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CARRIER SCREENING REPORT

Patient	Sample	Referring Doctor
Patient Name: 5579 Donor **Date of Birth:** 7[redacted]1979 **Reference #:** P0755769 **Indication:** Carrier Testing **Test Type:** Custom Carrier Screen (ECS)	**Specimen Type:** Saliva **Lab #:** 19019285CS **Date Collected:** 3/8/2019 **Date Received:** 3/9/2019 **Final Report:** 3/25/2019	**Lorraine Bonner, M.D.** **The Sperm Bank of California** **2115 Milvia Street** **Suite 201** **Berkeley, CA 94704** Fax: **510-841-0332**

RESULT SUMMARY

NEGATIVE for diseases tested ✓ CLane MS CGC 3/26/19

Please note that it is not possible to perform Tay-Sachs enzyme analysis on saliva samples, and therefore this test does not include enzyme analysis for Tay-Sachs disease.

MAR 27 2019

Spinal Muscular Atrophy

NEGATIVE for spinal muscular atrophy
SMN1 Copy Number: 2
SMN2 Copy Number: 1
c.*3+80T>G: Negative

Negative copy number result
Decreased risk of being an *SMN1* silent (2+0) carrier (see *SMA Table*)

Genes analyzed: *SMN1* (NM_000344.3) and *SMN2* (NM_017411.3)
Inheritance: Autosomal Recessive

Recommendations
Consideration of residual risk by ethnicity after a negative carrier screen is recommended, especially in the case of a positive family history for spinal muscular atrophy.

Interpretation
This patient is negative for loss of *SMN1* copy number. Complete loss of *SMN1* is causative in spinal muscular atrophy (SMA). Two copies of *SMN1* were detected in this individual, which significantly reduces the risk of being an SMA carrier. Parallel testing to assess the presence of an *SMN1* duplication allele was also performed to detect a single nucleotide polymorphism (SNP), c.*3+80T>G, in intron 7 of the *SMN1* gene. This individual was found to be negative for this change and is therefore, at a decreased risk of being a silent (2+0) carrier, see *SMA Table* for residual risk estimates based on ethnicity.

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Patient:	5579 Donor	DOB:	7[redacted]1979	Lab #:	19019285CS

Custom Carrier Screen (ECS)

Negative: No clinically significant variant(s) detected

Gene(s) analyzed: *CFTR, GBA, HEXA, BLM, ASPA, IKBKAP, FANCC, SMPD1,* and *MCOLN1*

Recommendations:
Consideration of residual risk by ethnicity after a negative carrier screen is recommended, especially in the case of a positive family history for a specific disorder.

Interpretation:
Screening for the presence of pathogenic variants in the *CFTR, GBA, HEXA, BLM, ASPA, IKBKAP, FANCC, SMPD1,* and *MCOLN1* genes which are associated with cystic fibrosis, Gaucher disease, Tay-Sachs disease, Bloom syndrome, Canavan disease, familial dysautonomia, Fanconi anemia, group C, Niemann-Pick disease, types A/B and mucolipidosis, type IV, respectively, was performed by next generation sequencing and possibly genotyping for select variants on DNA extracted from this patient's sample. No clinically significant variants were detected during this analysis.

Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for the disorder(s) tested. Please see table of residual risks for specific detection rates and residual risk by ethnicity. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

Comments:
This carrier screening test masks likely benign variants and variants of uncertain significance (VUS) if there are any. Only known pathogenic variants or likely pathogenic variants which are expected to result in deleterious effects on protein function are reported. If reporting of likely benign variants and VUS is desired in this patient, please contact the laboratory (tel. 212-241-2537) to request an amended report.

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

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Table of Residual Risks by Ethnicity

Please note: This table displays residual risks after a negative result for each of the genes and corresponding disorders. If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	Analytical Detection Rate
Bloom Syndrome (AR) NM_000057.2	*BLM*	African	1 in 532	99%	1 in 53,100	99%
		Ashkenazi Jewish	1 in 117	99%	1 in 11,700	
		East Asian	1 in 337	99%	1 in 33,600	
		Finnish	1 in 712	99%	1 in 71,100	
		Caucasian	1 in 358	95%	1 in 7,400	
		Latino	1 in 495	99%	1 in 49,400	
		South Asian	1 in 636	95%	1 in 12,500	
		Worldwide	1 in 357	97%	1 in 11,800	
Canavan Disease (AR) NM_000049.2	*ASPA*	African	1 in 741	98%	1 in 37,000	98%
		Ashkenazi Jewish	1 in 50	98%	1 in 2,400	
		Finnish	1 in 241	98%	1 in 12,000	
		Caucasian	1 in 486	88%	1 in 4,000	
		Latino	1 in 899	87%	1 in 7,100	
		South Asian	1 in 1923	61%	1 in 5,000	
		Worldwide	1 in 393	92%	1 in 5,200	
Cystic Fibrosis (AR) NM_000492.3 Exception: Exon 10	*CFTR*	African	1 in 58	91%	1 in 630	99%
		Ashkenazi Jewish	1 in 24	98%	1 in 1,200	
		East Asian	1 in 277	80%	1 in 1,400	
		Finnish	1 in 75	93%	1 in 1,100	
		Caucasian	1 in 23	95%	1 in 440	
		Latino	1 in 40	96%	1 in 1,000	
		South Asian	1 in 73	91%	1 in 800	
		Worldwide	1 in 33	94%	1 in 500	
Familial Dysautonomia (AR) NM_003640.3	*IKBKAP*	African	1 in 409	99%	1 in 40,800	99%
		Ashkenazi Jewish	1 in 35	99%	1 in 3,400	
		East Asian	1 in 784	99%	1 in 78,300	
		Finnish	1 in 707	99%	1 in 70,600	
		Caucasian	1 in 506	99%	1 in 50,500	
		Latino	1 in 801	99%	1 in 80,000	
		South Asian	1 in 855	99%	1 in 85,400	
		Worldwide	1 in 345	99%	1 in 34,400	
Fanconi Anemia, Group C (AR) NM_000136.2	*FANCC*	African	1 in 486	87%	1 in 3,700	99%
		Ashkenazi Jewish	1 in 82	99%	1 in 8,100	
		East Asian	1 in 344	99%	1 in 34,300	
		Finnish	1 in 1188	99%	1 in 119,000	
		Caucasian	1 in 431	98%	1 in 11,600	
		Latino	1 in 1121	99%	1 in 112,000	
		South Asian	1 in 1025	99%	1 in 102,000	
		Worldwide	1 in 444	97%	1 in 13,700	
Gaucher Disease (AR) NM_000157.3	*GBA*	Caucasian	1 in 164	87%	1 in 1,300	95%
		Ashkenazi Jewish	1 in 15	95%	1 in 280	
		Worldwide	1 in 158	86%	1 in 1,100	
Mucolipidosis IV (AR) NM_020533.2	*MCOLN1*	African	1 in 2037	99%	1 in 204,000	99%
		Ashkenazi Jewish	1 in 92	99%	1 in 9,100	
		Caucasian	1 in 1166	88%	1 in 9,400	
		Latino	1 in 1537	63%	1 in 4,100	
		South Asian	1 in 2565	83%	1 in 14,700	
		Worldwide	1 in 926	86%	1 in 6,500	

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Niemann-Pick Disease, Type A/B (AR) NM_000543.4	*SMPD1*	African	1 in 120	90%	1 in 1,100	99%
		Ashkenazi Jewish	1 in 98	99%	1 in 9,700	
		East Asian	1 in 81	94%	1 in 1,300	
		Finnish	1 in 2230	99%	1 in 223,000	
		Caucasian	1 in 350	81%	1 in 1,800	
		Latino	1 in 499	87%	1 in 4,000	
		South Asian	1 in 327	76%	1 in 1,300	
		Worldwide	1 in 240	88%	1 in 1,900	

Spinal Muscular Atrophy (AR) NM_000344.3 / NM_017411.3 — *SMN1/SMN2*

	Carrier Frequency	Detection Rate	Residual Risk After Negative Result (2 Copies)*	Detection Rate with *SMN1* c.3*+80T>G (2 Copies)	Residual Risk c.3*+80T>G Negative (2 Copies)	Residual Risk c.3*+80T>G Positive (2 Copies)	Residual Risk with ≥ 3 copies of *SMN1*
African American	1 in 85	71%	1 in 150	91%	1 in 455	1 in 49	1 in 4,300
Ashkenazi Jewish	1 in 76	90%	1 in 872	93%	1 in 978	1 in 10	1 in 4,800
East Asian	1 in 53	94%	1 in 864	95%	1 in 901	1 in 12	1 in 4,900
Caucasian	1 in 48	96%	1 in 803	95%	1 in 894	1 in 23	1 in 4,900
Latino	1 in 63	91%	1 in 609	94%	1 in 930	1 in 47	1 in 4,800
South Asian	1 in 103	87%	1 in 637	87%	1 in 637	1 in 608	1 in 4,700
Sephardic Jewish	1 in 34	96%	1 in 696	97%	1 in 884	1 in 12	1 in 4,900

*Residual risk with two copies *SMN1* detected using dosage sensitive methods. The presence of three or more copies of *SMN1* reduces the risk of being an *SMN1* carrier between 5 - 10 fold, depending on ethnicity.

Tay-Sachs Disease (AR) NM_000520.4	*HEXA*	African	1 in 216	99%*	1 in 21,500	99%
		Ashkenazi Jewish	1 in 30	99%*	1 in 2,900	
		East Asian	1 in 210	99%*	1 in 20,900	
		Finnish	1 in 399	99%*	1 in 39,800	
		Caucasian	1 in 90	97%*	1 in 3,400	
		Latino	1 in 243	89%*	1 in 2,200	
		South Asian	1 in 416	70%*	1 in 1,400	
		Worldwide	1 in 121	96%*	1 in 3,200	
		French Canadian - Gaspesie	1 in 13	99%*	1 in 1,200	
		French Canadian - Other	1 in 73	99%*	1 in 7,200	
		Irish	1 in 41	90%*	1 in 400	
		Sephardic Jewish – Moroccan and Iraqi	1 in 125	99%*	1 in 12,400	

*Carrier detection by HEXA enzyme analysis has a detection rate of approximately 98%.
AR: Autosomal Recessive

This case has been reviewed and electronically signed by Ruth Kornreich, Ph.D., FACMG, Laboratory Director

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.

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Patient:	5579 Donor	DOB:	7/●1979	Lab #:	19019285CS

Test Methods and Comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Fragile X CGG Repeat Analysis (Analytical Detection Rate >99%)
PCR amplification using Asuragen, Inc. AmplideX® *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* CGG repeats in the premutation and full mutation size range were further analyzed by Southern blot analysis to assess the size and methylation status of the *FMR1* CGG repeat.

Genotyping (Analytical Detection Rate >99%)
Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY® System were used to identify variants that are complex in nature or are present in low copy repeats. Rare sequence variants may interfere with assay performance.

Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)
MLPA® probe sets and reagents from MRC-Holland were used for copy number analysis of specific targets versus known control samples. False positive or negative results may occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

For alpha thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. This test is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome, may not be detected. With the exception of triplications, other benign alpha-globin gene polymorphisms will not be reported. Analyses of *HBA1* and *HBA2* are performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For Duchenne muscular dystrophy, the copy numbers of all *DMD* exons were analyzed. Potentially pathogenic single exon deletions and duplications are confirmed by a second method. Analysis of *DMD* is performed in association with sequencing of the coding regions.

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. These 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between *CYP21A2* and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals with SMA have an *SMN1* mutation that occurred *de novo*. Typically in these cases, only one parent is an SMA carrier.

The presence of the c.*3+80T>G (chr5:70,247,901T>G) variant allele in an individual with Ashkenazi Jewish or Asian ancestry is typically indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, c.*3+80T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic or Caucasian ancestry, the presence or absence of c.*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

Pathogenic or likely pathogenic sequence variants in exon 7 may be detected during testing for the c.*3+80T>G variant allele; these will be reported if confirmed to be located in SMN1 using locus-specific Sanger primers

MLPA for Gaucher disease (*GBA*), cystic fibrosis (*CFTR*), and non-syndromic hearing loss (*GJB2/GJB6*) will only be performed if indicated for confirmation of detected CNVs. If *GBA* analysis was performed, the copy numbers of exons 1, 3, 4, and 6 - 10 of the *GBA* gene (of 11 exons total) were analyzed. If *CFTR* analysis was performed, the copy numbers of all 27 *CFTR* exons were analyzed. If *GJB2/GJB6* analysis was performed, the copy number of the two *GJB2* exons were analyzed, as well as the presence or absence of the two upstream deletions of the *GJB2* regulatory region, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854).

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Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)
NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect™QXT technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a number of UTR, intronic or promoter regions that contain previously reported mutations. Samples were pooled and sequenced on the Illumina HiSeq 2500 platform in the Rapid Run mode or the Illumina NovaSeq platform in the Xp workflow, using 100 bp paired-end reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. The exons contained within these regions are noted within Table 1 (as "Exceptions") and will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas, or regions that fall into the Exceptions mentioned above. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition, a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants (Richards et al, 2015). All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis will not be reported.

Copy Number Variant Analysis (Analytical Detection Rate >95%)
Large duplications and deletions were called from the relative read depths on an exon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either a custom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenic single-exon CNVs will be confirmed and reported, if detected.

Exon Array (Confirmation method) (Accuracy >99%)
The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each array matrix has approximately 180,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGH probes are enriched to target the exonic regions of the genes in this panel.

Quantitative PCR (Confirmation method) (Accuracy >99%)
The relative quantification PCR is utilized on a Roche Universal Library Probe (UPL) system, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probe sets that specific to the target region and a control region with known genomic copy number. Relative genomic copy numbers are calculated based on the standard ΔΔCt formula.

Long-Range PCR (Analytical Detection Rate >99%)
Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2*, *HBA1* and *HBA2* and *GBA*. The PCR products were then prepared for short-read NGS sequencing and sequenced. Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with the sequencing output to analyze the results. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where two copies of a gene are located on the same chromosome in tandem, only the second copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However, because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in the tandem allele and this patient is therefore less likely to be a carrier. When an

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individual carries both a duplication allele and a pathogenic variant, or multiple pathogenic variants, the current analysis may not be able to determine the phase (cis/trans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing is required to determine the carrier status.

Residual Risk Calculations
Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >28,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the *a priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Sanger Sequencing (Confirmation method) (Accuracy >99%)
Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Tay-Sachs Disease (TSD) Enzyme Analysis (Analytical Detection Rate ≥98%)
Hexosaminidase activity and Hex A% activity were measured by a standard heat-inactivation, fluorometric method using artificial 4-MU-β-N-acetyl glucosaminide (4-MUG) substrate. This assay is highly sensitive and accurate in detecting Tay-Sachs carriers and individuals affected with TSD. Normal ranges of Hex A% activity are 55.0-72.0 for white blood cells and 58.0-72.0 for plasma. It is estimated that less than 0.5% of Tay-Sachs carriers have non-carrier levels of percent Hex A activity, and therefore may not be identified by this assay. In addition, this assay may detect individuals that are carriers of or are affected with Sandhoff disease. False positive results may occur if benign variants, such as pseudodeficiency alleles, interfere with the enzymatic assay. False negative results may occur if both *HEXA* and *HEXB* pathogenic or pseudodeficiency variants are present in the same individual.

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

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Additional disease-specific references available upon request.

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1428 Madison Ave, Atran Bldg
Rm 2-25 New York, NY 10029

CLIA #: 33D2097541
T: 800-298-6470
F: 212-241-0139
www.sema4genomics.com