Patient:
DOE, JANE

Accession: NXGMDX-999999
Created: January 01, 20XX
Patient Gender: Female
Date Of Birth: January 01, 19XX
Specimen Type: Whole blood
Collection Date: January 01, 20XX **Received Date:** January 02, 20XX

Receiving Physicians:
Dr. Smith

Receiving Facilities: SAMPLE REPORT

RESULTS SUMMARY

SUPER PANEL 145

Positive

- Usher syndrome type IIA/USH2A-related disorders

Positive carrier status for any conditions included on the ordered panel are detailed in the report. Sequence and/or copy number analysis did not identify any other known pathogenic variants for the remaining genetic conditions included on this panel. Please see the additional pages for information on the patient's results as well as the list of the genes and corresponding disease included on this test panel.

POSITIVE TEST INTERPRETATION

Super Panel 145

Usher syndrome type IIA/USH2A-related disorders - Positive

USH2A (NM_206933.4):c.5167G>C(p.Gly1723Arg), Pathogenic, Heterozygous
This individual is a carrier of a pathogenic *USH2A* gene variant, heterozygous for c.5167G>C (p.Gly1723Arg, G1723R). Risk for offspring is dependent upon the partner's carrier status.

Recommend consultation with a genetic counselor and testing of the patient's partner, if not already completed or underway.

PMID: 9536098, 17576681, 24944099, 25404053, 26667666

Signed by: SAMPLE REPORT
Signed on: SAMPLE REPORT
Accession #: NXGMDX-999999
Patient DOB: January 01, 19XX

801 Broadway NW, Suite# 203
Grand Rapids MI 49504
Phone: 855-77-NxGen
Fax: 616-710-4667

NEGATIVE TEST INTERPRETATION

Alpha Thalassemia
This individual possesses four intact alpha-globin genes, considered a normal copy number, and therefore is unlikely to be a carrier for Alpha Thalassemia. No alpha-globin gene rearrangements (deletions or duplications) or point mutations were detected.

Fragile X and FMR1 Related Disorders
This patient's *FMR1* gene alleles have 30 and 43 CGG repeats respectively. Based on these results, this individual is at low risk to have a child with Fragile X or an *FMR1*-related disorder.

Spinal Muscular Atrophy (SMA)
This individual possesses 2 copies of the survival motor neuron I (*SMN1*) gene, considered a normal copy number, and therefore is unlikely to be a carrier for Spinal Muscular Atrophy (SMA). Sequencing of *SMN1* exons 1-7 for pathogenic point mutations, as well as the silent carrier markers g.27134T>G and g.27706_27707delAT, was also performed and no variants were identified. This individual also possesses 2 copies of the survival motor neuron II (*SMN2*) gene. An increased number of *SMN2* copy numbers (3 or greater) can decrease the severity of SMA in affected individuals, however, *SMN2* copy number does not influence carrier status in the presence of one or more copies of *SMN1*.

801 Broadway NW, Suite# 203
Grand Rapids MI 49504
Phone: 855-77-NxGen
Fax: 616-710-4667

METHODS AND LIMITATIONS

Super Panel 145

DNA is isolated from the sample. Exons, untranslated regions, and 25 flanking intronic bases of interest in the *ABCC8, ABCD1, ACADM, ACADS, ACADVL, ACAT1, ADA, ADAMTS2, AGA, AGL, AGXT, AIRE, ALDH3A2, ALDOB, ALPL, AMT, ARSA, ASL, ASPA, ASS1, ATM, ATP7B, BBS1, BBS10, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BTD, CAPN3, CBS, CDH23, CFTR, CHM, CLN3, CLN5, CLN8, CLRN1, CNGB3, COL4A4, CPT1A, CPT2, CTNS, CTSK, DHCR7, DHDDS, DLD, DPYD, ELP1, F11, FAH, FANCC, FKTN, FOXI1, G6PC, G6PD, GAA, GALC, GALK1, GALT, GBA, GBE1, GCDH, GLDC, GJB2, GJB3, GJB6, GNE, GRHPR, HADHA, HBA1/HBA2, HBB, HEXA, HGD, HLCS, HMGCL, HSD17B4, IDUA, IVD, KCNJ10, LAMA3, LAMB3, LAMC2, LIPA, MAN2B1, MCCC1, MCCC2, MCOLN1, MEFV, MKKS, MLC1, MMAA, MMAB, MMACHC, MMADHC, MPI, MPL, MTRR, MTTP, MYO7A, NBN, NEB, NOLA3, NPC1, NPHS1, NPHS2, NR0B1, OPA3, PAH, PCCA, PCCB, PCDH15, PEX1, PEX7, PHGDH, PKHD1, PMM2, POMGNT1, PPT1, PROP1, PYGM, RMRP, RS1, SACS, SGCA, SGCB, SLC12A6, SLC17A5, SLC22A5, SLC25A13, SLC26A2, SLC26A4, SLC35A3, SLC37A4, SMN1, SMN2, SMPD1, STAR, SUMF1, TAT, TH, TMEM216, TPP1, TTPA, USH2A, and VPS13B* genes are amplified enzymatically and subjected to Next Generation DNA Sequencing (NGS) on an Ion Torrent Instrument at NxGen MDx. Due to the existence of paralogs, repetitive sequence, or high GC content, *CLN3, HBA1, HBA2, NEB, SMN1*, and *SMN2* do not have the same coverage as most of the genes included in this panel; however, our residual risk reflects the actual risk based on coverage and additional assays described below are performed to increase the sensitivity beyond what is feasible by NGS alone. More than 99% of all bases in the coverage areas are sequenced at greater than the 20X minimum read depth. Sanger sequencing is performed to confirm suspect variant calls.

The DNA sequences were assembled and aligned against reference gene sequences based on the human genome build GRCh37/UCSC hg 19 and analyzed for sequence variants using Torrent Suite software (ThermoFisher). Sequence variants are annotated and classified in the context of ACMG-AMP guidelines based on review of resources including ClinVar, the CFTR2 Database, population allele frequency data, in silico predictive tools, as well as relevant clinical and functional studies in the literature. Large deletions are not detected by this assay. *CFTR* intron 9 poly T tract genotyping is reported when R117H is present. *CFTR* intron 9 TG/T genotyping is reported by request only.

Alpha thalassemia deletion/duplication testing is also performed on DNA isolated from the sample. DNA from the sample is amplified enzymatically by multiplex ligation-dependent probe amplification (MLPA) and analyzed on a capillary electrophoresis instrument. This test was developed and its performance determined by NxGen MDx to identify gene dosage variants of the alpha globin gene cluster.

In addition to *SMN1* and *SMN2* sequencing as described above, *SMN1* and *SMN2* gene dosage are determined using Taqman quantitative PCR analysis. Variants in the *SMN2* gene are only reported when a single copy of *SMN1* is identified, as *SMN2* variants are not deleterious when more than one copy of *SMN1* is identified.

Gene specific *FMR1* PCR determines the total number of CGG and AGG triplet repeats in the 5' UTR. Sizing is accurate ±1 repeat. Premutation carriers between 55 and 90 CGG repeats are reflexively analyzed for AGG triplet repeats.

Disease susceptibility, pseudodeficiency alleles, benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Common *ACADS* disease susceptibility alleles G209S and R171W are not evaluated as part of this screening as presence of these variants in the absence of additional genetic and environmental factors cannot accurately determine risk for developing SCAD. Genetic carrier screening does not assess all inherited forms of intellectual disability, birth defects, and genetic disease. A family history of any of these conditions may warrant additional evaluation. Sanger confirmation studies are performed when both members of a reproductive couple are suspected carriers of the same condition.

This test has not been cleared or approved by the U.S. Food and Drug Administration. However, the FDA has determined that such a clearance or approval is not necessary. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing. This test was performed at NxGen MDx, located at 801 Broadway Suite 203, Grand Rapids, Michigan-49504. CLIA Number: 23D2059943.

Hemoglobinopathy evaluation is performed on the Capillarys 3 system by dilution, injection by aspiration, followed by high voltage protein separation and direct detection of the hemoglobin-protein fractions at 415 nm. The resultant electropherogram is evaluated for pattern abnormalities, and the individual peaks are quantified by Sebia Phoresis software. Only fractions that impact carrier status of diseases in this panel are reported.

Consultation with a genetics professional is recommended for interpretation of all results. The chance of false positive or false negative results cannot be completely excluded. Although DNA based testing provides highly accurate genotyping, rare diagnostic errors may occur. Examples include misinterpretation because of polymorphisms or rare genetic variants, blood transfusion, mislabeled specimens, amplicon drop outs, or erroneous representation of family relationships. Bone marrow transplants from allogeneic donors will interfere with testing.

NxGen MDx

Want to Tell Your Story?

Scan the QR code below to share your experience with us.

801 Broadway NW, Suite# 203
Grand Rapids MI 49504
Phone: 855-77-NxGen
Fax: 616-710-4667

RESIDUAL RISKS | Super Panel 145

NxGen MDx

Disease Name: *GENE*

3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) Lyase Deficiency: *HMGCL*
•**General Population:** 1/24950

3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, *MCCC1*-Related: *MCCC1*
•**General Population:** 1/494

3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, *MCCC2*-Related: *MCCC2*
•**General Population:** 1/784

3-Methylglutaconic Aciduria Type III (Costeff Optic Atrophy): *OPA3*
•**General Population:** 1/49900

Abetalipoproteinemia: *MTTP*
•**General Population:** 1/49900

Achromatopsia: *CNGB3*
•**General Population:** 1/9200

***ADA*-Related Conditions:** *ADA*
•**General Population:** 1/4323

Alkaptonuria: *HGD*
•**General Population:** 1/2490

Alpha-Thalassemia/Alpha-Globin Triplication: *HBA1/HBA2*
•**General Population:** 1/2400

Alpha-Mannosidosis: *MAN2B1*
•**General Population:** 1/35300

Alport Syndrome, *COL4A4*-Related: *COL4A4*
•**General Population:** 1/35200

Andermann Syndrome: *SLC12A6*
•**General Population:** 1/49900

Argininosuccinic Aciduria: *ASL*
•**General Population:** 1/12900

Arthrogryposis, Intellectual Disability, and Seizures (AMRS): *SLC35A3*
•**General Population:** 1/49900

Aspartylglucosaminuria: *AGA*
•**General Population:** 1/49900

Ataxia-Telangiectasia: *ATM*
•**General Population:** 1/4950

Ataxia with Vitamin E Deficiency: *TTPA*
•**General Population:** 1/4990

Autoimmune Polyendocrinopathy with Candidiasis and Ectodermal Dystrophy: *AIRE*
•**General Population:** 1/7450

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS): *SACS*
•**General Population:** 1/49900

Bardet-Biedl Syndrome, *BBS1*-Related: *BBS1*
•**General Population:** 1/32900

Bardet-Biedl Syndrome, *BBS10*-Related: *BBS10*
•**General Population:** 1/35300

Bardet-Biedl Syndrome, *MKKS*-Related: *MKKS*
•**General Population:** 1/49900

Beta-Ketothiolase Deficiency: *ACAT1*
•**General Population:** 1/2936

Beta-Thalassemia: *HBB*
•**General Population:** 1/960

Biotinidase Deficiency: *BTD*
•**General Population:** 1/12400

Bloom Syndrome: *BLM*
•**General Population:** 1/49900

Canavan Disease: *ASPA*
•**General Population:** 1/7900

Carnitine Palmitoyltransferase I Deficiency: *CPT1A*
•**General Population:** 1/4990

Carnitine Palmitoyltransferase II Deficiency: *CPT2*
•**General Population:** 1/3620

Cartilage-Hair Hypoplasia, Anauxetic Dysplasia Spectrum Disorders: *RMRP*
•**General Population:** 1/49900

Choroideremia: *CHM**
•**General Population:** Reduced

Citrin Deficiency: *SLC25A13*
•**General Population:** 1/7130

Citrullinemia Type 1: *ASS1*
•**General Population:** 1/2976

Cohen Syndrome: *VPS13B*
•**General Population:** 1/1849

Combined Pituitary Hormone Deficiency: *PROP1*
•**General Population:** 1/6100

Congenital Amegakaryocytic Thrombocytopenia: *MPL*
•**General Population:** 1/9980

Congenital Disorder of Glycosylation Type Ia: *PMM2*
•**General Population:** 1/18900

Congenital Disorder of Glycosylation Type Ib: *MPI*
•**General Population:** 1/49900

Cystic Fibrosis and Other *CFTR*-Related Disorders: *CFTR*
•**General Population:** 1/4400

Cystinosis: *CTNS*
•**General Population:** 1/15700

D-Bifunctional Protein Deficiency: *HSD17B4*
•**General Population:** 1/15700

***DHDDS*-Related Disorders (including Retinitis Pigmentosa 59):** *DHDDS*
•**General Population:** 1/49900

Dihydrolipoamide Dehydrogenase Deficiency: *DLD*
•**General Population:** 1/24950

Dihydropyrimidine Dehydrogenase Deficiency: *DPYD*
•**General Population:** 1/2120

Dyskeratosis Congenita, Autosomal Recessive: *NOLA3 (NOP10)*
•**General Population:** 1/24900

Ehlers-Danlos Syndrome, Dermatosparaxis Type: *ADAMTS2*
•**General Population:** 1/4159

Familial Dysautonomia: *ELP1*
•**General Population:** 1/49900

Familial Hyperinsulinism *ABCC8*-Related: *ABCC8*
•**General Population:** 1/5868

Familial Mediterranean Fever: *MEFV*
•**General Population:** 1/6300

Fanconi Anemia Type C: *FANCC*
•**General Population:** 1/5200

***FKTN*-Related Disorders (including Walker-Warburg Syndrome):** *FKTN*
•**General Population:** 1/49900

Fragile X Syndrome and *FMR1*-Related Disorders: *FMR1**
•**General Population:** Reduced

Galactokinase Deficiency: *GALK1*
•**General Population:** 1/12100

Galactosemia, *GALT*-Related: *GALT*
•**General Population:** 1/4950

* This condition is inherited in an X-linked manner.

This is a reference table for the risk calculations for all conditions tested when no pathogenic variant is found. Since negative results do not completely rule out the possibility of being a carrier, the residual risk represents the patient's post-test likelihood of being a carrier based on the general population carrier frequency

Gaucher Disease: *GBA*
•**General Population:** 1/15700

Glucose-6-Phosphate Dehydrogenase Deficiency: *G6PD**
•**General Population:** Reduced

Glutaric Acidemia Type 1: *GCDH*
•**General Population:** 1/2868

Glycine Encephalopathy, *AMT*-Related: *AMT*
•**General Population:** 1/32400

Glycine Encephalopathy, *GLDC*-Related: *GLDC*
•**General Population:** 1/2734

Glycogen Storage Disease Type IA: *G6PC*
•**General Population:** 1/3580

Glycogen Storage Disease Type IB: *SLC37A4*
•**General Population:** 1/17450

Glycogen Storage Disease Type II (Pompe Disease): *GAA*
•**General Population:** 1/1415

Glycogen Storage Disease Type III: *AGL*
•**General Population:** 1/3160

Glycogen Storage Disease Type IV/Adult Polyglucosan Body Disease: *GBE1*
•**General Population:** 1/5515

Glycogen Storage Disease Type V: *PYGM*
•**General Population:** 1/15700

GRACILE Syndrome/*BCS1L*-Related Disorders: *BCS1L*
•**General Population:** 1/49900

Hemophilia C/Factor XI Deficiency: *F11*
•**General Population:** 1/49900

Hereditary Fructose Intolerance: *ALDOB*
•**General Population:** 1/12100

Holocarboxylase Synthetase Deficiency: *HLCS*
•**General Population:** 1/22300

Homocystinuria Due to Cystathionine Beta-Synthase Deficiency: *CBS*
•**General Population:** 1/4460

Homocystinuria, Cobalamin E Type: *MTRR*
•**General Population:** 1/9980

Hypophosphatasia: *ALPL*
•**General Population:** 1/2980

Inclusion Body Myopathy 2: *GNE*
•**General Population:** 1/890

Isovaleric Acidemia: *IVD*
•**General Population:** 1/24900

Joubert Syndrome 2/*TMEM216*-Related Disorders: *TMEM216*
•**General Population:** 1/49900

Junctional Epidermolysis Bullosa, *LAMA3*-Related: *LAMA3*
•**General Population:** 1/24950

Junctional Epidermolysis Bullosa, *LAMB3*-Related: *LAMB3*
•**General Population:** 1/31600

Junctional Epidermolysis Bullosa, *LAMC2*-Related: *LAMC2*
•**General Population:** 1/8318

Krabbe Disease: *GALC*
•**General Population:** 1/350

Limb-Girdle Muscular Dystrophy Type 2A: *CAPN3*
•**General Population:** 1/832

Limb-Girdle Muscular Dystrophy Type 2D: *SGCA*
•**General Population:** 1/12476

Limb-Girdle Muscular Dystrophy Type 2E: *SGCB*
•**General Population:** 1/7129

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency: *HADHA*
•**General Population:** 1/35000

Lysosomal Acid Lipase Deficiency: *LIPA*
•**General Population:** 1/11100

Maple Syrup Urine Disease Type IA: *BCKDHA*
•**General Population:** 1/37200

Maple Syrup Urine Disease Type IB: *BCKDHB*
•**General Population:** 1/34500

Medium Chain Acyl-CoA Dehydrogenase Deficiency: *ACADM*
•**General Population:** 1/2768

Megalencephalic Leukoencephalopathy with Subcortical Cysts Type 1: *MLC1*
•**General Population:** 1/8318

Metachromatic Leukodystrophy, *ARSA*-Related: *ARSA*
•**General Population:** 1/1980

Methylmalonic Acidemia with Homocystinuria, Cobalamin C Type: *MMACHC*
•**General Population:** 1/12200

Methylmalonic Acidemia with Homocystinuria, Cobalamin D Type: *MMADHC*
•**General Population:** 1/49900

Methylmalonic Acidemia, *MMAA*-Related: *MMAA*
•**General Population:** 1/31500

Methylmalonic Acidemia, *MMAB*-Related: *MMAB*
•**General Population:** 1/45500

Mucolipidosis Type IV: *MCOLN1*
•**General Population:** 1/49900

Mucopolysaccharidosis Type I (Hurler Syndrome): *IDUA*
•**General Population:** 1/4900

Multiple Sulfatase Deficiency: *SUMF1*
•**General Population:** 1/1427

Nemaline Myopathy 2: *NEB*
•**General Population:** 1/3140

Nephrotic Syndrome/Congenital Finnish Nephrosis, *NPHS1*-Related: *NPHS1*
•**General Population:** 1/49900

Nephrotic Syndrome/Steroid-Resistant Nephrotic Syndrome, *NPHS2*-Related: *NPHS2*
•**General Population:** 1/49900

Neuronal Ceroid Lipofuscinosis, *CLN3*-Related: *CLN3*
•**General Population:** 1/5726

Neuronal Ceroid Lipofuscinosis, *CLN5*-Related: *CLN5*
•**General Population:** 1/7130

Neuronal Ceroid Lipofuscinosis, *PPT1*-Related: *PPT1*
•**General Population:** 1/9900

Neuronal Ceroid Lipofuscinosis, *TPP1*-Related: *TPP1*
•**General Population:** 1/8300

Neuronal Ceroid Lipofuscinosis/Northern Epilepsy, *CLN8*-Related: *CLN8*
•**General Population:** 1/6634

Niemann-Pick Type A/B: *SMPD1*
•**General Population:** 1/4980

Niemann-Pick Type C, *NPC1*-Related: *NPC1*
•**General Population:** 1/1820

Nijmegen Breakage Syndrome: *NBN*
•**General Population:** 1/49900

Nonsyndromic Hearing Loss: *GJB2/GJB3/GJB6*
GJB2
•**General Population:** 1/2450
GJB3
•**General Population:** 1/49900
GJB6
•**General Population:** 1/4990

Pendred Syndrome: *SLC26A4/FOXI1/KCNJ10*
SLC26A4
•**General Population:** 1/1580
FOXI1
•**General Population:** 1/49900
KCNJ10 (also associated with SeSAME syndrome)
•**General Population:** 1/49900

Phenylalanine Hydroxylase Deficiency: *PAH*
•**General Population:** 1/5700

Phosphoglycerate Dehydrogenase Deficiency/ Neu-Laxova Syndrome: *PHGDH*
•**General Population:** 1/9980

Polycystic Kidney Disease, Autosomal Recessive: *PKHD1*
•**General Population:** 1/1380

***POMGNT1*-Related Disorders:** *POMGNT1*
•**General Population:** 1/24950

Primary Carnitine Deficiency: *SLC22A5*
•**General Population:** 1/234

Primary Hyperoxaluria Type 1, *AGXT*-Related: *AGXT*
•**General Population:** 1/13400

Primary Hyperoxaluria Type 2, *GRHPR*-Related: *GRHPR*
•**General Population:** 1/49900

Propionic Acidemia, *PCCA*-Related: *PCCA*
•**General Population:** 1/4460

Propionic Acidemia, *PCCB*-Related: *PCCB*
•**General Population:** 1/22300

Pseudocholinesterase Deficiency: *BCHE*
•**General Population:** 1/2100

Pycnodysostosis: *CTSK*
•**General Population:** 1/43700

Rhizomelic Chondrodysplasia Punctata Type 1/Refsum Disease: *PEX7*
•**General Population:** 1/3900

Short Chain Acyl-CoA Dehydrogenase Deficiency: *ACADS*
•**General Population:** 1/9300

Sialic Acid Storage Disorders: *SLC17A5*
•**General Population:** 1/3565

Sickle Cell (HbS) and HbC Disease: *HBB*
•**General Population:** 1/1800

Sjögren-Larsson Syndrome: *ALDH3A2*
•**General Population:** 1/16634

***SLC26A2*-Related Disorders:** *SLC26A2*
•**General Population:** 1/3140

Smith-Lemli-Opitz Syndrome: *DHCR7*
•**General Population:** 1/1750

Spinal Muscular Atrophy: *SMN1/SMN2*
•**General Population:** 1/5300

Tay-Sachs Disease: *HEXA*
•**General Population:** 1/24900

Tyrosine Hydroxylase Deficiency: *TH*
•**General Population:** 1/49900

Tyrosinemia Type I: *FAH*
•**General Population:** 1/2480

Tyrosinemia Type II: *TAT*
•**General Population:** 1/2490

Usher Syndrome Type IB/*MYO7A*-Related Disorders: *MYO7A*
•**General Population:** 1/19900

Usher Syndrome Type ID/*CDH23*-Related Disorders: *CDH23*
•**General Population:** 1/1059

Usher Syndrome Type IF/*PCDH15*-Related Disorders: *PCDH15*
•**General Population:** 1/1079

Usher Syndrome Type IIA/*USH2A*-Related Disorders: *USH2A*
•**General Population:** 1/1850

Usher Syndrome Type IIIA: *CLRN1*
•**General Population:** 1/53400

Very Long Chain Acyl-CoA Dehydrogenase Deficiency: *ACADVL*
•**General Population:** 1/9900

Wilson Disease: *ATP7B*
•**General Population:** 1/4450

X-Linked Adrenoleukodystrophy: *ABCD1**
•**General Population:** Reduced

X-Linked Congenital Adrenal Hypoplasia: *NR0B1**
•**General Population:** Reduced

X-Linked Juvenile Retinoschisis: *RS1**
•**General Population:** Reduced

Zellweger Syndrome Spectrum, *PEX1*-Related: *PEX1*
•**General Population:** 1/2860

Usher Syndrome, *USH2A*-Related Disorders

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Usher syndrome, *USH2A*-related disorders. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Usher syndrome, *USH2A*-related disorders for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for Usher syndrome, *USH2A*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

Usher Syndrome, *USH2A*-Related Disorders Explained

What is Usher Syndrome, *USH2A*-Related Disorders?

Usher syndrome, *USH2A*-related disorders is a group of inherited disorders characterized by mild-to-severe hearing loss and vision loss that worsen over time. Individuals with this type of Usher syndrome typically have hearing loss at birth that can get more severe over time. Vision loss begins in adolescence or adulthood and progressively worsens over time.

Prognosis

Individuals have hearing and vision impairment. The condition does not affect a person's life expectancy or intelligence.

Treatment

Treatment is mostly supportive, as there is no cure. Optimizing communication is important. Some individuals may benefit from hearing aids or cochlear implantation and speech training to normalize language, while others opt to learn sign language. Routine eye exams are recommended.

Resources

Usher Syndrome Coalition
https://www.usher-syndrome.org/

National Institute on Deafness and Other Communication Disorders
https://www.nidcd.nih.gov/health/usher-syndrome

National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2023