Oakdale, Connecticut 06370

Appointment Date: 11/23/2022

Patient: [redacted]
DOB: [redacted], 4-month-old male
In attendance: [redacted] (Parents)
Accession No. [redacted]

Reason for Referral: Pediatrician referral for Confirmation of child's karyotype
Date Collected: 08/15/2022
Date Reported: 10/10/2022
Results: Chromosomal microarray (CMA) of 47,XXY consistent with Klinefelter's Syndrome

Family History for both patient and partner: Unremarkable (see pedigree)
Ethnicity: Both parents report they are Caucasian

Discussion:
[redacted] had a noninvasive prenatal test (NIPT) during her pregnancy whereby she was told there was a high probability for the fetus to have the above finding. She was offered amniocentesis to confirm the finding but she refused given the risk for that procedure and wanted to wait for the birth of her child to have the results confirmed. She did develop preeclampsia and delivered the baby by C-Section at 36 weeks with a birth weight of 5 lbs.,10 oz.

The above results were discussed in that Klinefelter syndrome occurs as a result of a random error causing a male to be born with an extra sex (X) chromosome. It is not an inherited condition. We shared that between 1/500 to 1/1000 males are born with this finding and many do not find out about the diagnosis until adulthood. We discussed that signs and symptoms vary widely among individuals with this diagnosis whereby a potential range of possibilities exist from normal development to issues that can include cognitive, social, emotional and/or behavioral problems with best possible outcomes occurring through early intervention. Follow up through puberty is also recommended given the risk for male infertility. Parents have appointments already set up for their son to see a Pediatric Geneticist and an Endocrinologist so that further clinical evaluation and follow-up can be done to offset any problems that may arise as the child grows.

Recommendations:
We did discuss and do recommend that should the parents have any future pregnancies; the maternal age needs to be taken into account given that her risk for a child with a chromosomal abnormality increases the older the mother is and while NIPT is available it is still considered a screening tool and not a diagnostic test. As was offered to her with her previous pregnancy, confirmation of any abnormal NIPT is dependent upon amniocentesis for a definitive diagnosis.

Summary:
Confirmation of the child's chromosomal results were given along with discussion of possible outcomes for the child's growth and development into adulthood. Clinical follow-up is recommended.

Future pregnancies should be monitored for chromosomal abnormalities whereby the maternal age is more of a risk over that of already having a child with a chromosomal abnormality. Amniocentesis is still the gold standard to diagnose a chromosomal abnormality during pregnancy given that NIPT is not a diagnostic test.

Thank you for your referral and we remain available.

Genetic Counseling was based on information provided at the time of counseling and/or information provided by the laboratory at the time of testing. Because genetic information and recommendations are continuously evolving, updates will be available over time. Genetic counseling is not meant to replace medical advice as offered through one's physician(s).

Yours truly,

Vivien Diaz-Barrios, MS, LCGC
Vivien Diaz-Barrios, MS, LCGC
Board Certified and Licensed Genetic Counselor
Licensed in New Jersey

session time: 30 minutes
ICD-10 Code:
Q99.9 chromosomal abnormality, not elsewhere classified

cc. Medical Provider: W. Connor, MD
cc: patient's parents
attachment: Pedigree

Patient: [redacted]
Parents: [redacted]

DOB: [redacted]
Genetic Counselor: V. Diaz-Barrios, MS, LCGC