



Patient Information:
10907, Donor
DOB: [REDACTED]
Sex: M
MR#:
Patient#: FT-PT8746817

Partner Information:
Not Tested

Physician:
Kuan, James
ATTN: Kirk, Ashley
Phoenix Sperm Bank
4915 25th Avenue NE, Ste 204W
Seattle, WA 98105
Phone: (206) 588-1484

Laboratory:
Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Lawrence M. Weiss, MD
Report Date: **Jun 13, 2024**

Accession:
FT-7030649
Test#: FT-TS14853508
Specimen Type: Blood (EDTA)
Collected: May 28, 2024

Accession:
N/A

FINAL RESULTS



Carrier for genetic conditions in **multiple** genes.
Genetic counseling is recommended.

TEST PERFORMED

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders)
(515 Gene Panel; gene sequencing with deletion and duplication analysis)

| Condition and Gene | Inheritance | 10907, Donor | Partner |
|-----------------------------------------------------------|-------------|-------------------------------------|---------|
| Oculocutaneous albinism, type IV <i>SLC45A2</i> | AR | ⊕ Carrier c.365A>G (p.Asn122Ser) | N/A |
| Hemoglobin C disease <i>HBB</i> | AR | ⊕ Carrier c.19G>A (p.Glu7Lys) | N/A |

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for carrier mutations in 2 genes. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- Testing for copy number changes in the SMN1 gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)



OCULOCUTANEOUS ALBINISM, TYPE IV

| Patient | 10907, Donor | Partner |
|-----------------|--------------------------------------------------------|---------|
| Result | ⊕ Carrier | N/A |
| Variant Details | SLC45A2 (NM_016180.5) c.365A>G (p.Asn122Ser) | N/A |

What is Oculocutaneous albinism, type IV?

Oculocutaneous Albinism, Type IV (OCA4) is part of a group of genetic conditions that affect the pigmentation (color) in the skin, hair, and eyes. Affected individuals typically have very fair skin, white or light colored hair, and vision problems. Vision problems include reduced sharpness, rapid, involuntary movements of the eyes (nystagmus), and increased sensitivity to light. Additionally, those who have OCA4 are more sensitive to the sun and at an increased risk for skin damage and skin cancer (melanoma).

What is my risk of having an affected child?

Oculocutaneous albinism, type IV is inherited in an autosomal recessive manner. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

Prognosis is generally good for appropriately managed individuals with OCA4 and lifespan is normal. There is not a cure for OCA4, but management can include vision aids and measures to protect the skin. Regular visits to the ophthalmologist or dermatologist are recommended.

What mutation was detected?

The detected heterozygous variant was NM_016180.5:c.365A>G (p.Asn122Ser). This variant, p.Asn122Ser, has previously been reported in the compound heterozygous state in individuals with albinism (PubMed: [29345414](#), Invitae, personal communication). Additionally, this variant has been identified by this laboratory in the compound heterozygous state an individual with symptoms consistent with a SLC45A2-related condition. Another variant at this position in the gene (p.Asn122Lys) has been associated with albinism, suggesting that a change at this position adversely affects protein structure and/or function and is potentially disease-causing (PubMed: [34838614](#)). The laboratory classifies this variant as likely pathogenic.



HEMOGLOBIN C DISEASE

| Patient | 10907, Donor | Partner |
|-----------------|-------------------------------------------------|---------|
| Result | ⊕ Carrier | N/A |
| Variant Details | HBB (NM_000518.5) c.19G>A (p.Glu7Lys) | N/A |

What is Hemoglobin C disease?

Hemoglobin C disease (Hb C disease) is an inherited blood disorder caused by having two copies of Hemoglobin C (HbC). Some individuals with hemoglobin C disease have a reduced amount of red blood cells and have mild hemolytic anemia, while others do not experience any symptoms. However, HbC can cause more severe symptoms if inherited with sickle cell or beta thalassemia trait.

The inheritance of one copy each of HbC and the sickle cell variant (HbS) results in Hb S-C disease, which is a milder form of sickle cell disease. Sickle cell disease is an inherited blood disease characterized by anemia, pain crisis, susceptibility to infection, and organ damage. Sickle cell disease affects a protein called hemoglobin, found in blood. Under conditions of low oxygen, the abnormal hemoglobin causes red blood cells to form a sickle shape, rather than their normal round shape. Sickled red blood cells break down prematurely, leading to anemia. These cells can also stick together and can block blood vessels, causing pain and resulting in inadequate blood supply to the parts of the body that can lead to organ damage.

The inheritance of one copy each of HbC and beta thalassemia trait result in Hemoglobin C/β Thalassemia, which is a type of beta thalassemia. Beta thalassemia is a blood disorder that reduces the production of hemoglobin. Depending on the type of beta thalassemia trait inherited with HbC, individuals can develop mild to moderate anemia, failure to thrive, and jaundice. Some individuals may develop an enlarged spleen, liver and heart, as well as bony abnormalities.

What is my risk of having an affected child?

Hemoglobin C disease is inherited in an autosomal recessive manner. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

Individuals with hemoglobin C disease can have mild anemia but are not concerned for serious health issues. Some individuals with hemoglobin C disease never experience any symptoms in their lifetime. However, the symptoms of sickle cell disease and beta thalassemia are more severe. Treatment for sickle cell disease includes hydration and pain management for pain crises, antibiotics, and medications to reduce episodes of blood vessel blockage. Blood transfusion may be required in severe cases. The most common treatment for beta thalassemia is blood transfusions. Depending on the severity of the disorder, frequent blood transfusions need to be followed by chelation therapy to remove the buildup of toxic metals in the blood that can cause organ damage.

What mutation was detected?

The detected heterozygous variant was NM_000518.5:c.19G>A (p.Glu7Lys). This variant, also known as p.Glu6Lys or hemoglobin C, is one of the most common abnormal structural hemoglobin variants globally reported in multiple individuals affected with hemoglobin C and other hemoglobinopathies (PubMed: [8294201](#), [20301551](#), [26372199](#), [23297836](#), [27117572](#), [2888754](#), [22028795](#), [21228398](#), [30604644](#), [28121068](#)). Functional studies have demonstrated that this variant results in a reduction of the overall surface hydrophobicity and affects the kinetic properties of the hemoglobin protein (PubMed: [2888754](#)). This variant is classified as "Pathogenic" in ClinVar, with multiple submitters in agreement (ClinVar: 15126). The laboratory classifies this variant as pathogenic.



GENES TESTED:

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) - 515 Genes

This analysis was run using the Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) gene list. 515 genes were tested with 99.5% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCC2, ABCC8, ACAD9, ACADM, ACADVL, ACAT1, ACOX1, ACSF3, ADA, ADAMTS2, ADAMTSL4, ADGRG1, ADGRV1, AGA, AGL, AGPS, AGXT, AHI1, AIPL1, AIRE, ALDH3A2, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, AMN, AMT, ANO10, AP1S1, AQP2, ARG1, ARL6, ARSA, ARSB, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7B, ATP8B1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BLM, BLOC1S3, BLOC1S6, BMP1, BRIP1, BSND, CAD, CANT1, CAPN3, CASQ2, CBS, CC2D1A, CC2D2A, CCDC103, CCDC39, CCDC88C, CD3D, CD3E, CD40, CD59, CDH23, CEP152, CEP290, CERKL, CFTR, CHAT, CHRNE, CHRNG, CIITA, CLCN1, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL27A1, COL4A3, COL4A4, COL7A1, COX15, CPS1, CPT1A, CPT2, CRB1, CRTAP, CRYL1, CTNS, CTSA, CTSC, CTSD, CTSK, CYBA, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, CYP7B1, DBT, DCAF17, DCLRE1C, DDX11, DGAT1, DGUOK, DHCR7, DHDDS, DLD, DLL3, DNAH11, DNAH5, DNAI1, DNAI2, DNMT3B, DOK7, DUOX2, DYNC2H1, DYSF, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELP1, EPG5, ERCC2, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, FAH, FAM161A, FANCA, FANCC, FANCD2, FANCE, FANCG, FANCI, FANCL, FBP1, FBXO7, FH, FKBP10, FKRP, FKTN, FMO3, FOXN1, FOXRED1, FRAS1, FREM2, FUCA1, G6PC, G6PC3, GAA, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GDF5, GFM1, GHR, GJB2, GJB6, GLB1, GLDC, GLE1, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GRHRP, GRIP1, GSS, GUCY2D, GUSB, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HJV, HLCS, HMGCL, HMOX1, HOGA1, HPD, HPS1, HPS3, HPS4, HPS5, HPS6, HSD17B3, HSD17B4, HSD3B2, HYAL1, HYLS1, IDUA, IGHMBP2, IKBKB, IL7R, INVS, ITGA6, ITGB3, ITGB4, IVD, JAK3, KCNJ1, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LARGE1, LCA5, LDLR, LDLRAP1, LHX3, LIFR, LIG4, LIPA, LMBRD1, LOXHD1, LPL, LRAT, LRP2, LRP3, LRP4, LRP5, LRP6, LRP7, LRP8, LRP9, LRP10, LRP11, LRP12, LRP13, LRP14, LRP15, LRP16, LRP17, LRP18, LRP19, LRP20, 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otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

ALG1: Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 6-13 of the ALG1 gene (NM_019109.4). **CEP290:** Copy number analysis for exons 8-13 and exons 39-42 may have reduced sensitivity in the CEP290 gene. Confirmation of these exons are limited to individuals with a positive personal history of CEP290-related conditions and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **CFTR:** Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either. **CRYL1:** As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP11B2:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP21A2:** Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. **DDX11:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in the DDX11 gene. **DUOX2:** The current testing method is not able to reliably detect variants in exons 6-8 of the DUOX2 gene (NM_014080.5) due to significant interference by the highly homologous gene, DUOX1. **FANCD2:** Due to pseudogene interference, copy-number-variants within exon 14-17 of the FANCD2 gene (NM_033084.4) are not evaluated and detection of single-nucleotide variants and small insertions/deletions in this region is not guaranteed. **GALT:** In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state. See GeneReviews for more information: <https://www.ncbi.nlm.nih.gov/books/NBK1518/> **GBA:** Significant pseudogene interference and/or reciprocal exchanges between the GBA gene and its pseudogene, GBAP1, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of this individual's clinical findings, biochemical profile, and family history. The current testing method cannot detect copy-neutral rearrangements between the pseudogene and the functional gene, which have been reported in very rare cases of Gaucher disease (PubMed: 21704274). **HBA1:** Significant interference



from highly homologous regions in exons 1-2 of the HBA1 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. HBA2: Significant interference from highly homologous regions in exons 1-2 of the HBA2 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. HSD17B4: Copy number analysis for exons 4-6 may have reduced sensitivity in the HSD17B4 gene. Confirmation of these exons are limited to individuals with a positive personal history of D-bifunctional protein deficiency and Perrault syndrome and/or individuals carrying a pathogenic/likely pathogenic sequence variant. LMBRD1: Copy number analysis for exons 9-12 may have reduced sensitivity in the LMBRD1 gene. Confirmation of these exons are limited to individuals with a positive personal history of combined methylmalonic aciduria and homocystinuria and/or individuals carrying a pathogenic/likely pathogenic sequence variant. MTHFR: As recommended by ACMG, the two common polymorphisms in the MTHFR gene - c.1286A>C (p.Glu429Ala, also known as c.1298A>C) and c.665C>T (p.Ala222Val, also known as c.677C>T) - are not reported in this test due to lack of sufficient clinical utility to merit testing (PubMed: [23288205](#)). NEB: This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. NPHS2: If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. OTOA: Due to pseudogene interference, our current testing method is not able to reliably detect variants in exons 20-28 (NM_144672.3) in the OTOA gene. SMN1: The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: [20301526](#)). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported. TERT: The TERT promoter region is analyzed for both sequencing and copy number variants. TYR: Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 4-5 of the TYR gene (NM_000372.5). VPS45: LoF is not a known disease mechanism. WRN: Due to the interference by highly homologous regions within the WRN gene, our current testing method has less sensitivity to detect variants in exons 10-11 of WRN (NM_000553.6).

SIGNATURE:



Geetu Mendiratta-Vij, PhD, FACMG, CGMBS on 6/13/2024
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed and its performance characteristics determined by Fulgent Therapeutics LLC CAP #8042697 CLIA #05D2043189; 4399 Santa Anita Ave., El Monte, CA, 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at [626-350-0537](tel:626-350-0537) or by email at info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes on this test please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)



Report Status FINAL

Route 2017 Ordered by:
Phoenix Sperm Bank
1492 S Mill Ave
Suite 306
Tempe, AZ 85281



James Kuan, MD

Patient Information: [REDACTED] **10907, DONOR**

Account: 18131
ID/MR#: 10907
Patient Lab ID:
665762cb92b514092df45a27

Collected: 05/28/2024 11:32 AM
Received: 05/29/2024 09:39 AM
Reported: 06/06/2024 04:51 PM

Order #: 181310000117 / NL101069791
DOB: [REDACTED] **Age:** [REDACTED]
Sex: M
Patient Phone: 602-888-7255

PL

GENETICS

Accession #:
CG240005819

Cell Type/Source:
Blood

Clinician Provided Information:
DONOR TESTING

Chromosome Analysis: Routine Blood

Analysis Details:

Metaphases/Cells Counted : 20
Metaphases/Cells Analyzed : 5
Metaphases Karyotyped : 4

PV

Results:

NORMAL MALE KARYOTYPE

PV

46,XY

Interpretation:
Normal

PV

PV

Normal karyotype at the band level 550 or above as determined by the trypsin-Giemsa method. There was no evidence for a chromosome abnormality within the limits of the band level and technology utilized in this study.

PHA-stimulated lymphocyte chromosome analysis is an accurate technique to detect many constitutional chromosome abnormalities. More extensive investigation may be required to detect mosaicism or subtle structural rearrangement. It also should be noted that this type of testing does not rule out the possibility of mendelian, mitochondrial, multifactorial or environmental etiologies.

Comments:

remote: iic

PV

Cytogenetics Director:

PV

Electronically signed by Guang Liu MD, ABMGG, FACMG, Genetics/Genomics Director
Verified 06/06/24

Tests Ordered: Chromosome Analysis: Routine Blood

Unless otherwise noted, testing performed by: Sonora Quest Laboratories, 424 S 56th St, Phoenix, AZ 85034 800.766.6721
Testing noted as PV performed by: Genetics/Genomics Div., Sonora Quest Laboratories, 424 S. 56th St, Phoenix, AZ 85034 602.685.5700

End of Report

10907, DONOR Order #: 181310000117 / NL101069791 - FINAL Report

L=Low, H=High, C=Critical Abnormal, CL=Critical Low, CH=Critical High, *=Comment

Distribution #: 715345224-38914257

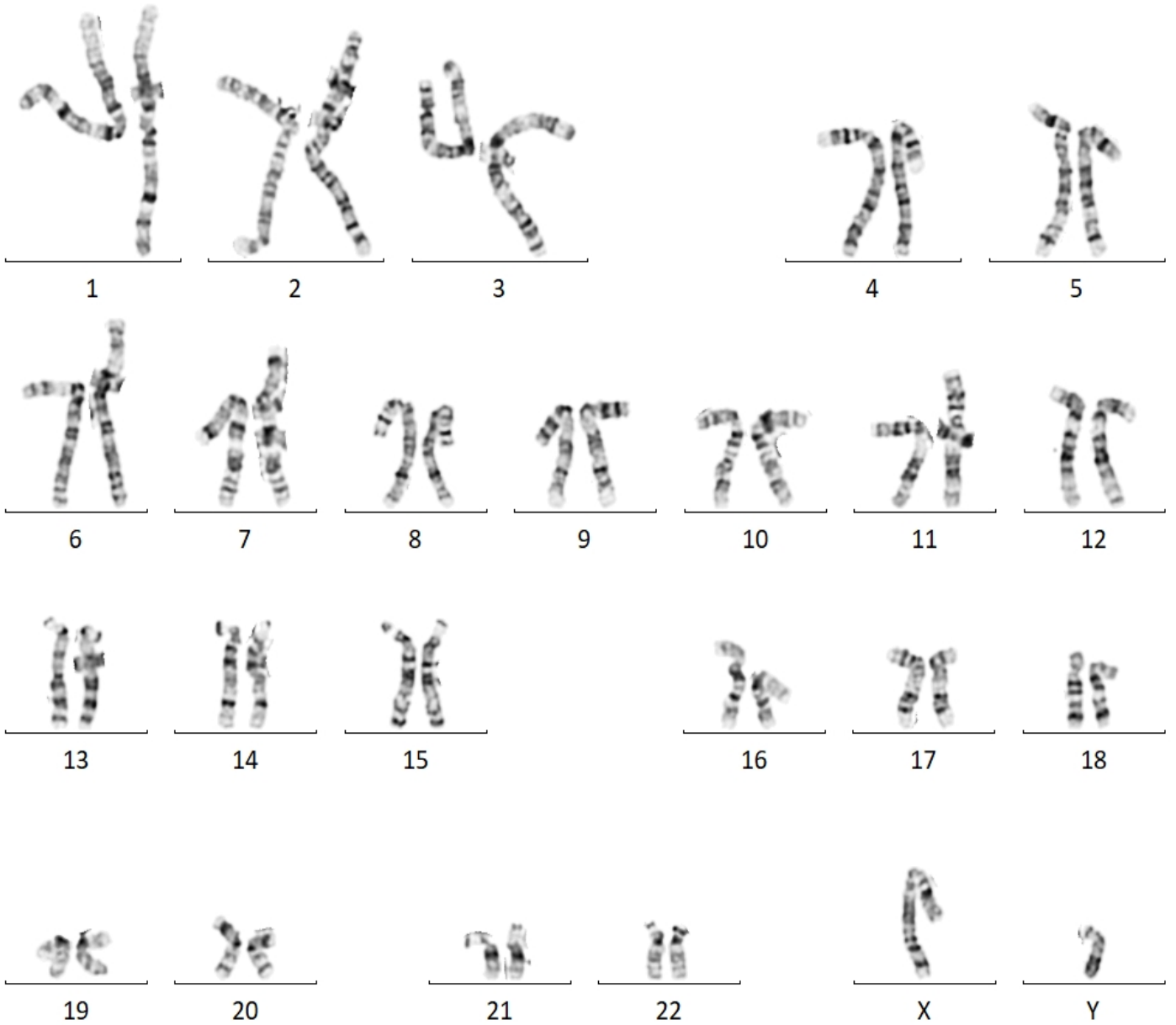


Result Report

Produced by AutoDist On 06/06/2024 04:51 PM

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ATTACHMENTS



10907, DONOR Order #: 181310000117 / NL101069791 - FINAL Report

L=Low, H=High, C=Critical Abnormal, CL=Critical Low, CH=Critical High, *=Comment

Distribution #: 715345224-38914257



Result Report

Produced by AutoDist On 06/06/2024 04:51 PM

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