



RESULTS RECIPIENT
SEATTLE SPERM BANK
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Report Date: 03/16/2022

MALE
DONOR 10632
DOB: [REDACTED]
Ethnicity: Southeast Asian
Sample Type: EDTA Blood
Date of Collection:
Date Received: 02/25/2022
Date Tested: 03/03/2022
Barcode: 11004512970442
Accession ID: CSLEJ4U9LZ2NAQ6
Indication: Egg or sperm donor

FEMALE
N/A

Foresight® Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

RESULTS SUMMARY

Risk Details	DONOR 10632	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested)	N/A
POSITIVE: CARRIER GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness Reproductive Risk: 1 in 140 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_004004.5(GJB2):c.109G>A (V37I) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER Citrullinemia Type 1 Reproductive Risk: 1 in 480 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000050.4(ASS1):c.910C>T (R304W) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".
POSITIVE: CARRIER Calpainopathy Reproductive Risk: 1 in 560 Inheritance: Autosomal Recessive	⊕ CARRIER* CAPN3 Exon 20-Partial Exon 24 del [NM_000070.2:c.2115+111_*6del] †	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

†Likely to have a negative impact on gene function.

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 12.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may



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also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness

Reproductive risk: 1 in 140
Risk before testing: 1 in 4,600

Gene: GJB2 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 10632	No partner tested
Result	Carrier	N/A
Variant(s)	NM_004004.5(GJB2):c.109G>A(V37I) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of GJB2-related DFNB1 nonsyndromic hearing loss and deafness. Carriers generally do not experience symptoms. V37I is typically associated with bilateral mild to moderate and slowly progressive hearing loss.	N/A
Detection rate	>99%	N/A
Exons tested	NM_004004:1-2.	N/A

What Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

DFNB1 nonsyndromic hearing loss and deafness is an inherited condition in which an individual has mild to severe hearing loss, usually, from birth. It is caused by mutations in *GJB2* (which encodes the protein connexin 26) and *GJB6* (which encodes connexin 30). The condition does not typically worsen over time, but in some cases may be slowly progressive. The word "nonsyndromic" refers to the fact that there are no other symptoms or systems of the body involved with the disease. Unlike some other forms of hearing loss, DFNB1 nonsyndromic hearing loss and deafness does not affect balance or movement. The degree of hearing loss is difficult to predict based on which genetic mutation one has. Even if members of the same family are affected by DFNB1 nonsyndromic hearing loss and deafness, the degree of hearing loss may vary among them.

How Common Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

In the United States, the United Kingdom, France, Australia, and New Zealand, approximately 14 in 100,000 individuals have DFNB1 nonsyndromic hearing loss and deafness. This may be an underestimate as individuals with a mild presentation may not be diagnosed. Roughly 1 in 33 Caucasian individuals are carriers of the mutation that causes the condition.

While this condition is most recognized in the Caucasian population, it has also been observed in other ethnicities.

How Is GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness Treated?

Individuals with DFNB1 nonsyndromic hearing loss and deafness may show improvement by using hearing aids. For those with profound deafness, cochlear implants may also be helpful. They may also want to consider enrolling in an educational program for the hearing impaired.

What is the Prognosis for an Individual with GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness?

While an individual with GJB2-related DFNB1 nonsyndromic hearing loss and deafness will have mild to severe hearing loss, it does not affect lifespan and does not affect any other part of the body.

POSITIVE: CARRIER

Citrullinemia Type 1

Reproductive risk: 1 in 480
Risk before testing: 1 in 58,000

Gene: ASS1 | Inheritance Pattern: Autosomal Recessive

Patient	DONOR 10632	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000050.4(ASS1):c.910C>T(R304W) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of citrullinemia type 1. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000050:3-16.	N/A

What Is Citrullinemia Type 1?

Citrullinemia type I is an inherited condition in which ammonia and other toxic substances build up in the blood, causing life-threatening complications shortly after birth. It is caused by mutations in the *ASS1* gene. Citrullinemia type I belongs to a group of diseases known as urea cycle disorders. When the body consumes protein, it also produces excess nitrogen. Under normal circumstances, the body converts that nitrogen to urea, which is then excreted in the urine. People with citrullinemia type I are deficient in an enzyme known as argininosuccinate synthase, which is needed for this vital process, leading to a buildup of ammonia and other urea cycle byproducts in the body. The excess ammonia is harmful to the nervous system, causing many of the disease's symptoms.

Infants with citrullinemia type I appear normal at birth. However, within the first week of life most will become lethargic and display poor feeding, vomiting, and seizures that often lead to unconsciousness, stroke, increased pressure around the brain, and death if untreated.

A milder form of type I citrullinemia may develop later in childhood or adulthood, but this form of the disease is less common. This later-onset form is associated with intense headaches, partial loss of vision, problems with balance and muscle coordination (ataxia), episodes of elevated ammonia that are similar to the classic form, and lethargy. Women with the later-onset form may have an onset of severe symptoms during pregnancy or postpartum.

How Common Is Citrullinemia Type 1?

Scientists estimate that 1 in 57,000 births are affected by citrullinemia type I.

How Is Citrullinemia Type 1 Treated?

The goals of treatment for citrullinemia type I are to regulate the amount of ammonia in the blood. Physicians adhere to certain protocols to control the body's ammonia levels. These protocols use medication, dialysis, and a specifically prescribed diet. Children with citrullinemia will need to be monitored closely by a physician specializing in metabolic disorders. Physicians will also monitor and attempt to relieve any excess of pressure around the brain.

Lifelong management involves strict dietary management in addition to oral administration of sodium phenylbutyrate or glycerol phenylbutyrate and L-carnitine to prevent systemic hypocarnitinemia. Liver transplantation may be warranted.

What Is the Prognosis for a Person with Citrullinemia Type 1?

The prognosis for a child with citrullinemia type I is not well established. Without treatment, the longest-known survival was 17 days. With treatment, these children can survive for an unknown period of time, but they will have significant mental and neurological impairment.

Initial neurologic findings associated with the milder, late-onset form may be more subtle than those seen in the acute neonatal form because of the older age of affected individuals.

POSITIVE: CARRIER

Calpainopathy

Reproductive risk: 1 in 560
Risk before testing: 1 in 78,000

Gene: CAPN3 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 10632	No partner tested
Result	👤 Carrier	N/A
Variant(s)	CAPN3 Exon 20-Partial Exon 24 del [NM_000070.2:c.2115+111_*6del] †	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of calpainopathy. Carriers generally do not experience symptoms.	N/A
Detection rate	99%	N/A
Exons tested	NM_000070:1-24.	N/A

†Likely to have a negative impact on gene function.

What is Calpainopathy?

Calpainopathy (previously known as limb-girdle muscular dystrophy type 2A, or LGMD2A) is a spectrum of disorders that cause muscle breakdown (atrophy) and weakness. Calpainopathy is caused by harmful genetic changes (mutations) in the *CAPN3* gene. The primary symptom is worsening (progressive) muscle weakness of the hip, shoulder, and abdomen. The rate at which the muscles weaken can vary greatly, but many experience weakness to a point where a wheelchair becomes necessary. Other features include enlarged calf muscles, shortening and hardening of muscles leading to rigid joints (contractures), curvature of the spine (scoliosis), and prominence (winging) of the shoulder blades. Calpainopathy does not affect intelligence or mental function. Some individuals with the disease can have a mild course where they do not show symptoms (asymptomatic), while others may have severe symptoms that can be fatal. Failure to get enough oxygen to the lungs (respiratory failure) is the most common cause of death. The symptoms of the disease can vary greatly from person to person (even among people in the same family). The age that symptoms begin is also quite varied, with some individuals showing muscle weakness beginning in childhood. The most common age of onset is in the early teens. Genetic testing cannot predict how severe individuals will be affected.

How common is Calpainopathy?

The exact prevalence of calpainopathy is difficult to determine because of the wide range of symptoms. The estimated prevalence of calpainopathy in the population is approximately 1 in 80,000 individuals. Calpainopathy is more common among individuals of French Reunion Island and Indiana Amish descent.

How is Calpainopathy treated?

There is no cure for calpainopathy. Physical therapy helps patients to retain muscle strength and mobility for as long as possible. Mobility aids, such as walkers, canes, braces, and wheelchairs, may become necessary. If muscle weakness begins to affect the ability to breathe, a machine that assists with breathing (a ventilator) may be needed. Cardiac surveillance is recommended, and those who develop heart problems will need to see a heart specialist (a cardiologist) for treatment. Some individuals may need surgery if they develop scoliosis or contractures.

What is the prognosis for an individual with Calpainopathy?

The outlook for a person with calpainopathy varies. Generally, the earlier symptoms begin, the faster they progress. Some people with the disease experience only mild symptoms and may have near-normal strength. Others with a mild course may remain able to walk for 30 years or more after symptoms appear. People with more severe disease typically may need to use a wheelchair as early as 10 years after their diagnosis. There is evidence that symptoms progress faster in males than in females.

Methods and Limitations

DONOR 10632 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

Sequencing with copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of ≥75%. Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants may interfere with this analysis. Some individuals with two copies of *SMN1* are “silent” carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two copies of *SMN1*.

Analysis of homologous regions

A combination of high-throughput sequencing, read-depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss-of-function variants in certain genes that have homology to other genomic regions. The precise breakpoints of large deletions in these genes cannot be determined but are instead estimated from copy number analysis. Pseudogenes may interfere with this analysis, especially when many pseudogene copies are present.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient CAH, depending on the chromosomal location of the variants (phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be “silent” carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are based only on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided.

Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e. pathogenic or likely pathogenic) are reported. Benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in more patients. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If an updated report is issued, the variants reported may change based on their current classification. This can include changes to the variants displayed in gene specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

Limitations

The MWH Foresight Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. 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 (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between the chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported, if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined by, Myriad Women's Health, 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 evaluation. CLIA Number: #05D1102604.



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Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no, clinical significance.

If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

Resources

GENOME CONNECT | <http://www.genomeconnect.org>

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

SENIOR LABORATORY DIRECTOR

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Erik Zmuda, PhD, Diplomate of the American Board of Medical Genetics and Genomics, CGMB on Mar 16, 2022

Conditions Tested

6-pyruvoyl-tetrahydropterin Synthase Deficiency - Gene: PTS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000317:1-6. **Detection Rate:** Southeast Asian >99%.

Adenosine Deaminase Deficiency - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000022:1-12. **Detection Rate:** Southeast Asian 98%.

Alpha Thalassemia, HBA1/HBA2-related - Genes: HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. Exons: NM_000517:1-3; NM_000558:1-3. Variants (16): -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Southeast Asian >99%.

Alpha-mannosidosis - Gene: MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000528:1-23. **Detection Rate:** Southeast Asian >99%.

Alpha-sarcoglycanopathy - Gene: SGCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000023:1-9. **Detection Rate:** Southeast Asian >99%.

Alstrom Syndrome - Gene: ALMS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015120:1-23. **Detection Rate:** Southeast Asian >99%.

Andermann Syndrome - Gene: SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_133647:1-25. **Detection Rate:** Southeast Asian >99%.

Argininemia - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000045:1-8. **Detection Rate:** Southeast Asian 97%.

Argininosuccinic Aciduria - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001024943:1-16. **Detection Rate:** Southeast Asian >99%.

Aspartylglucosaminuria - Gene: AGA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000027:1-9. **Detection Rate:** Southeast Asian >99%.

Ataxia with Vitamin E Deficiency - Gene: TTPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000370:1-5. **Detection Rate:** Southeast Asian >99%.

Ataxia-telangiectasia - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000051:2-63. **Detection Rate:** Southeast Asian 96%.

ATP7A-related Disorders - Gene: ATP7A. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000052:2-23. **Detection Rate:** Southeast Asian 90%.

Autoimmune Polyglandular Syndrome Type 1 - Gene: AIRE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000383:1-14. **Detection Rate:** Southeast Asian >99%.

Autosomal Recessive Osteopetrosis Type 1 - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006019:2-20. **Detection Rate:** Southeast Asian 96%.

Autosomal Recessive Polycystic Kidney Disease, PKHD1-related - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_138694 2-67. **Detection Rate:** Southeast Asian >99%.

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - Gene: SACS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_014363 2-10. **Detection Rate:** Southeast Asian 99%.

Bardet-Biedl Syndrome, BBS1-related - Gene: BBS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024649:1-17. **Detection Rate:** Southeast Asian >99%.

Bardet-Biedl Syndrome, BBS10-related - Gene: BBS10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024685:1-2. **Detection Rate:** Southeast Asian >99%.

Bardet-Biedl Syndrome, BBS12-related - Gene: BBS12. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_152618:2. **Detection Rate:** Southeast Asian >99%.

Bardet-Biedl Syndrome, BBS2-related - Gene: BBS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_031885:1-17. **Detection Rate:** Southeast Asian >99%.

BCS1L-related Disorders - Gene: BCS1L. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004328:3-9. **Detection Rate:** Southeast Asian >99%.

Beta-sarcoglycanopathy - Gene: SGCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000232:1-6. **Detection Rate:** Southeast Asian >99%.

Biotinidase Deficiency - Gene: BTD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000060:1-4. **Detection Rate:** Southeast Asian >99%.

Bloom Syndrome - Gene: BLM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000057:2-22. **Detection Rate:** Southeast Asian >99%.

Calpainopathy - Gene: CAPN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000070:1-24. **Detection Rate:** Southeast Asian 99%.

Canavan Disease - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000049:1-6. **Detection Rate:** Southeast Asian 98%.

Carbamoylphosphate Synthetase I Deficiency - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001875:1-38. **Detection Rate:** Southeast Asian >99%.

Carnitine Palmitoyltransferase IA Deficiency - Gene: CPT1A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001876:2-19. **Detection Rate:** Southeast Asian >99%.

Carnitine Palmitoyltransferase II Deficiency - Gene: CPT2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000098:1-5. **Detection Rate:** Southeast Asian >99%.

Cartilage-hair Hypoplasia - Gene: RMRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NR_003051:1. **Detection Rate:** Southeast Asian >99%.

Cerebrotendinous Xanthomatosis - Gene: CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000784:1-9. **Detection Rate:** Southeast Asian >99%.

Citrullinemia Type 1 - Gene: ASS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000050:3-16. **Detection Rate:** Southeast Asian >99%.

CLN3-related Neuronal Ceroid Lipofuscinosis - Gene: CLN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001042432 2-16. **Detection Rate:** Southeast Asian >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - Gene: CLN5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006493:1-4. **Detection Rate:** Southeast Asian >99%.

CLN8-related Neuronal Ceroid Lipofuscinosis - Gene: CLN8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_018941:2-3. **Detection Rate:** Southeast Asian >99%.

Cohen Syndrome - Gene: VPS13B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017890:2-62. **Detection Rate:** Southeast Asian 97%.

COL4A3-related Alport Syndrome - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000091:1-52. **Detection Rate:** Southeast Asian 94%.

COL4A4-related Alport Syndrome - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000092:2-48. **Detection Rate:** Southeast Asian >99%.

Combined Pituitary Hormone Deficiency, PROP1-related - Gene: PROP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006261:1-3. **Detection Rate:** Southeast Asian >99%.

Congenital Adrenal Hyperplasia, CYP11B1-related - Gene: CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000497:1-9. **Detection Rate:** Southeast Asian 97%.

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. Detection Rate: Southeast Asian 88%.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000303:1-8. Detection Rate: Southeast Asian >99%.

Congenital Disorder of Glycosylation Type Ic - Gene: ALG6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_013339:2-15. Detection Rate: Southeast Asian >99%.

Congenital Disorder of Glycosylation, MPI-related - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002435:1-8. Detection Rate: Southeast Asian >99%.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_025136:1-2. Detection Rate: Southeast Asian >99%.

Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. Detection Rate: Southeast Asian >99%.

Cystinosis - Gene: CTNS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004937:3-12. Detection Rate: Southeast Asian >99%.

D-bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000414:1-24. Detection Rate: Southeast Asian 98%.

Delta-sarcoglycanopathy - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000337:2-9. Detection Rate: Southeast Asian 96%.

Dihydroliipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000108:1-14. Detection Rate: Southeast Asian >99%.

Dysferlinopathy - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003494:1-55. Detection Rate: Southeast Asian 98%.

Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - Gene: DMD. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_004006:1-79. Detection Rate: Southeast Asian 99%.

ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000124:2-21. Detection Rate: Southeast Asian 96%.

ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000082:1-12. Detection Rate: Southeast Asian 97%.

EVC-related Ellis-van Creveld Syndrome - Gene: EVC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153717:1-21. Detection Rate: Southeast Asian 96%.

EVC2-related Ellis-van Creveld Syndrome - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_147127:1-22. Detection Rate: Southeast Asian 98%.

Fabry Disease - Gene: GLA. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000169:1-7. Detection Rate: Southeast Asian 98%.

Familial Dysautonomia - Gene: ELP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003640:2-37. Detection Rate: Southeast Asian >99%.

Familial Hyperinsulinism, ABCC8-related - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000352:1-39. Detection Rate: Southeast Asian >99%.

Familial Hyperinsulinism, KCNJ11-related - Gene: KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000525:1. Detection Rate: Southeast Asian >99%.

Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000243:1-10. Detection Rate: Southeast Asian >99%.

Fanconi Anemia Complementation Group A - Gene: FANCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000135:1-43. Detection Rate: Southeast Asian 92%.

Fanconi Anemia, FANCC-related - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000136:2-15. Detection Rate: Southeast Asian >99%.

FKRP-related Disorders - Gene: FKRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_024301:4. Detection Rate: Southeast Asian >99%.

FKTN-related Disorders - Gene: FKTN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001079802:3-11. Detection Rate: Southeast Asian >99%.

Free Sialic Acid Storage Disorders - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012434:1-11. Detection Rate: Southeast Asian 98%.

Galactokinase Deficiency - Gene: GALK1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000154:1-8. Detection Rate: Southeast Asian >99%.

Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000155:1-11. Detection Rate: Southeast Asian >99%.

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. Detection Rate: Southeast Asian 87%.

Gaucher Disease - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. Variants (10): D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. Detection Rate: Southeast Asian 60%.

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004004:1-2. Detection Rate: Southeast Asian >99%.

GLB1-related Disorders - Gene: GLB1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000404:1-16. Detection Rate: Southeast Asian >99%.

Glutaric Acidemia, GCDH-related - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000159:2-12. Detection Rate: Southeast Asian >99%.

Glycine Encephalopathy, AMT-related - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000481:1-9. Detection Rate: Southeast Asian >99%.

Glycine Encephalopathy, GLDC-related - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000170:1-25. Detection Rate: Southeast Asian 94%.

Glycogen Storage Disease Type Ia - Gene: G6PC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000151:1-5. Detection Rate: Southeast Asian 98%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001164277 3-11. Detection Rate: Southeast Asian >99%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000642:2-34. Detection Rate: Southeast Asian >99%.

GNE Myopathy - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001128227:1-12. Detection Rate: Southeast Asian >99%.

GNPTAB-related Disorders - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024312:1-21. Detection Rate: Southeast Asian >99%.

HADHA-related Disorders - Gene: HADHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000182:1-20. Detection Rate: Southeast Asian >99%.

Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000518:1-3. Detection Rate: Southeast Asian >99%.

Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000035:2-9. Detection Rate: Southeast Asian >99%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000520:1-14. **Detection Rate:** Southeast Asian >99%.

HMG-CoA Lyase Deficiency - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000191:1-9. **Detection Rate:** Southeast Asian >99%.

Holocarboxylase Synthetase Deficiency - Gene: HLCS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000411:4-12. **Detection Rate:** Southeast Asian >99%.

Homocystinuria, CBS-related - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000071:3-17. **Detection Rate:** Southeast Asian >99%.

Hydroletharus Syndrome - Gene: HYL1. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_145014:4. **Detection Rate:** Southeast Asian >99%.

Hypophosphatasia - Gene: ALPL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000478:2-12. **Detection Rate:** Southeast Asian >99%.

Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002225:1-12. **Detection Rate:** Southeast Asian >99%.

Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001173990:1-5. **Detection Rate:** Southeast Asian >99%.

Junctional Epidermolysis Bullosa, LAMA3-related - Gene: LAMA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000227:1-38. **Detection Rate:** Southeast Asian >99%.

Junctional Epidermolysis Bullosa, LAMB3-related - Gene: LAMB3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000228:2-23. **Detection Rate:** Southeast Asian >99%.

Junctional Epidermolysis Bullosa, LAMC2-related - Gene: LAMC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_005562:1-23. **Detection Rate:** Southeast Asian >99%.

Krabbe Disease - Gene: GALC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000153:1-17. **Detection Rate:** Southeast Asian >99%.

Leigh Syndrome, French-Canadian Type - Gene: LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_133259:1-38. **Detection Rate:** Southeast Asian >99%.

Lipoid Congenital Adrenal Hyperplasia - Gene: STAR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000349:1-7. **Detection Rate:** Southeast Asian >99%.

Lysosomal Acid Lipase Deficiency - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000235:2-10. **Detection Rate:** Southeast Asian 98%.

Maple Syrup Urine Disease Type Ia - Gene: BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000709:1-9. **Detection Rate:** Southeast Asian >99%.

Maple Syrup Urine Disease Type Ib - Gene: BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_183050:1-10. **Detection Rate:** Southeast Asian >99%.

Maple Syrup Urine Disease Type II - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001918:1-11. **Detection Rate:** Southeast Asian 97%.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000016:1-12. **Detection Rate:** Southeast Asian >99%.

Megalencephalic Leukoencephalopathy with Subcortical Cysts - Gene: MLC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015166:2-12. **Detection Rate:** Southeast Asian >99%.

Metachromatic Leukodystrophy - Gene: ARSA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000487:1-8. **Detection Rate:** Southeast Asian >99%.

Methylmalonic Acidemia, cblA Type - Gene: MMAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_172250:2-7. **Detection Rate:** Southeast Asian >99%.

Methylmalonic Acidemia, cblB Type - Gene: MMAB. Autosomal Recessive.

Sequencing with copy number analysis. Exons: NM_052845:1-9. **Detection Rate:** Southeast Asian >99%.

Methylmalonic Acidemia, MMUT-related - Gene: MMUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000255:2-13. **Detection Rate:** Southeast Asian >99%.

Methylmalonic Aciduria and Homocystinuria, cblC Type - Gene: MMACHC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015506:1-4. **Detection Rate:** Southeast Asian >99%.

MKS1-related Disorders - Gene: MKS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017777:1-18. **Detection Rate:** Southeast Asian >99%.

Mucopolidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** Southeast Asian 98%.

Mucopolidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_020533:1-14. **Detection Rate:** Southeast Asian >99%.

Mucopolysaccharidosis Type I - Gene: IDUA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000203:1-14. **Detection Rate:** Southeast Asian >99%.

Mucopolysaccharidosis Type II - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000202:1-9. **Detection Rate:** Southeast Asian 89%.

Mucopolysaccharidosis Type IIIA - Gene: SGSH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000199:1-8. **Detection Rate:** Southeast Asian >99%.

Mucopolysaccharidosis Type IIIB - Gene: NAGLU. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000263:1-6. **Detection Rate:** Southeast Asian >99%.

Mucopolysaccharidosis Type IIIC - Gene: HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_152419:1-18. **Detection Rate:** Southeast Asian >99%.

Muscular Dystrophy, LAMA2-related - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000426:1-43,45-65. **Detection Rate:** Southeast Asian 98%.

MYO7A-related Disorders - Gene: MYO7A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000260:2-49. **Detection Rate:** Southeast Asian >99%.

NEB-related Nemaline Myopathy - Gene: NEB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001271208:3-80,117-183. **Detection Rate:** Southeast Asian 92%.

Nephrotic Syndrome, NPHS1-related - Gene: NPHS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004646:1-29. **Detection Rate:** Southeast Asian >99%.

Nephrotic Syndrome, NPHS2-related - Gene: NPHS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_014625:1-8. **Detection Rate:** Southeast Asian >99%.

Neuronal Ceroid Lipofuscinosis, CLN6-related - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017882:1-7. **Detection Rate:** Southeast Asian >99%.

Niemann-Pick Disease Type C1 - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000271:1-25. **Detection Rate:** Southeast Asian >99%.

Niemann-Pick Disease Type C2 - Gene: NPC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006432:1-5. **Detection Rate:** Southeast Asian >99%.

Niemann-Pick Disease, SMPD1-related - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000543:1-6. **Detection Rate:** Southeast Asian >99%.

Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002485:1-16. **Detection Rate:** Southeast Asian >99%.

Ornithine Transcarbamylase Deficiency - Gene: OTC. X-linked Recessive.

Sequencing with copy number analysis. Exons: NM_000531:1-10. **Detection Rate:** Southeast Asian 97%.

PCCA-related Propionic Acidemia - Gene: PCCA. Autosomal Recessive.

Sequencing with copy number analysis. Exons: NM_000282:1-24. **Detection Rate:** Southeast Asian 95%.

PCCB-related Propionic Acidemia - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000532:1-15. **Detection Rate:** Southeast Asian >99%.

PCDH15-related Disorders - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_033056:2-33. **Detection Rate:** Southeast Asian 93%.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000441:2-21. **Detection Rate:** Southeast Asian >99%.

Peroxisome Biogenesis Disorder Type 1 - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000466:1-24. **Detection Rate:** Southeast Asian >99%.

Peroxisome Biogenesis Disorder Type 3 - Gene: PEX12. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000286:1-3. **Detection Rate:** Southeast Asian >99%.

Peroxisome Biogenesis Disorder Type 4 - Gene: PEX6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000287:1-17. **Detection Rate:** Southeast Asian 97%.

Peroxisome Biogenesis Disorder Type 5 - Gene: PEX2. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000318:4. **Detection Rate:** Southeast Asian >99%.

Peroxisome Biogenesis Disorder Type 6 - Gene: PEX10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153818:1-6. **Detection Rate:** Southeast Asian >99%.

Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000277:1-13. **Detection Rate:** Southeast Asian >99%.

POMGNT-related Disorders - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017739:2-22. **Detection Rate:** Southeast Asian 96%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. **Detection Rate:** Southeast Asian >99%.

PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000310:1-9. **Detection Rate:** Southeast Asian >99%.

Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003060:1-10. **Detection Rate:** Southeast Asian >99%.

Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000030:1-11. **Detection Rate:** Southeast Asian >99%.

Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012203:1-9. **Detection Rate:** Southeast Asian >99%.

Primary Hyperoxaluria Type 3 - Gene: HOGA1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_138413:1-7. **Detection Rate:** Southeast Asian >99%.

Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000396:2-8. **Detection Rate:** Southeast Asian >99%.

Pyruvate Carboxylase Deficiency - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000920:3-22. **Detection Rate:** Southeast Asian >99%.

Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000288:1-10. **Detection Rate:** Southeast Asian >99%.

RTEL1-related Disorders - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032957:2-35. **Detection Rate:** Southeast Asian >99%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. **Detection Rate:** Southeast Asian 98%.

Short-chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000017:1-10. **Detection Rate:** Southeast Asian >99%.

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000382:1-10. **Detection Rate:** Southeast Asian 96%.

SLC26A2-related Disorders - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000112:2-3. **Detection Rate:** Southeast Asian >99%.

Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001360:3-9. **Detection Rate:** Southeast Asian >99%.

Spastic Paraplegia Type 15 - Gene: ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015346:2-42. **Detection Rate:** Southeast Asian >99%.

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** Southeast Asian 93%.

Spondylthoracic Dysostosis - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001039958:1-2. **Detection Rate:** Southeast Asian >99%.

TGM1-related Autosomal Recessive Congenital Ichthyosis - Gene: TGM1.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000359 2-15. **Detection Rate:** Southeast Asian >99%.

TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000391:1-13. **Detection Rate:** Southeast Asian >99%.

Tyrosine Hydroxylase Deficiency - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_199292:1-14. **Detection Rate:** Southeast Asian >99%.

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000137:1-14. **Detection Rate:** Southeast Asian >99%.

Tyrosinemia Type II - Gene: TAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000353:2-12. **Detection Rate:** Southeast Asian >99%.

USH1C-related Disorders - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_005709:1-21. **Detection Rate:** Southeast Asian >99%.

USH2A-related Disorders - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_206933:2-72. **Detection Rate:** Southeast Asian 98%.

Usher Syndrome Type 3 - Gene: CLRN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_174878:1-3. **Detection Rate:** Southeast Asian >99%.

Very-long-chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000018:1-20. **Detection Rate:** Southeast Asian >99%.

Wilson Disease - Gene: ATP7B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000053:1-21. **Detection Rate:** Southeast Asian >99%.

X-linked Adrenal Hypoplasia Congenita - Gene: NROB1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** Southeast Asian 97%.

X-linked Adrenoleukodystrophy - Gene: ABCD1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000033:1-6. **Detection Rate:** Southeast Asian 77%.

X-linked Alport Syndrome - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000495:1-51. **Detection Rate:** Southeast Asian 96%.

X-linked Juvenile Retinoschisis - Gene: RS1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000330:1-6. **Detection Rate:** Southeast Asian 98%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 03/16/2022

MALE
DONOR 10632
DOB [REDACTED]
Ethnicity: Southeast Asian
Barcode: 11004512970442

FEMALE
N/A

X-linked Myotubular Myopathy - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000252:2-15. **Detection Rate:** Southeast Asian 96%.

X-linked Severe Combined Immunodeficiency - Gene: IL2RG. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000206:1-8. **Detection Rate:** Southeast Asian >99%.

Xeroderma Pigmentosum Group A - Gene: XPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000380:1-6. **Detection Rate:** Southeast Asian >99%.

Xeroderma Pigmentosum Group C - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004628:1-16. **Detection Rate:** Southeast Asian 97%.

Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' 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 given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '+' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Disease	DONOR 10632 Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	1 in 33,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 34,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	1 in 800,000	1 in 150,000
Autoimmune Polyglandular Syndrome Type 1	1 in 18,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 39,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 18,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	CAPN3 Exon 20-Partial Exon 24 del [NM_000070.2:c.2115+111_*6del] †	1 in 560
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 18,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	NM_000050.4(ASS1):c.910C>T(R304W) heterozygote †	1 in 480
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 13,000	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 5,800	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 35,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related	1 in 8,400	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 410	1 in 80,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10632 Residual Risk	Reproductive Risk
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 9,000	< 1 in 1,000,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 13,000	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,400	< 1 in 1,000,000
ERCC8-related Disorders	1 in 12,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 80,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 50,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 3,100	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 32,000	< 1 in 1,000,000
FTKN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 44,000	< 1 in 1,000,000
Galactosemia	1 in 11,000	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 2,600	< 1 in 1,000,000
Gaucher Disease	1 in 310	1 in 150,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	NM_004004.5(GJB2):c.109G>A(V37I) heterozygote [†]	1 in 140
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	< 1 in 1,000,000
Glycine Encephalopathy, GLDC-related	1 in 4,100	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 8,700	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	< 1 in 50,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 25,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 1,700	1 in 120,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 27,000	< 1 in 1,000,000
Hydroletharus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 23,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 26,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 31,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 17,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	< 1 in 34,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 19,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 36,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 5,700	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 6,000	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000

Disease	DONOR 10632 Residual Risk	Reproductive Risk
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, MMUT-related	1 in 6,300	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucopolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	< 1 in 1,000,000	1 in 300,000
Mucopolysaccharidosis Type IIIA	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 26,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 400,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 17,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	< 1 in 50,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	1 in 140,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 6,400	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 10,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 16,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 13,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 20,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	1 in 18,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 9,700	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	< 1 in 50,000	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 700	1 in 150,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 5,900	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 03/16/2022

MALE
DONOR 10632
DOB: [REDACTED]
Ethnicity: Southeast Asian
Barcode: 11004512970442

FEMALE
N/A

Disease	DONOR 10632 Residual Risk	Reproductive Risk
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 14,000	< 1 in 1,000,000
Wilson Disease	1 in 6,500	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 50,000
X-linked Myotubular Myopathy	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	1 in 200,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000

Patient name:	Donor 10632	Sample type:	Blood	Report date:	26-JAN-2023
DOB:	██████████	Sample collection date:	18-JAN-2023	Invitae #:	RQ4563994
Sex assigned at birth:	Male	Sample accession date:	19-JAN-2023	Clinical team:	Guadalupe Martinez Jeffrey Olliffe
Gender:					
Patient ID (MRN):					

Reason for testing

Gamete donor

Test performed

Invitae Carrier Screen


RESULT: NEGATIVE

This carrier test evaluated 1 gene(s) for genetic changes (variants) that are associated with an increased risk of having a child with a genetic condition. Knowledge of carrier status for one of these conditions may provide information that can be used to assist with family planning and/or preparation. Carrier screening is not intended for diagnostic purposes. To identify a potential genetic basis for a condition in the individual being tested, diagnostic testing for the gene(s) of interest is recommended.

This test did not identify any genetic changes in the gene(s) analyzed that are currently recognized as clinically significant. This negative result reduces, but does not eliminate, the chance that this individual is a carrier for conditions caused by any of the genes tested. This individual may still be a carrier for a genetic condition that is not evaluated by this test.

Next steps

- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called “residual risk.” See the Carrier detection rates and residual risks document.
- Discussion with a physician and/or genetic counselor is recommended to further review the implications of this test result and to understand these results in the context of any family history of a genetic condition.
- All patients, regardless of result, may wish to consider additional screening for hemoglobinopathies by complete blood count (CBC) and hemoglobin electrophoresis, if this has not already been completed.
- Individuals can register their tests at <https://www.invitae.com/patients/> to access online results, educational resources, and next steps.



Patient name: Donor 10632 DOB: [REDACTED]

Invitae #: RQ4563994

Residual risk

No carrier test can detect 100% of carriers. There still remains a small risk of being a carrier after a negative test (residual risk). Residual risk values assume a negative family history and are inferred from published carrier frequencies and estimated detection rates based on testing technologies used at Invitae. You can view Invitae's complete Carrier detection rates and residual risks document (containing all carrier genes) online at <https://www.invitae.com/carrier-residual-risks/>. Additionally, the order-specific information for this report is available to download in the portal (under this order's documents) or can be requested by contacting Invitae Client Services. The complete Carrier detection rates and residual risks document will not be applicable for any genes with specimen-specific limitations in sequencing and/or deletion/duplication coverage. Please see the final bullet point in the Limitations section of this report to view if this specimen had any gene-specific coverage gaps.



Patient name: Donor 10632 DOB: [REDACTED]

Invitae #: RQ4563994

Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details. Results are negative, unless otherwise indicated in the report.

GENE	TRANSCRIPT
GBE1	NM_000158.3

Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with $\geq 50\times$ depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated in the Genes analyzed section. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Invitae utilizes a classification methodology to identify next-generation sequencing (NGS)-detected variants that require orthogonal confirmation (Lincoln, et al. J Mol Diagn. 2019 Mar;21(2):318-329.). Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For GBA the reference genome has been modified to mask the sites of polymorphic paralog sequence variants (PSVs) in both the gene and pseudogene. For CYP21A2 and GBA, if one or more reportable variants, gene conversion, or fusion event is identified via our NGS pipeline (see Limitations), these variants are confirmed by PacBio sequencing of an amplicon generated by long-range PCR and subsequent short-range PCR. In some cases, it may not be possible to disambiguate between the gene and pseudogene. For GJB2, the reportable range includes large upstream deletions overlapping GJB6. For HBA1/2, the reference genome has been modified to force some sequencing reads derived from HBA1 to align to HBA2, and variant calling algorithms are modified to support an expectation of 4 alleles in these regions. HBA1/2 copy number calling is performed by a custom hypothesis testing algorithm which generates diplotype calls. If sequence data for a sample does not support a unique high confidence match from among hypotheses tested, that sample is flagged for manual review. Copy number variation is only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the $\alpha 3.7$ subtypes, and all $\alpha 3.7$ variants are called as HBA1 deletions. This assay may not detect overlapping copy gain and copy loss events when the breakpoints of those events are similar. For FMR1, triplet repeats are detected by PCR with fluorescently labeled primers followed by capillary electrophoresis. Reference ranges: Normal: <45 CGG repeats, intermediate: 45-54 CGG repeats, premutation: 55-200 CGG repeats, full mutation: >200 CGG repeats. For alleles with 55-90 triplet repeats, the region surrounding the FMR1 repeat is amplified by PCR. The PCR amplicons are then processed through PacBio SMRTBell library prep and sequenced using PacBio long read technology. The number of AGG interruptions within the 55-90 triplet repeat is read directly from the resulting DNA sequences. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).
- This report only includes variants that have a clinically significant association with the conditions tested as of the report date. Variants of uncertain significance, benign variants, and likely benign variants are not included in this report. However, if additional evidence becomes available to indicate that the clinical significance of a variant has changed, Invitae may update this report and provide notification.
- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at <http://www.ncbi.nlm.nih.gov/pubmed>.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (<http://exac.broadinstitute.org>), gnomAD (<http://gnomad.broadinstitute.org>), and dbSNP (<http://ncbi.nlm.nih.gov/SNP>).

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Invitae. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.



Patient name: Donor 10632

DOB: [REDACTED]

Invitae #: RQ4563994

Limitations

- Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination.

This report has been reviewed and approved by:



Matteo Vatta, Ph.D., FACMG
Clinical Molecular Geneticist

This table displays residual risks after a negative result for each of the genes and corresponding disorders. The values provided assume a negative family history and the absence of symptoms for each disorder. For genes associated with both dominant and recessive inheritance, the numbers in this table apply to the recessive condition(s) associated with the gene, unless otherwise noted. Residual risk values are provided for disorders when carrier frequency is greater than 1 in 500. For disorders with carrier frequency equal to, or less than, 1 in 500, residual risk is considered to be reduced substantially. When provided, residual risk values are inferred from published carrier frequencies, and estimated detection rates are based on testing technologies used at Invitae. Residual risks are provided only as a guide for assessing approximate risk given a negative result; values may vary based on the ethnic background(s) of an individual. For any genes marked with an asterisk*, refer to the Limitations section of the patient report for detailed coverage information. In the case of a sample-specific limitation, "N/A" indicates that a residual risk value could not be calculated. AR = autosomal recessive, XL = X-linked, AD = autosomal dominant.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
GBE1-related conditions (AR) NM_000158.3	GBE1	Pan-ethnic	1 in 387	99%	1 in 38600

Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 055-944-5005-0

Patient Name: **10632, DONOR**

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU22-17049 L

Indications: NOT GIVEN

Account Number: 02003540

Ordering Physician: **J OLLIFFE**

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 02/24/2022

Date Received: 02/26/2022

Date Reported: **03/12/2022**

Test: **Chromosome, Blood, Routine**

Cells Counted: 35

Cells Analyzed: 20

Cells Karyotyped: 2

Band Resolution: 500

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Technical Component-Processing performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Anjen Chenn, M.D., Ph.D.

Technical Component-Chromosome analysis performed at Virtual Scientific, Inc., CLIA# 37D2202662. 148 Cubbage Dr. Rincon, GA 31326. Laboratory Director Lisa R Smith, Ph.D.

Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 055-944-5005-0

Patient Name: 10632, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU22-17049 L

Account Number: 02003540

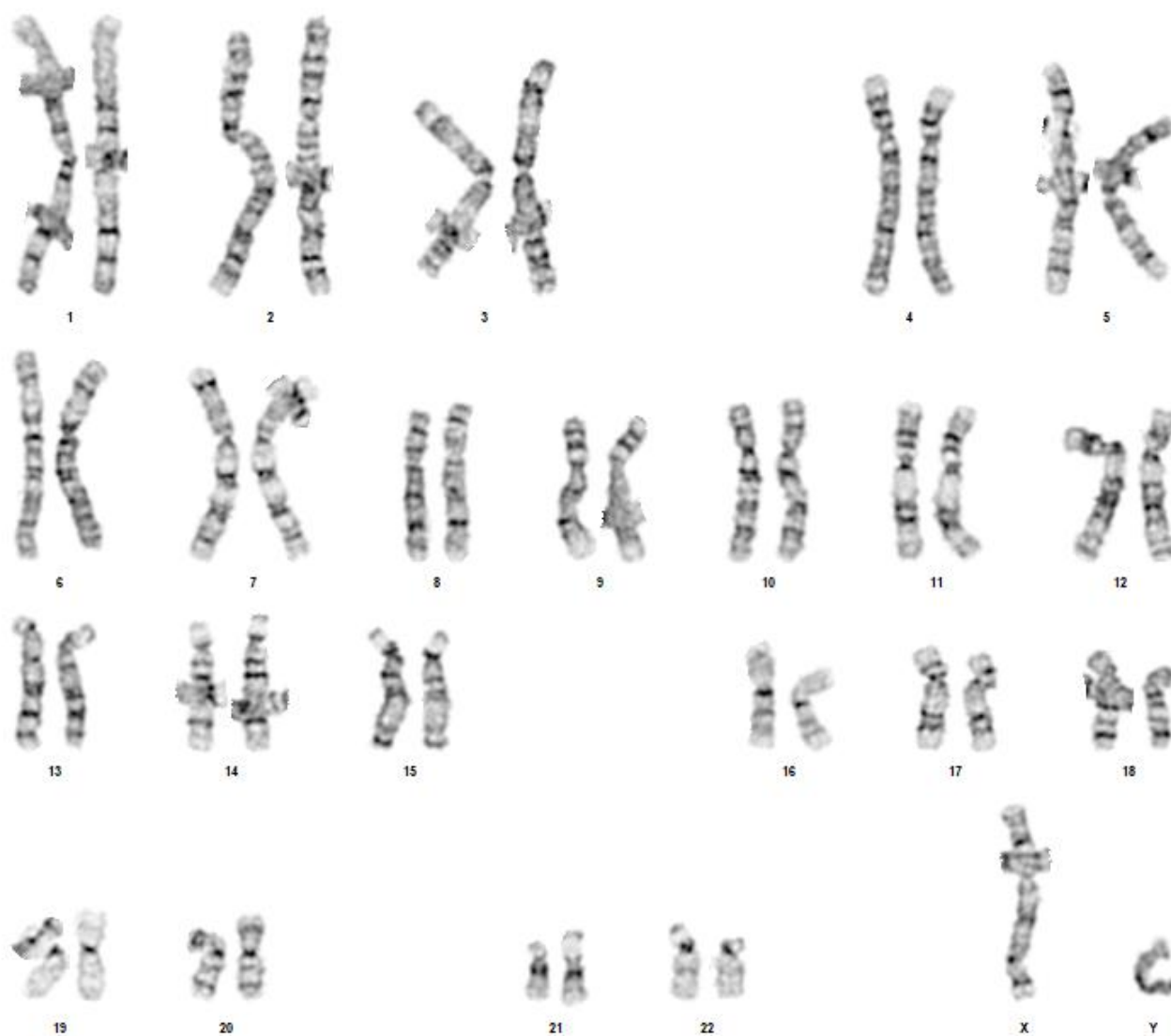
Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/24/2022

Date Received: 02/26/2022





Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 055-944-5005-0

Patient Name: **10632, DONOR**

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU22-17049 L

Account Number: 02003540

Ordering Physician: **J OLLIFFE**

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 02/24/2022

Date Received: 02/26/2022

Inder K. Gadi, PhD, FACMG

Anjen Chenn, M.D., Ph.D.

Medical Director

Stuart Schwartz, PhD

National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive , RTP , NC , 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709 Medical Director, Anjen Chenn, M D , Ph D
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings

This document contains private and confidential health information **protected by state and federal law.**

10632, Donor

Patient ID:

Specimen ID: 055 944 3947 0

DOB:

Age:

Sex: Male

Patient Report

Account Number: 02003540

Ordering Physician: J OLLIFFE



Ordered Items: LP+12AC+CBC/D/Plt+UA+Rh+ABO...

Date Collected: 02/24/2022

Date Received: 02/25/2022

Date Reported: 03/01/2022

Fasting: No

LP+12AC+CBC/D/Plt+UA+Rh+ABO...

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Glucose ⁰¹	79		mg/dL	65-99
Uric Acid ⁰¹	7.2		mg/dL	3.8-8.4
Therapeutic target for gout patients: <6.0				
BUN ⁰¹	18		mg/dL	6-20
▲ Creatinine ⁰¹	1.32 High		mg/dL	0.76-1.27
Effective February 28, 2022 Labcorp will begin reporting the 2021 CKD-EPI creatinine equation that estimates kidney function without a race variable.				
eGFR If NonAfricn Am	72		mL/min/1.73	>59
eGFR If Africn Am	83		mL/min/1.73	>59
In accordance with recommendations from the NKF-ASN Task force, Labcorp is in the process of updating its eGFR calculation to the 2021 CKD-EPI creatinine equation that estimates kidney function without a race variable.				
Calcium ⁰¹	9.6		mg/dL	8.7-10.2
Protein, Total ⁰¹	7.1		g/dL	6.0-8.5
Albumin ⁰¹	4.4		g/dL	4.1-5.2
Bilirubin, Total ⁰¹	0.5		mg/dL	0.0-1.2
Alkaline Phosphatase ⁰¹	77		IU/L	44-121
LDH ⁰¹	199		IU/L	121-224
AST (SGOT) ⁰¹	32		IU/L	0-40
ALT (SGPT) ⁰¹	35		IU/L	0-44
Cholesterol, Total ⁰¹	192		mg/dL	100-199
▲ Triglycerides ⁰¹	209 High		mg/dL	0-149
▼ HDL Cholesterol ⁰¹	37 Low		mg/dL	>39
▲ LDL Chol Calc (NIH)	118 High		mg/dL	0-99
LDL/HDL Ratio	3.2		ratio	0.0-3.6
Please Note: ⁰¹				

LDL/HDL Ratio

	Men	Women
1/2 Avg. Risk	1.0	1.5
Avg. Risk	3.6	3.2
2X Avg. Risk	6.2	5.0
3X Avg. Risk	8.0	6.1

Hgb Fractionation by CE: ⁰²				
Hgb F ⁰²	0.0		%	0.0-2.0
Hgb A ⁰²	97.4		%	96.4-98.8
Hgb A2 ⁰²	2.6		%	1.8-3.2
Hgb S ⁰²	0.0		%	0.0
Interpretation: ⁰²				

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LP+12AC+CBC/D/Plt+UA+Rh+ABO... (Cont.)

Normal hemoglobin present; no hemoglobin variant or thalassemia observed.

ABO Grouping ⁰¹	O		
Rh Factor ⁰¹	Positive		
Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.			
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CBC, Platelet Ct, and Diff ⁰¹			
WBC ⁰¹	5.9	x10E3/uL	3.4-10.8
▲ RBC ⁰¹	6.45	High	x10E6/uL 4.14-5.80
▲ Hemoglobin ⁰¹	19.3	High	g/dL 13.0-17.7
▲ Hematocrit ⁰¹	56.3	High	% 37.5-51.0
MCV ⁰¹	87	fL	79-97
MCH ⁰¹	29.9	pg	26.6-33.0
MCHC ⁰¹	34.3	g/dL	31.5-35.7
RDW ⁰¹	14.2	%	11.6-15.4
Platelets ⁰¹	251	x10E3/uL	150-450
Neutrophils ⁰¹	53	%	Not Estab.
Lymphs ⁰¹	31	%	Not Estab.
Monocytes ⁰¹	11	%	Not Estab.
Eos ⁰¹	4	%	Not Estab.
Basos ⁰¹	1	%	Not Estab.
Neutrophils (Absolute) ⁰¹	3.1	x10E3/uL	1.4-7.0
Lymphs (Absolute) ⁰¹	1.8	x10E3/uL	0.7-3.1
Monocytes(Absolute) ⁰¹	0.7	x10E3/uL	0.1-0.9
Eos (Absolute) ⁰¹	0.2	x10E3/uL	0.0-0.4
Baso (Absolute) ⁰¹	0.1	x10E3/uL	0.0-0.2
Immature Granulocytes ⁰¹	0	%	Not Estab.
Immature Grans (Abs) ⁰¹	0.0	x10E3/uL	0.0-0.1
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Urinalysis Gross Exam ⁰¹			
Specific Gravity ⁰¹	1.023		1.005-1.030
pH ⁰¹	6.0		5.0-7.5
Urine-Color ⁰¹	Yellow		Yellow
Appearance ⁰¹	Clear		Clear
WBC Esterase ⁰¹	Negative		Negative
Protein ⁰¹	Negative		Negative/Trace
Glucose ⁰¹	Negative		Negative
Ketones ⁰¹	Negative		Negative
Occult Blood ⁰¹	Negative		Negative
Bilirubin ⁰¹	Negative		Negative
Urobilinogen,Semi-Qn ⁰¹	0.2	mg/dL	0.2-1.0
Nitrite, Urine ⁰¹	Negative		Negative
Microscopic Examination ⁰¹			
Microscopic not indicated and not performed.			