RESULTS RECIPIENT
UND CENTER FOR FAMILY MEDICINE
Attn: Jacqueline Quisno
701 E Rosser Ave
Bismarck, ND 58501
Phone: (701) 751-6743
Fax: (701) 751-9560
NPI: 1326016163
Report Date: 09/25/2023

MALE
CARSON KUNZ
DOB: 07/01/1990
Ethnicity: Mixed or Other Caucasian
Sample Type: EDTA Blood
Date of Collection: 09/15/2023
Date Received: 09/18/2023
Date Tested: 09/24/2023
Barcode: 11004513356767
Indication: Other genetic carrier status, Other: Z31.83

FEMALE
BROOKLYN KUNZ
DOB: 01/06/1993
Ethnicity: Mixed or Other Caucasian
Sample Type: EDTA Blood
Date of Collection: 09/14/2023
Date Received: 09/15/2023
Date Tested: 09/20/2023
Barcode: 11004513358240
Indication: Other genetic carrier status, Other: Z13.71

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	CARSON KUNZ	BROOKLYN KUNZ
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel **(175 conditions tested)**	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel Fragile X Syndrome **(176 conditions tested)**
POSITIVE: CARRIER Congenital Adrenal Hyperplasia, CYP21A2-related **Reproductive Risk: 1 in 5,100** **Inheritance: Autosomal Recessive**	**⊕ CARRIER*** NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote	**⊖ NEGATIVE** No disease-causing mutations detected.
POSITIVE: CARRIER Cystinosis **Reproductive Risk: 1 in 89,000** **Inheritance: Autosomal Recessive**	**⊖ NEGATIVE** No disease-causing mutations detected.	**⊕ CARRIER*** NM_004937.2(CTNS):c.(?_-36009)_(848_?)del(aka 57 kb deletion) heterozygote

*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 11.

CLINICAL NOTES

- Genetic counseling is recommended for reproductive risk assessment in patients with known carrier status.

NEXT STEPS

- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

DISCLAIMERS

- The terms 'male', 'female', 'he', 'she', 'women', and 'men' refer to sex assigned at birth.

POSITIVE: CARRIER

Congenital Adrenal Hyperplasia, CYP21A2-related

Reproductive risk: 1 in 5,100
Risk before testing: 1 in 12,000

Gene: CYP21A2 | **Inheritance Pattern:** Autosomal Recessive

Patient	CARSON KUNZ	BROOKLYN KUNZ
Result	⊕ Carrier	⊖ Negative
Variant(s)	NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote	No disease-causing mutations detected.
Methodology	Analysis of homologous regions (v4.0)	Analysis of homologous regions (v3.2)
Interpretation	This individual is a carrier of congenital adrenal hyperplasia, CYP21A2-related. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T(V282L) is a non-classic congenital adrenal hyperplasia, CYP21A2-related mutation.	This does not rule out the possibility of being a carrier. The post-test risk of being a carrier, assuming a negative family history, is 1 in 1,300.
Detection rate	96%	96%
Variants tested	CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G.	CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G.

What Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

Congenital adrenal hyperplasia (CAH) refers to a group of genetic disorders that affect the body's adrenal glands. The adrenal glands regulate essential functions in the body, including the production of several important hormones. CAH occurs when the adrenal glands are unable to produce these hormones properly, resulting in a hormone imbalance. CAH, CYP21A2-related is caused by mutations in the *CYP21A2* gene. The *CYP21A2* gene produces the 21-hydroxylase enzyme. Another name for this disorder is 21-hydroxylase-deficient CAH (21-OHD CAH).

When the 21-hydroxylase enzyme is missing or present at low levels, the adrenal glands are unable to produce two critical hormones, cortisol and aldosterone. The body responds to this deficiency by producing an excess of male sex hormones, called androgens. Collectively, the excess androgen production and hormone deficiencies can lead to a variety of medical problems, which vary in severity depending on the form of CAH. CAH associated with *CYP21A2* (21-OHD CAH) has two major forms: classic and non-classic.

CLASSIC FORM

The most severe form referred to as classic 21-OHD CAH, can be further divided into two different subtypes: salt wasting and simple virilizing (non-salt wasting) types. The classic salt-wasting type is associated with near-to-complete deficiency of the 21-hydroxylase enzyme, resulting in the complete inability to produce the hormones cortisol and aldosterone. In this type, the body cannot retain enough sodium (salt) and when too much salt is lost in the urine, it may lead to dehydration, vomiting, diarrhea, poor growth, heart-rhythm abnormalities (arrhythmias), and shock (salt wasting). If not properly treated, salt wasting can lead to death in some cases.

Additionally, female newborns often have external genitals that do not clearly appear either male or female (ambiguous genitalia), whereas male newborns may present with enlarged genitals. Signs of early puberty and the exaggerated development of male characteristics (virilization) occur in both males and females with CAH. These symptoms may include: rapid growth and development

in early childhood, but shorter-than-average height in adulthood, abnormal menstruation cycles for females, excess facial hair for females, early facial-hair growth for males, severe acne, and infertility in both men and women. Male characteristics such as muscle bulk and a deep voice can occur in females and in boys (masculinization).

The simple virilizing type of CAH is associated with partial 21-hydroxylase deficiency. Unlike the salt-wasting type, individuals with this condition typically do not experience severe and life-threatening sodium-deficiency symptoms as newborns. However, the majority of female newborns with this type will have ambiguous genitalia, and both male and female children may show signs of early puberty.

NON-CLASSIC FORM

The non-classic type (late-onset type) is the the least-severe form of 21-OHD CAH and is caused by a mild deficiency of the 21-hydroxylase enzyme. Individuals with this type may start experiencing symptoms related to excess androgen production in childhood, adolescence, or adulthood. Both males and females may exhibit rapid growth in childhood, shorter-than-average stature in adulthood, virilization, and infertility. Additionally, girls may experience symptoms of masculinization and abnormal menstruation. However, some individuals with non-classic CAH may never know they are affected because the symptoms are so mild.

How Common Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

The incidence of 21-OHD CAH varies by type and ethnicity. The incidence for the classic form is approximately 1 in 15,000 births worldwide. The prevalence of the classic form varies from 1 in 300 for Yupik Eskimos in Alaska to 1 in 21,000 in Japanese. The non-classic form of 21-OHD CAH is much more common, with an incidence of approximately 1 in 1000 births. The prevalence of the non-classic form is much higher in some ethnicities, namely in the Ashkenazi Jewish (1 in 27), Hispanic (1 in 40), Slavic (1 in 50), and Italian (1 in 300) ethnicities. Mutations in *CYP21A2* account for about 90% of CAH cases.

How Is Congenital Adrenal Hyperplasia, CYP21A2-Related Treated?

Currently, there is no cure for CAH. However, treatments are available to address some of the associated symptoms. Patients benefit from taking hormone-replacement medications, which work to increase levels of deficient hormones and suppress the overproduction of male hormones. Most individuals with classic CAH will need to take hormone medications for the rest of their lives. Those with the less-severe forms of CAH are sometimes able to stop taking these medications in adulthood and are typically treated with lower doses. Some individuals with non-classic CAH do not require any treatment. A multidisciplinary team of physicians, including an endocrinologist, will need to monitor the medication dosage, medication side effects, growth, and sexual development of patients who continue to receive treatment.

Newborn females with ambiguous genitalia may need surgery to correct the function and appearance of the external genitalia. Surgery, if needed, is most often performed during infancy, but can be performed later in life. Treatments provided during pregnancy may reduce the degree of virilization in female fetuses. However, because the long-term safety of prenatal treatment is unknown, these therapies are considered experimental and are not recommended by professional guidelines.

What Is the Prognosis for an Individual with Congenital Adrenal Hyperplasia, CYP21A2-Related?

With early diagnosis and proper medication management, most individuals with 21-OHD CAH will have a normal life expectancy. Early death can occur during periods of significant sodium loss (salt crises) if medication dosage is not adequately adjusted, especially during times of illness or trauma. Problems with growth and development, ambiguous genitalia, and virilization are monitored by physicians on an ongoing basis. Females with 21-OHD CAH can become pregnant, but fertility is reduced.

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MALE
CARSON KUNZ
DOB: 07/01/1990
Ethnicity: Mixed or Other Caucasian
Barcode: 11004513356767

FEMALE
BROOKLYN KUNZ
DOB: 01/06/1993
Ethnicity: Mixed or Other Caucasian
Barcode: 11004513358240

POSITIVE: CARRIER

Cystinosis

Reproductive risk: 1 in 89,000
Risk before testing: 1 in 200,000

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

Patient	CARSON KUNZ	BROOKLYN KUNZ
Result	⊖ Negative	⊕ Carrier
Variant(s)	No disease-causing mutations detected.	NM_004937.2(CTNS):c.(?_-36009)_(848_?)del(aka 57 kb deletion) heterozygote
Methodology	Sequencing with copy number analysis (v4.0)	Sequencing with copy number analysis (v3.1)
Interpretation	This does not rule out the possibility of being a carrier. The post-test risk of being a carrier, assuming a negative family history, is 1 in 22,000.	This individual is a carrier of cystinosis. Carriers generally do not experience symptoms.
Detection rate	>99%	>99%
Exons tested	NM_004937:3-12.	NM_004937:3-12.

What is Cystinosis?

Cystinosis is an inherited disease that causes the amino acid cysteine to accumulate within body cells and form crystals which can damage the body's organs, particularly the kidneys and eyes. Without treatment, children with the condition will experience kidney failure around the age of 10.

There are three forms of cystinosis. The most severe form, nephropathic cystinosis, appears in infants. It causes poor growth and renal tubular Fanconi syndrome, a kidney disorder in which the organ eliminates certain essential nutrients and minerals. The loss of these nutrients inhibits normal body growth and may result in soft, bowed bones. Cysteine crystals also accumulate in the eyes, causing photophobia, an extreme sensitivity to light. Other symptoms may include muscle wasting, difficulty swallowing, diabetes, an underactive thyroid gland, and nervous system problems.

Less severe forms of the disease cause symptoms to begin later in life and may not affect the kidneys.

How common is Cystinosis?

Cystinosis affects approximately 1 in 200,000 people. The disease is most common in Brittany, France, where it affects 1 in 26,000.

How is Cystinosis treated?

Thanks to a drug called cysteamine, cystinosis has become easier to manage. Taken orally in capsules (brand name: Cystagon), it reduces the accumulation of cysteine crystals in the body. The drug has been shown to delay or prevent kidney failure and improve growth rates in children. Cysteamine eye drops have been successful in relieving photophobia in people with cystinosis, although they are not yet approved by the FDA for that purpose.

Nutritional monitoring is important in children with cystinosis. These children require a large amount of water to prevent dehydration. Supplements of several vitamins and minerals are also recommended for most people with the disease. Human growth hormone treatments have been shown to help people with cystinosis reach normal height.

Kidney transplants are an option for people with cystinosis. Cysteine crystals will not build up in the newly transplanted kidney, although they may still affect other organs of the body.

What is the prognosis for a person with Cystinosis?

Cystagon has extended the lifespan of people with cystinosis, but exact lifespan is not known. Some people with the disease have lived into their 50s.

Methods and Limitations

CARSON KUNZ [Foresight Carrier Screen]: Sequencing with copy number analysis (v4.0), spinal muscular atrophy (v4.0), analysis of homologous regions (v4.0), and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v4.0) (Assay(s): DTS v4.0).
BROOKLYN KUNZ [Foresight Carrier Screen]: Sequencing with copy number analysis (v3.1), triplet repeat detection, spinal muscular atrophy (v3.0), analysis of homologous regions (v3.2), and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v3.2) (Assay(s): DTS v3.2, fragile x).

Sequencing with copy number analysis (v3.1)

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.

Sequencing with copy number analysis (v4.0)

Hybridization capture-based target enrichment, 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 rare instances where genomic features (e.g., homopolymers) or other variables compromise calling fidelity, the affected regions 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 types of homology impede reliable variant detection may be assayed using alternate technology or may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with a sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel with a sensitivity of ≥75%. Selected founder deletions may be detected at higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from tiled regions and, when available, using junction reads. Only exons included in the region affected by the copy number variant (CNV) are included 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.

Triplet repeat detection

Polymerase chain reaction (PCR) with fluorescently labeled primers is used to amplify the CGG repeat region in the 5' UTR of *FMR1* (NM_002024.4: c.1-131CGG[1_n]), and PCR products are sized using capillary electrophoresis. Reported sizes are accurate to ±1 repeat for normal or intermediate alleles and ± two repeats for premutation alleles. Alleles above 200 CGG repeats (full mutations), while identified, are not specifically sized and will be reported as ">200" CGG repeats. In an unknown number of cases, other genetic variation may interfere with CGG repeat analysis. Other *FMR1* pathogenic variation will not be detected. *FMR1* promoter methylation is not analyzed. Allele size mosaicism may not be detected, as the test has been calibrated to yield results that are equivalent to the results from Southern blot. Opt-in testing of *FMR1* AGG interruptions is available for results showing between 50 and 54 CGG repeats. Automatic reflex testing of AGG interruptions is performed for results showing between 55 and 90 CGG repeats. AGG interruption analysis is performed by a reference laboratory, and methods are provided in the appended report, when applicable. This assay is designed to detect germline (constitutional) variation of the CGG repeat in the 5' UTR of *FMR1*; gonadal mosaicism will not be detected. Results assume a normal karyotype. Sex chromosome variations and aneuploidies may affect the accuracy of this assay.

Spinal muscular atrophy (v3.0)

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*.

Spinal muscular atrophy (v4.0)

Targeted copy number analysis via hybridization capture-based target enrichment and high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. In an unknown number of cases, other genetic variation 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 apparent copies of *SMN1*. Further, individuals who are negative for the g.27134T>G SNP, and who are reported as having two copies of the *SMN1* gene may have additional *SMN1* gene copies. If additional unreported *SMN1* copies are present, the reported residual risk for these individuals may be overestimated. Other rare carrier states, where complex exchanges exist between gene copies or chromosomes, may not be detected by the assay.

Analysis of homologous regions (v3.2)

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.

Analysis of homologous regions (v4.0)

Hybridization capture-based target enrichment, high-throughput sequencing, targeted genotyping, and read-depth-based copy number analysis are used to determine the number of functional gene copies and/or the presence of selected variants in genes that have significant 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 congenital adrenal hyperplasia (CAH), depending on the chromosomal location of the variants (i.e., 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 rare carrier states, where complementary changes exist between gene copies or 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 estimates on the report are based 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 (v3.2)

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.

Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v4.0)

Hybridization capture-based target enrichment, high-throughput sequencing, and 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 exons listed in the assay specifications plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., 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 rare carrier states, where complementary changes exist between gene copies or chromosomes, may not be detected by the assay.

Interpretation of reported variants (v3.2)

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.

Interpretation of reported variants (v4.0)

The interpretation and classification of variants reflect the current state of Myriad's scientific understanding based on information available at the time of variant assessment. 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, likely 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 over time 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 additional individuals. 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 a report is updated or re-issued for other reasons, the variants reported may change based on their classification at the time of re-issue. 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 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 reside on the same chromosome or different chromosomes (i.e., phase). This assay is not validated to detect sex chromosome copy number variations; however, limited sex chromosome analysis is performed for quality control purposes. If present, sex chromosome variations including copy number variations, aneuploidies, aneusomies, rearrangements, or other structural changes may significantly reduce test sensitivity and accuracy of risk estimates. Variant interpretation and residual and reproductive risk estimates assume a normal karyotype. The Foresight™ Carrier Screen reports carrier status for only genes/phenotypes specified by the ordering healthcare provider. Other heritable and non-heritable conditions and defects exist that are not addressed by this test. Furthermore, not all forms of genetic variation are detected by this assay (e.g., 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 genes or 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.

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.

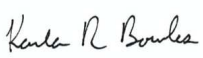
If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

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 John Alexander, PhD, FACMG, CGMB on Sep 25, 2023

Conditions Tested

6-pyruvoyl-tetrahydropterin Synthase Deficiency - **Gene:** PTS. Autosomal Recessive. **Exons:** NM_000317:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Adenosine Deaminase Deficiency - **Gene:** ADA. Autosomal Recessive. **Exons:** NM_000022:1-12. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Alpha Thalassemia, HBA1/HBA2-related - **Genes:** HBA1, HBA2. Autosomal Recessive. **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:** Not calculated due to rarity of disease in this individual's reported ethnicity.
Carson (Male): Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v4.0).
Brooklyn (Female): Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (v3.2).

Alpha-mannosidosis - **Gene:** MAN2B1. Autosomal Recessive. **Exons:** NM_000528:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Alpha-sarcoglycanopathy - **Gene:** SGCA. Autosomal Recessive. **Exons:** NM_000023:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Alstrom Syndrome - **Gene:** ALMS1. Autosomal Recessive. **Exons:** NM_015120:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Andermann Syndrome - **Gene:** SLC12A6. Autosomal Recessive. **Exons:** NM_133647:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Argininemia - **Gene:** ARG1. Autosomal Recessive. **Exons:** NM_000045:1-8. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Argininosuccinic Aciduria - **Gene:** ASL. Autosomal Recessive. **Exons:** NM_001024943:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Aspartylglucosaminuria - **Gene:** AGA. Autosomal Recessive. **Exons:** NM_000027:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Ataxia with Vitamin E Deficiency - **Gene:** TTPA. Autosomal Recessive. **Exons:** NM_000370:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Ataxia-telangiectasia - **Gene:** ATM. Autosomal Recessive. **Exons:** NM_000051:2-63. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. **Exons:** NM_000052:2-23. **Detection Rate:** Mixed or Other Caucasian 90%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Autoimmune Polyglandular Syndrome Type 1 - **Gene:** AIRE. Autosomal Recessive. **Exons:** NM_000383:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. **Exons:** NM_006019:2-20. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Autosomal Recessive Polycystic Kidney Disease, PKHD1-related - **Gene:** PKHD1. Autosomal Recessive. **Exons:** NM_138694:2-67. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - **Gene:** SACS. Autosomal Recessive. **Exons:** NM_014363:2-10. **Detection Rate:** Mixed or Other Caucasian 99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Bardet-Biedl Syndrome, BBS1-related - **Gene:** BBS1. Autosomal Recessive. **Exons:** NM_024649:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Bardet-Biedl Syndrome, BBS10-related - **Gene:** BBS10. Autosomal Recessive. **Exons:** NM_024685:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Bardet-Biedl Syndrome, BBS12-related - **Gene:** BBS12. Autosomal Recessive. **Exon:** NM_152618:2. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Bardet-Biedl Syndrome, BBS2-related - **Gene:** BBS2. Autosomal Recessive. **Exons:** NM_031885:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

BCS1L-related Disorders - **Gene:** BCS1L. Autosomal Recessive. **Exons:** NM_004328:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Beta-sarcoglycanopathy - **Gene:** SGCB. Autosomal Recessive. **Exons:** NM_000232:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Biotinidase Deficiency - **Gene:** BTD. Autosomal Recessive. **Exons:** NM_000060:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Bloom Syndrome - **Gene:** BLM. Autosomal Recessive. **Exons:** NM_000057:2-22. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Calpainopathy - **Gene:** CAPN3. Autosomal Recessive. **Exons:** NM_000070:1-24. **Detection Rate:** Mixed or Other Caucasian 99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Canavan Disease - **Gene:** ASPA. Autosomal Recessive. **Exons:** NM_000049:1-6. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Carbamoylphosphate Synthetase I Deficiency - **Gene:** CPS1. Autosomal Recessive. **Exons:** NM_001875:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Carnitine Palmitoyltransferase IA Deficiency - **Gene:** CPT1A. Autosomal Recessive. **Exons:** NM_001876:2-19. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Carnitine Palmitoyltransferase II Deficiency - **Gene:** CPT2. Autosomal Recessive. **Exons:** NM_000098:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Cartilage-hair Hypoplasia - **Gene:** RMRP. Autosomal Recessive. **Exon:** NR_003051:1. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Cerebrotendinous Xanthomatosis - **Gene:** CYP27A1. Autosomal Recessive. **Exons:** NM_000784:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Citrullinemia Type 1 - **Gene:** ASS1. Autosomal Recessive. **Exons:** NM_000050:3-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

CLN3-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN3. Autosomal Recessive. **Exons:** NM_001042432:2-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

CLN5-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN5. Autosomal Recessive. **Exons:** NM_006493:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

CLN8-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN8. Autosomal Recessive. **Exons:** NM_018941:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. **Exons:** NM_017890:2-62. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

COL4A3-related Alport Syndrome - **Gene:** COL4A3. Autosomal Recessive. **Exons:** NM_000091:1-52. **Detection Rate:** Mixed or Other Caucasian 94%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

COL4A4-related Alport Syndrome - **Gene:** COL4A4. Autosomal Recessive. **Exons:** NM_000092:2-48. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Combined Pituitary Hormone Deficiency, PROP1-related - **Gene:** PROP1. Autosomal Recessive. **Exons:** NM_006261:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Congenital Adrenal Hyperplasia, CYP11B1-related - **Gene:** CYP11B1. Autosomal Recessive. **Exons:** NM_000497:1-9. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Congenital Adrenal Hyperplasia, CYP21A2-related - **Gene:** CYP21A2. Autosomal Recessive. **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:** Mixed or Other Caucasian 96%.
Carson (Male): Analysis of homologous regions (v4.0).
Brooklyn (Female): Analysis of homologous regions (v3.2).

Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. Autosomal Recessive. **Exons:** NM_000303:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Congenital Disorder of Glycosylation Type Ic - **Gene:** ALG6. Autosomal Recessive. **Exons:** NM_013339:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Congenital Disorder of Glycosylation, MPI-related - **Gene:** MPI. Autosomal Recessive. **Exons:** NM_002435:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Costeff Optic Atrophy Syndrome - **Gene:** OPA3. Autosomal Recessive. **Exons:** NM_025136:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Cystic Fibrosis - **Gene:** CFTR. Autosomal Recessive. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Cystinosis - **Gene:** CTNS. Autosomal Recessive. **Exons:** NM_004937:3-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

D-bifunctional Protein Deficiency - **Gene:** HSD17B4. Autosomal Recessive. **Exons:** NM_000414:1-24. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Delta-sarcoglycanopathy - **Gene:** SGCD. Autosomal Recessive. **Exons:** NM_000337:2-9. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Dihydrolipoamide Dehydrogenase Deficiency - **Gene:** DLD. Autosomal Recessive. **Exons:** NM_000108:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Dysferlinopathy - **Gene:** DYSF. Autosomal Recessive. **Exons:** NM_003494:1-55. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - **Gene:** DMD. X-linked Recessive. **Exons:** NM_004006:1-79. **Detection Rate:** Mixed or Other Caucasian 99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

ERCC6-related Disorders - **Gene:** ERCC6. Autosomal Recessive. **Exons:** NM_000124:2-21. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

ERCC8-related Disorders - **Gene:** ERCC8. Autosomal Recessive. **Exons:** NM_000082:1-12. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

EVC-related Ellis-van Creveld Syndrome - **Gene:** EVC. Autosomal Recessive. **Exons:** NM_153717:1-21. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

EVC2-related Ellis-van Creveld Syndrome - **Gene:** EVC2. Autosomal Recessive. **Exons:** NM_147127:1-22. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Fabry Disease - **Gene:** GLA. X-linked Recessive. **Exons:** NM_000169:1-7. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Familial Dysautonomia - **Gene:** ELP1. Autosomal Recessive. **Exons:** NM_003640:2-37. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Familial Hyperinsulinism, ABCC8-related - **Gene:** ABCC8. Autosomal Recessive. **Exons:** NM_000352:1-39. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Familial Hyperinsulinism, KCNJ11-related - **Gene:** KCNJ11. Autosomal Recessive. **Exon:** NM_000525:1. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Familial Mediterranean Fever - **Gene:** MEFV. Autosomal Recessive. **Exons:** NM_000243:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Fanconi Anemia Complementation Group A - **Gene:** FANCA. Autosomal Recessive. **Exons:** NM_000135:1-43. **Detection Rate:** Mixed or Other Caucasian 92%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Fanconi Anemia, FANCC-related - **Gene:** FANCC. Autosomal Recessive. **Exons:** NM_000136:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

FKRP-related Disorders - **Gene:** FKRP. Autosomal Recessive. **Exon:** NM_024301:4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

FKTN-related Disorders - **Gene:** FKTN. Autosomal Recessive. **Exons:** NM_001079802:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Fragile X Syndrome - **Gene:** FMR1. X-linked Dominant. **Variant (1):** FMR1 CGG repeat number. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Not tested.
Brooklyn (Female): Triplet repeat detection.

Free Sialic Acid Storage Disorders - **Gene:** SLC17A5. Autosomal Recessive. **Exons:** NM_012434:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Galactokinase Deficiency - **Gene:** GALK1. Autosomal Recessive. **Exons:** NM_000154:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Galactosemia - **Gene:** GALT. Autosomal Recessive. **Exons:** NM_000155:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Gamma-sarcoglycanopathy - **Gene:** SGCG. Autosomal Recessive. **Exons:** NM_000231:2-8. **Detection Rate:** Mixed or Other Caucasian 87%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Gaucher Disease - **Gene:** GBA1. Autosomal Recessive. **Variants (10):** D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. **Detection Rate:** Mixed or Other Caucasian 60%.
Carson (Male): Analysis of homologous regions (v4.0).
Brooklyn (Female): Analysis of homologous regions (v3.2).

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - **Gene:** GJB2. Autosomal Recessive. **Exons:** NM_004004:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

GLB1-related Disorders - **Gene:** GLB1. Autosomal Recessive. **Exons:** NM_000404:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glutaric Acidemia, GCDH-related - **Gene:** GCDH. Autosomal Recessive. **Exons:** NM_000159:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glycine Encephalopathy, AMT-related - **Gene:** AMT. Autosomal Recessive. **Exons:** NM_000481:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glycine Encephalopathy, GLDC-related - **Gene:** GLDC. Autosomal Recessive. **Exons:** NM_000170:1-25. **Detection Rate:** Mixed or Other Caucasian 94%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glycogen Storage Disease Type Ia - **Gene:** G6PC1. Autosomal Recessive. **Exons:** NM_000151:1-5. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. Autosomal Recessive. **Exons:** NM_001164277:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Glycogen Storage Disease Type III - **Gene:** AGL. Autosomal Recessive. **Exons:** NM_000642:2-34. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

GNE Myopathy - **Gene:** GNE. Autosomal Recessive. **Exons:** NM_001128227:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

GNPTAB-related Disorders - **Gene:** GNPTAB. Autosomal Recessive. **Exons:** NM_024312:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

HADHA-related Disorders - **Gene:** HADHA. Autosomal Recessive. **Exons:** NM_000182:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. Autosomal Recessive. **Exons:** NM_000518:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Hereditary Fructose Intolerance - **Gene:** ALDOB. Autosomal Recessive. **Exons:** NM_000035:2-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. Autosomal Recessive. **Exons:** NM_000520:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

HMG-CoA Lyase Deficiency - **Gene:** HMGCL. Autosomal Recessive. **Exons:** NM_000191:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Holocarboxylase Synthetase Deficiency - **Gene:** HLCS. Autosomal Recessive. **Exons:** NM_000411:4-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Homocystinuria, CBS-related - **Gene:** CBS. Autosomal Recessive. **Exons:** NM_000071:3-17. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Hydrolethalus Syndrome - **Gene:** HYLS1. Autosomal Recessive. **Exon:** NM_145014:4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Hypophosphatasia - **Gene:** ALPL. Autosomal Recessive. **Exons:** NM_000478:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Isovaleric Acidemia - **Gene:** IVD. Autosomal Recessive. **Exons:** NM_002225:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Joubert Syndrome 2 - **Gene:** TMEM216. Autosomal Recessive. **Exons:** NM_001173990:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Junctional Epidermolysis Bullosa, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. **Exons:** NM_000227:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. **Exons:** NM_000228:2-23. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. **Exons:** NM_005562:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Krabbe Disease - **Gene:** GALC. Autosomal Recessive. **Exons:** NM_000153:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Leigh Syndrome, French-Canadian Type - **Gene:** LRPPRC. Autosomal Recessive. **Exons:** NM_133259:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Lipoid Congenital Adrenal Hyperplasia - **Gene:** STAR. Autosomal Recessive. **Exons:** NM_000349:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Lysosomal Acid Lipase Deficiency - **Gene:** LIPA. Autosomal Recessive. **Exons:** NM_000235:2-10. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Maple Syrup Urine Disease Type Ia - **Gene:** BCKDHA. Autosomal Recessive. **Exons:** NM_000709:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Maple Syrup Urine Disease Type Ib - **Gene:** BCKDHB. Autosomal Recessive. **Exons:** NM_183050:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Maple Syrup Urine Disease Type II - **Gene:** DBT. Autosomal Recessive. **Exons:** NM_001918:1-11. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. Autosomal Recessive. **Exons:** NM_000016:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Megalencephalic Leukoencephalopathy with Subcortical Cysts - **Gene:** MLC1. Autosomal Recessive. **Exons:** NM_015166:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Metachromatic Leukodystrophy - **Gene:** ARSA. Autosomal Recessive. **Exons:** NM_000487:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Methylmalonic Acidemia, cblA Type - **Gene:** MMAA. Autosomal Recessive. **Exons:** NM_172250:2-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Methylmalonic Acidemia, cblB Type - **Gene:** MMAB. Autosomal Recessive. **Exons:** NM_052845:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Methylmalonic Acidemia, MMUT-related - **Gene:** MMUT. Autosomal Recessive. **Exons:** NM_000255:2-13. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Methylmalonic Aciduria and Homocystinuria, cblC Type - **Gene:** MMACHC. Autosomal Recessive. **Exons:** NM_015506:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

MKS1-related Disorders - **Gene:** MKS1. Autosomal Recessive. **Exons:** NM_017777:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucolipidosis III Gamma - **Gene:** GNPTG. Autosomal Recessive. **Exons:** NM_032520:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucolipidosis IV - **Gene:** MCOLN1. Autosomal Recessive. **Exons:** NM_020533:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucopolysaccharidosis Type I - **Gene:** IDUA. Autosomal Recessive. **Exons:** NM_000203:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucopolysaccharidosis Type II - **Gene:** IDS. X-linked Recessive. **Exons:** NM_000202:1-9. **Detection Rate:** Mixed or Other Caucasian 89%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucopolysaccharidosis Type IIIA - **Gene:** SGSH. Autosomal Recessive. **Exons:** NM_000199:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucopolysaccharidosis Type IIIB - **Gene:** NAGLU. Autosomal Recessive. **Exons:** NM_000263:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Mucopolysaccharidosis Type IIIC - **Gene:** HGSNAT. Autosomal Recessive. **Exons:** NM_152419:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Muscular Dystrophy, LAMA2-related - **Gene:** LAMA2. Autosomal Recessive. **Exons:** NM_000426:1-43,45-65. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

MYO7A-related Disorders - **Gene:** MYO7A. Autosomal Recessive. **Exons:** NM_000260:2-49. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

NEB-related Nemaline Myopathy - **Gene:** NEB. Autosomal Recessive. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** Mixed or Other Caucasian 92%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Nephrotic Syndrome, NPHS1-related - **Gene:** NPHS1. Autosomal Recessive. **Exons:** NM_004646:1-29. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Nephrotic Syndrome, NPHS2-related - **Gene:** NPHS2. Autosomal Recessive. **Exons:** NM_014625:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Neuronal Ceroid Lipofuscinosis, CLN6-related - **Gene:** CLN6. Autosomal Recessive. **Exons:** NM_017882:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Niemann-Pick Disease Type C1 - **Gene:** NPC1. Autosomal Recessive. **Exons:** NM_000271:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Niemann-Pick Disease Type C2 - **Gene:** NPC2. Autosomal Recessive. **Exons:** NM_006432:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Niemann-Pick Disease, SMPD1-related - **Gene:** SMPD1. Autosomal Recessive. **Exons:** NM_000543:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Nijmegen Breakage Syndrome - **Gene:** NBN. Autosomal Recessive. **Exons:** NM_002485:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Ornithine Transcarbamylase Deficiency - **Gene:** OTC. X-linked Recessive. **Exons:** NM_000531:1-10. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

PCCA-related Propionic Acidemia - **Gene:** PCCA. Autosomal Recessive. **Exons:** NM_000282:1-24. **Detection Rate:** Mixed or Other Caucasian 95%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

PCCB-related Propionic Acidemia - **Gene:** PCCB. Autosomal Recessive. **Exons:** NM_000532:1-15. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

PCDH15-related Disorders - **Gene:** PCDH15. Autosomal Recessive. **Exons:** NM_033056:2-33. **Detection Rate:** Mixed or Other Caucasian 93%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Pendred Syndrome - **Gene:** SLC26A4. Autosomal Recessive. **Exons:** NM_000441:2-21. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Peroxisome Biogenesis Disorder Type 1 - **Gene:** PEX1. Autosomal Recessive. **Exons:** NM_000466:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Peroxisome Biogenesis Disorder Type 3 - **Gene:** PEX12. Autosomal Recessive. **Exons:** NM_000286:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Peroxisome Biogenesis Disorder Type 4 - **Gene:** PEX6. Autosomal Recessive. **Exons:** NM_000287:1-17. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Peroxisome Biogenesis Disorder Type 5 - **Gene:** PEX2. Autosomal Recessive. **Exon:** NM_000318:4. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Peroxisome Biogenesis Disorder Type 6 - **Gene:** PEX10. Autosomal Recessive. **Exons:** NM_153818:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. Autosomal Recessive. **Exons:** NM_000277:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

POMGNT-related Disorders - **Gene:** POMGNT1. Autosomal Recessive. **Exons:** NM_017739:2-22. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Pompe Disease - **Gene:** GAA. Autosomal Recessive. **Exons:** NM_000152:2-20. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

PPT1-related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. Autosomal Recessive. **Exons:** NM_000310:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Primary Carnitine Deficiency - **Gene:** SLC22A5. Autosomal Recessive. **Exons:** NM_003060:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Primary Hyperoxaluria Type 1 - **Gene:** AGXT. Autosomal Recessive. **Exons:** NM_000030:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. Autosomal Recessive. **Exons:** NM_012203:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Primary Hyperoxaluria Type 3 - **Gene:** HOGA1. Autosomal Recessive. **Exons:** NM_138413:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Pycnodysostosis - **Gene:** CTSK. Autosomal Recessive. **Exons:** NM_000396:2-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Pyruvate Carboxylase Deficiency - **Gene:** PC. Autosomal Recessive. **Exons:** NM_000920:3-22. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. **Exons:** NM_000288:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

RTEL1-related Disorders - **Gene:** RTEL1. Autosomal Recessive. **Exons:** NM_032957:2-35. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. **Exons:** NM_000521:1-14. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Short-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. Autosomal Recessive. **Exons:** NM_000017:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. Autosomal Recessive. **Exons:** NM_000382:1-10. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

SLC26A2-related Disorders - **Gene:** SLC26A2. Autosomal Recessive. **Exons:** NM_000112:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. Autosomal Recessive. **Exons:** NM_001360:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Spastic Paraplegia Type 15 - **Gene:** ZFYVE26. Autosomal Recessive. **Exons:** NM_015346:2-42. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Spinal Muscular Atrophy - **Gene:** SMN1. Autosomal Recessive. **Variant (1):** SMN1 copy number. **Detection Rate:** Mixed or Other Caucasian 95%.
Carson (Male): Spinal muscular atrophy (v4.0).
Brooklyn (Female): Spinal muscular atrophy (v3.0).

Spondylothoracic Dysostosis - **Gene:** MESP2. Autosomal Recessive. **Exons:** NM_001039958:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

TGM1-related Autosomal Recessive Congenital Ichthyosis - **Gene:** TGM1. Autosomal Recessive. **Exons:** NM_000359:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

TPP1-related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. Autosomal Recessive. **Exons:** NM_000391:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Tyrosine Hydroxylase Deficiency - **Gene:** TH. Autosomal Recessive. **Exons:** NM_199292:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Tyrosinemia Type I - **Gene:** FAH. Autosomal Recessive. **Exons:** NM_000137:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. **Exons:** NM_000353:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

USH1C-related Disorders - **Gene:** USH1C. Autosomal Recessive. **Exons:** NM_005709:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

USH2A-related Disorders - **Gene:** USH2A. Autosomal Recessive. **Exons:** NM_206933:2-72. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Usher Syndrome Type 3 - **Gene:** CLRN1. Autosomal Recessive. **Exons:** NM_174878:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Very-long-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. Autosomal Recessive. **Exons:** NM_000018:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. **Exons:** NM_000053:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Adrenal Hypoplasia Congenita - **Gene:** NR0B1. X-linked Recessive. **Exons:** NM_000475:1-2. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Adrenoleukodystrophy - **Gene:** ABCD1. X-linked Recessive. **Exons:** NM_000033:1-6. **Detection Rate:** Mixed or Other Caucasian 77%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Alport Syndrome - **Gene:** COL4A5. X-linked Recessive. **Exons:** NM_000495:1-51. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Juvenile Retinoschisis - **Gene:** RS1. X-linked Recessive. **Exons:** NM_000330:1-6. **Detection Rate:** Mixed or Other Caucasian 98%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Myotubular Myopathy - **Gene:** MTM1. X-linked Recessive. **Exons:** NM_000252:2-15. **Detection Rate:** Mixed or Other Caucasian 96%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

X-linked Severe Combined Immunodeficiency - **Gene:** IL2RG. X-linked Recessive. **Exons:** NM_000206:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Xeroderma Pigmentosum Group A - **Gene:** XPA. Autosomal Recessive. **Exons:** NM_000380:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

Xeroderma Pigmentosum Group C - **Gene:** XPC. Autosomal Recessive. **Exons:** NM_004628:1-16. **Detection Rate:** Mixed or Other Caucasian 97%.
Carson (Male): Sequencing with copy number analysis (v4.0).
Brooklyn (Female): Sequencing with copy number analysis (v3.1).

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.

Disease	CARSON KUNZ Residual Risk	BROOKLYN KUNZ Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	Alpha globin status: aa/aa.	Alpha globin status: aa/aa.	Low
Alpha-mannosidosis	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 15,000	1 in 15,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 660,000	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 32,000	1 in 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 13,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 14,000	1 in 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 3,400	1 in 3,400	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related	1 in 8,400	1 in 8,400	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote †	1 in 1,300	1 in 5,100
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000

RESULTS RECIPIENT
UND CENTER FOR FAMILY MEDICINE
Attn: Jacqueline Quisno
NPI: 1326016163
Report Date: 09/25/2023

MALE
CARSON KUNZ
DOB: 07/01/1990
Ethnicity: Mixed or Other Caucasian
Barcode: 11004513356767

FEMALE
BROOKLYN KUNZ
DOB: 01/06/1993
Ethnicity: Mixed or Other Caucasian
Barcode: 11004513358240

Disease	CARSON KUNZ Residual Risk	BROOKLYN KUNZ Residual Risk	Reproductive Risk
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 3,000	1 in 3,000	< 1 in 1,000,000
Cystinosis	1 in 22,000	57 kb deletion heterozygote †	1 in 89,000
D-bifunctional Protein Deficiency	1 in 9,000	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 13,000	< 1 in 13,000	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,500	1 in 8,500	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 16,000	< 1 in 16,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Fragile X Syndrome	Not tested	Normal: 30 and 30 repeats	Not calculated
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 37,000	1 in 37,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,300	1 in 3,300	< 1 in 1,000,000
Gaucher Disease	1 in 260	1 in 260	1 in 280,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 2,500	< 1 in 1,000,000
GLB1-related Disorders	1 in 17,000	1 in 17,000	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	1 in 26,000	< 1 in 1,000,000
Glycine Encephalopathy, GLDC-related	1 in 2,500	1 in 2,500	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 8,700	1 in 8,700	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	1 in 23,000	1 in 23,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 3,700	1 in 3,700	< 1 in 1,000,000
Hereditary Fructose Intolerance	1 in 7,900	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	1 in 9,400	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 30,000	1 in 30,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	1 in 32,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 32,000	1 in 32,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	1 in 14,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 14,000	1 in 14,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 39,000	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 4,400	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000

myriad WOMEN'S HEALTH

RESULTS RECIPIENT
UND CENTER FOR FAMILY MEDICINE
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NPI: 1326016163
Report Date: 09/25/2023

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FEMALE
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DOB: 01/06/1993
Ethnicity: Mixed or Other Caucasian
Barcode: 11004513358240

Disease	CARSON KUNZ Residual Risk	BROOKLYN KUNZ Residual Risk	Reproductive Risk
Metachromatic Leukodystrophy	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	1 in 48,000	1 in 48,000	< 1 in 1,000,000
Methylmalonic Acidemia, MMUT-related	1 in 26,000	1 in 26,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis III Gamma	< 1 in 20,000	< 1 in 20,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	< 1 in 1,000,000	1 in 670,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIA	1 in 19,000	1 in 19,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 27,000	1 in 27,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	1 in 5,700	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 1,200	< 1 in 1,000,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	1 in 20,000	1 in 20,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 19,000	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
PCCA-related Propionic Acidemia	1 in 4,200	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 8,200	1 in 8,200	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 4,800	1 in 4,800	< 1 in 1,000,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 4,000	1 in 4,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 17,000	1 in 17,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	1 in 13,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	1 in 18,000	1 in 18,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 9,400	1 in 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	< 1 in 1,000,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 4,100	1 in 4,100	< 1 in 1,000,000

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Disease	CARSON KUNZ Residual Risk	BROOKLYN KUNZ Residual Risk	Reproductive Risk
Usher Syndrome Type 3	1 in 41,000	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	1 in 18,000	< 1 in 1,000,000
Wilson Disease	1 in 6,500	1 in 6,500	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 45,000	1 in 180,000
X-linked Alport Syndrome	Not calculated	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 670,000	< 1 in 1,000,000
X-linked Myotubular Myopathy	Not calculated	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	1 in 7,300	< 1 in 1,000,000