Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4940

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 09/28/22

Donor Reported Ancestry: English, Irish, Scottish, Swedish, German Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by genotyping for 99 variants in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/641
Hb Beta Chian-Related Hemoglobinopathy	Negative by genotyping for 28 variants in the HBB gene	1/290
Tay Sachs Disease Enzyme Analysis	Non-Carrier by Hexosaminidase A analysis	
Special Testing		
Genes: ACADM, PAH, LOXHD1	Negative by genotyping	See attached results for more information

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.**Donor residual risk is the chance the donor is still a carrier after testing negative.

Counsyl

RESULT RECIPIENT	MALE	FEMALE
FAIRF[redacted]YOBANK - [redacted]	4940 4940	N/A
Report Date: 09/05/2014	DOB: [redacted]	
	Ethnicity: Northern European	
	Sample Type: EDTA Blood	
	Date of Collection: 08/27/2014	
	Date Received: 08/29/2014	
	Date Tested: 09/04/2014	
	Barcode: [redacted]	
	Indication: Egg or Sperm Donor	

Family Prep Screen

NEGATIVE

ABOUT THIS TEST

The Counsyl Family Prep Screen (version 1.0) tests known mutations to help you learn about your chance to have a child with a genetic disease.

PANEL DETAILS

Fairfax Cryobank Fundamental Panel **(3 diseases tested)**

VERSION

4940 4940 **(Family Prep Screen 1.0)**

RESULTS SUMMARY

NEGATIVE

No known or potential disease-causing mutations were detected.

ENTERED
ON 9.9.14

CLINICAL NOTES

- None

NEXT STEPS

- If necessary, patients can discuss residual risks with their physician or a genetic counselor.
- To schedule a complimentary appointment to speak with a clinical expert about these results, please visit counsyl.com/my/consults/.

Methods and Limitations

4940 4940 [Family Prep Screen 1.0]: targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Copy number analysis: Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's workup. CLIA Number: **#05D1102604**.

LAB DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Jelena Brezo

Jelena Brezo, PhD, FACMG

Diseases Tested

Autosomal Recessive Disorders

TARGETED GENOTYPING

Cystic Fibrosis - Gene: CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542*, G551D, R553*, R560T, R1162*, W1282*, N1303K, c.1521_1523delCTT, c.1519_1521delATC, c.2052delA, c.3528delC, c.489+1G>T, c.579+1G>T, c.1585-1G>A, c.1766+1G>A, 2789+5G>A, c.2988+1G>A, 3849+10kbC>T, E60*, R75*, E92*, Y122*, G178R, R347H, Q493*, V520F, S549N, P574H, M1101K, D1152H, c.2012delT, c.262_263delTT, c.313delA, c.948delT, c.3744delA, c.3773dupT, c.1680-1G>A, 3272-26A>G, c.2051_2052delAAinsG, S549R, R117C, L206W, G330*, T338I, R352Q, S364P, G480C, C524*, S549R, Q552*, A559T, G622D, R709*, K710*, R764*, Q890*, R1066C, W1089*, Y1092X, R1158*, S1196*, W1204*, Q1238*, S1251N, S1255*, c.3067_3072del6, c.442delA, c.531delT, c.803delA, c.805_806delAT, c.1545_1546delTA, M607_Q643del, c.1911delG, c.1923_1931del9ins1, c.1976delA, c.3039delC, c.3536_3539delCCAA, c.3659delC, c.1155_1156dupTA, c.2052dupA, c.2175dupA, c.2738insG, 296+12T>C, c.273+1G>A, 405+3A>C, c.274-1G>A, 711+5G>A, c.580-1G>T, c.1766+1G>T, 1898+5G>T, Q996, c.325_327delTATinsG, 3849+4A>G, c.1075_1079del5ins5. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection rate:** Northern European 91%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. **Variants (28):** E7V, K18*, Q40*, c.126_129delCTTT, c.27dupG, IVS-II-654, IVS-II-745, c.315+1G>A, IVS-I-6, IVS-I-110, IVS-I-5, c.92+1G>A, -88C>T, -28A>G, -29A>G, c.25_26delAA, c.217dupA, c.316-2A>C, c.316-2A>G, G25, -87C>G, E7K, W16*, c.51delC, c.20delA, E27K, E122Q, E122K. **Detection rate:** Northern European 83%.

COPY NUMBER ANALYSIS

Spinal Muscular Atrophy - Gene: SMN1. **Variant (1):** SMN1 copy number. **Detection rate:** Northern European 95%.

Risk Calculations

Below are the risk calculations for all diseases tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	4940 4940 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000

Patient Information	Specimen Information	Client Information
ID, 4940 **DOB: Not Given** **AGE: Not Given** Gender: M Fasting: N Phone: NG Patient ID: NG	Specimen: NE251307B Requisition: 8105802 Collected: 08/27/2014 / 11:30 EDT Received: 08/27/2014 / 21:29 EDT Reported: 08/28/2014 / 13:50 EDT	Client #[redacted] STERN, HARVEY J FAIRFAX CRYO BANK [redacted]

COMMENTS: ADULT

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.21		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	15.9		13.2-17.1 g/dL	
HEMATOCRIT	46.6		38.5-50.0 %	
MCV	89.4		80.0-100.0 fL	
MCH	30.6		27.0-33.0 pg	
RDW	13.3		11.0-15.0 %	
HEMOGLOBIN A	96.8		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.2		1.8-3.5 %	
INTERPRETATION				
	Normal phenotype.			

PERFORMING SITE:
QHO QUEST DIAGNOSTICS HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044-3432 Laboratory Director: ANDREW S EDELMAN, MD PHD, CLIA: 39D0204404

ENTERED QH 9.9.14

Integrated GENETICS

Tay-Sachs Enzyme Analysis

Patient Name: 4940, 4940-140827
Referring Physician: [redacted]
Specimen #: [redacted] **Client #:** [redacted]
Patient ID: [redacted]

Fairfax Cryobank [redacted]

DOB: Not Given
SSN: ***-**-****

Date Collected: 08/27/2014
Date Received: 08/28/2014
Lab ID:
Hospital ID:
Specimen Type: **White Blood Cells**

RESULTS: **Hexosaminidase Activity : 1362 nmol/mg protein**
Hexosaminidase Percent A: 56.4

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥54%	≥54%
Expected Carrier Range:	Hex A	20 - 49%	20 - 49%

INTERPRETATION: NON CARRIER

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

ENTERED ON 9.9.14

Under the direction of: Stanford Marenberg, PHD, MBCC

Stanford Marenberg, Ph.D.

Date: 09/02/2014

Testing Performed At Esoterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank [redacted]

Address [redacted]

Reporting Phone # [redacted]

Patient name/Donor Alias	Donor # 4940	Patient DOB	N/A
Donor #	4940-140827	Specimen type	Peripheral Blood
Collection Date	08/27/2014	Accession #	14-148CG
Date Received	08/28/2014		

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	Type of banding	GTG
Cells analyzed	5	Band resolution	550
Cells karyotyped	2		
Modal chromosome #	46		

KARYOTYPE 46,XY

FISH

Probe(s)	N/A
Nuclei scored	N/A

INTERPRETATION

Normal male karyotype
No clonal numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments

ENTERED OH 9.9.14

Wayne S. Stanley, Ph.D., FACMG
Clinical Cytogeneticist

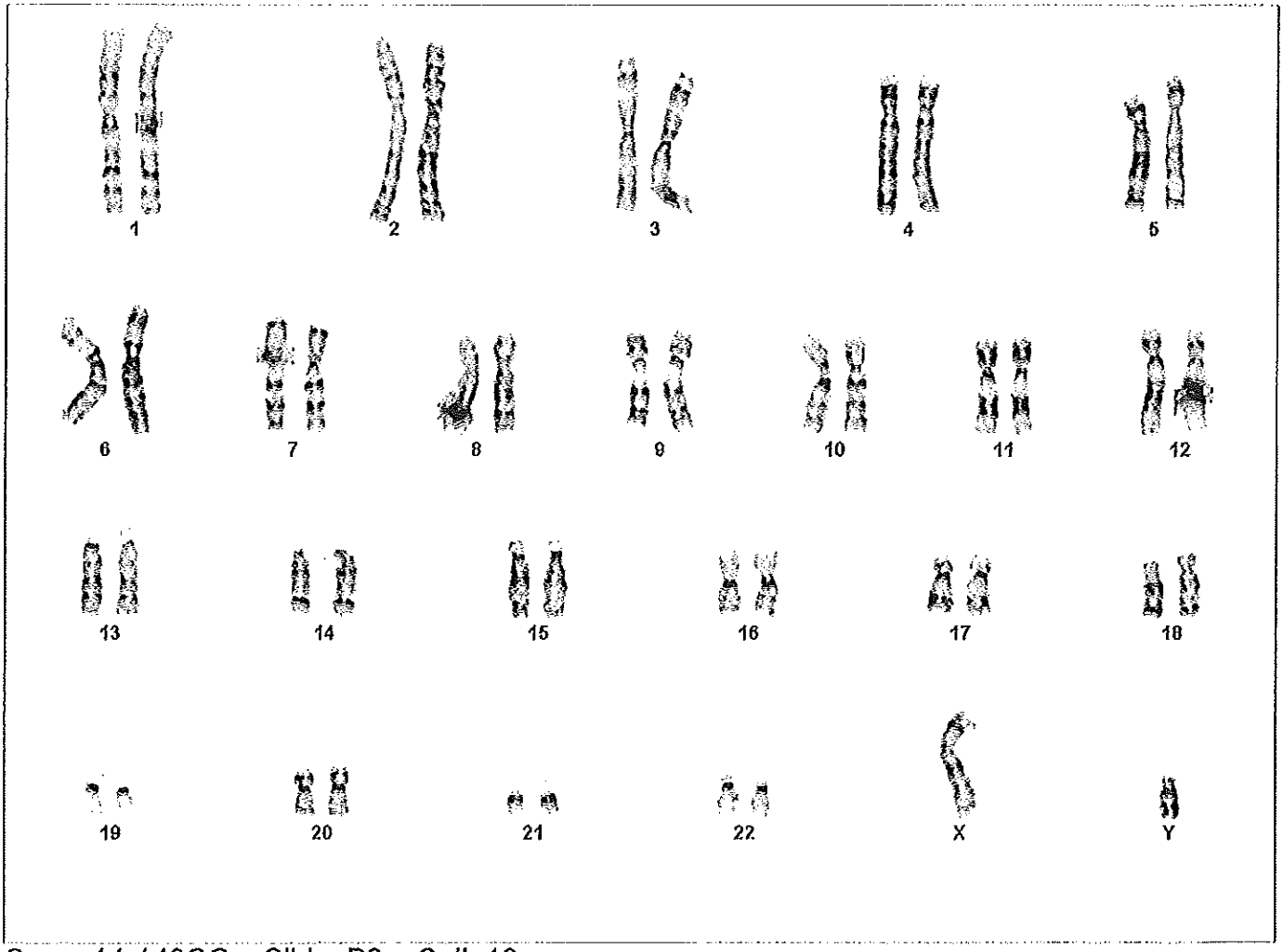
9/8/14
Date

Genetics and IVF Preimplantation Genetics Laboratory

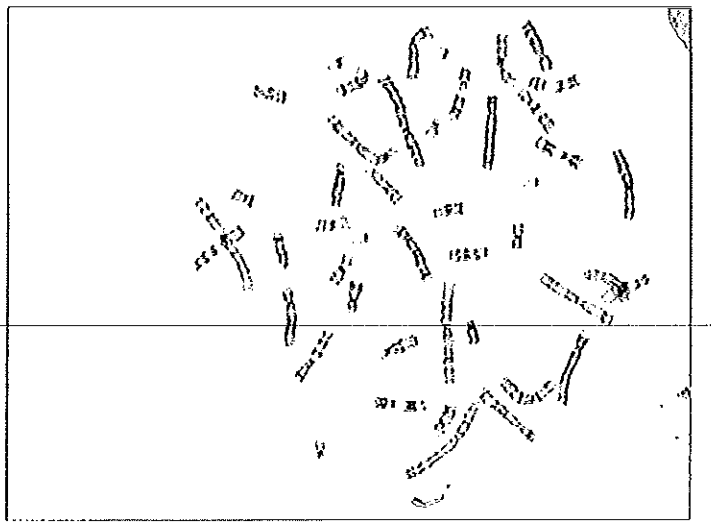
Patient name: DONOR # 4940

Case name: 14-148CG

46,XY



Case: 14-148CG Slide: B2 Cell: 18



Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
[redacted]
Report Generated: 06/03/2015

4940 Donor

DOB:
Gender: Male
Ethnicity: European
Procedure ID: 22585
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood , #24055
Specimen Collection: 05/27/2015
Specimen Received: 05/28/2015
Specimen Analyzed: 06/03/2015

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

4940 Donor was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♂ Male

Panel: Phenylalanine Hydroxylase Deficiency and Medium Chain Acyl-CoA Dehydrogenase Deficiency , Diseases Tested: 2, Mutations Tested: 25, Genes Tested: 2, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
Lab Technician Bo Chu

Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors.

The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

Diseases & Mutations Assayed

High Impact | Treatment Benefits | X-Linked | Moderate Impact

H	T	X	M	Disease	#	Mutations
●	●	○	○	Medium Chain Acyl-CoA Dehydrogenase Deficiency	8	♂ Genotyping | c.A985G (p.K329E), c.C362T (p.T121I), c.G583A (p.G195R), c.G799A (p.G267R), c.T199C (p.Y67H), c.C250T (p.L84F), c.C616T (p.R206C), c.G617A (p.C206H)
●	●	○	○	Phenylalanine Hydroxylase Deficiency	17	♂ Genotyping | c.1066-11G>A, c.1315+1G>A, c.A1241G (p.Y414C), c.C1222T (p.R408W), c.C754T (p.R252W), c.G1223A (p.R408Q), c.G473A (p.R158Q), c.G782A (p.R261Q), c.G814T (p.G272X), c.T143C (p.L48S), c.T194C (p.I65T), c.T896G (p.F299C), c.C842T (p.P281L), c.G838A (p.E280K), c.C117G (p.F39L), c.G3A (p.M1I), c.A1G (p.M1V)

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
3015 Williams Drive, #110, Fairfax, VA, 22031, US
Physician: [redacted]
Report Generated: 2017-03-31

4940 Donor

DOB:
Gender: Male
Ethnicity: European
Procedure ID: 22585
Kit Barcode: [redacted]
Specimen: Blood, #24055
Specimen Collection: 2015-05-27
Specimen Received: 2015-05-28
Specimen Analyzed: 2017-03-31

Partner Not Tested

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Mutations Tested: 2

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

4940 Donor was not identified to carry any of the mutation(s) tested.

No pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

For additional disease information, please visit recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

Assay performed by Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763
Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in the genes tested. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors. The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

This test was developed and its performance determined by Recombine, Inc., and it 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.

Diseases & Mutations Assayed

Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related (LOXHD1): Mutations (2): ♂ Genotyping | c.2008C>T (p.R670X), c.4714C>T (p.R1572X)

Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

Disease	Carrier Rate	Detection Rate	Residual Risk
Nonsyndromic Hearing Loss and Deafness: LOXHD1 Related	♂ Ashkenazi Jewish: 1/180	>99%	<1/18,000