
myriad
WOMEN'S HEALTH
UTMOST CONFIDENCE IN EVERY RESULT
Myriad Foresight® Carrier Screen

# ELEVATE QUALITY OF CARE WITH EXPANDED CARRIER SCREENING (ECS)

Carrier screening is used to identify couples who are at risk of passing inherited disorders to their children. Traditionally, carrier screening has been offered to patients based on their ethnic background or family history.

However, this approach can miss couples at risk of having a pregnancy affected by a genetic disease.

8 YEARS

**Did you know**

It takes an average of **8 years** to diagnose a rare genetic disease.[1]

## Collectively common

The total risk of serious disorders identified through ECS* is higher than the incidence of routinely screened for conditions.

**ECS** **1 in 300** pregnancies — **Approximately 1 in 300 pregnancies are affected by a condition screened by the Foresight Carrier Screen (ECS)[2]**

| DOWN SYNDROME | OPEN NEURAL TUBE DEFECTS | CYSTIC FIBROSIS |
|---|---|---|
| **1 in 800** births[3] | **1 in 1,000** births[4] | **1 in 3,500** births[5] |

Elevate quality of care by offering expanded carrier screening to patients regardless of family history or ethnicity.

*For persons receiving Foresight Universal (176 condition) screening. Modeled US population, excluding those with family history.

# LET'S MAKE ECS ROUTINE PRACTICE

In 2017, the American College of Obstetrics and Gynecology (ACOG) recognized expanded carrier screening as an acceptable screening strategy.[6]

## Consistency in care

Offering ECS routinely, regardless of family history or reported ethnicity, is the only screening approach that ensures consistent care for all patients. ACOG also acknowledges the need to streamline the screening approach to benefit more patients.

> Each health care provider should establish a standard approach that is consistently offered to and discussed with each patient.
>
> — ACOG 690[6]

## Choosing the Myriad Foresight® Carrier Screen

Selecting a lab that enables you to offer the best test while streamlining the work associated with detecting more carriers allows you to confidently integrate ECS into your practice.

### Panel with purpose

Have the utmost confidence in every result with the highest published at-risk couple detection for serious conditions

### Pioneer and leader

Trust in the only validated ECS panel in the US, backed by 20+ peer-reviewed publications and >900,000 patients screened

### Complete practice support

Make it easy to integrate routine screening in your practice with the support of Myriad Complete™

# A PANEL WITH PURPOSE: UNMATCHED DETECTION OF SERIOUS DISORDERS

## The Foresight Carrier Screen detection rates

The true goal of carrier screening is to detect at-risk couples of serious diseases. That's why we've designed the Foresight Carrier Screen to maximize detection rates for the diseases that matter the most.

**1 in 22**
**couples at-risk**
Leads the industry in helping providers identify couples at-risk for serious and actionable conditions.[2]

**>99%**
**for most genes**
The overwhelming majority of genes on the panel have detection rates >99%, which ensures utmost confidence in both positive and negative results.

## Prioritizing clinical significance in panel design

To identify appropriate diseases for our test panel, our team of experts evaluated >650 genes based on strict criteria. Our goal is to produce not simply more, but meaningful clinical information.

### The Process[7]

| | |
|---|---|
| **Severity** | Is this condition only mild? Or is it serious (moderate, severe or profound)?[8] |
| **Actionability** | Is this information helpful to patients? |
| **Prevalence** | Is the condition common enough to be of value? |
| **Sensitivity** | With the best technology available, how well can we identify carriers? |

**Foresight Carrier Screen (>176 diseases)**

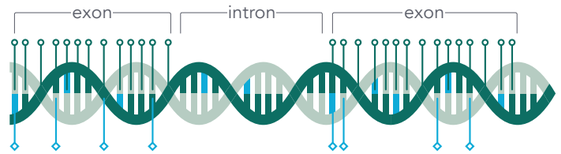
Using these criteria, we selected >176 diseases for the Foresight Carrier Screen that are serious, clinically-actionable, and prevalent, with maximum gene-level sensitivity.

Strict disease inclusion criteria ensure that we provide meaningful clinical information to you and your patients.

VARIANT IDENTIFICATION

### 1 Highly advanced methodology designed to maximize detection rates

Full-exon sequencing—the core methodology




○ **Full-exon sequencing** looks at the entire exon to identify all disease-causing mutations

◇ **Targeted sequencing** focuses only on specific areas of the exon where mutations are associated with a disease

### 2 Custom assays maximize detection rates for difficult-to-sequence genes

**Case example:** Congenital Adrenal Hyperplasia, CYP21A2-Related (CYP21A2)*


99% identical between pseudogene and gene

| Carrier Screen | Detection rate |
|---|---|
| Foresight Carrier Screen: | 88 – >99% |
| Panel without custom assays: | Not tested |

### 3 Panel-wide deletions calling provides an additional detection rate advantage

**Case example:** achieving >99% cystic fibrosis (CF) detection rate


| Carrier Screen | Result |
|---|---|
| Foresight Carrier Screen: | Carrier |
| Panel without deletions calling: | Non-Carrier |

VARIANT INTERPRETATION

### 4 Real-time curation unlocks the true potiential of full-exon sequencing

When a novel variant is detected during the sequencing process, our team of PhD scientists and genetic counselors investigate its pathogenicity, in accordance with ACMG and AMP guidelines.[9] A combination of manual review and sophisticated software are used to ensure a thorough exploration and analysis. Only known and likely pathogenic variants are reported.

# WHY DETECTION RATE MATTERS: INTERPRETING NEGATIVE RESULTS

The higher the detection rate for a gene, the lower the risk is to have a false negative for that condition.

**SHE'S A CARRIER FOR CYSTIC FIBROSIS**
(NORTHERN EUROPEAN DESCENT)

**HIS RESULT IS NEGATIVE**
(HISPANIC DESCENT)

## Implications: Can you trust his negative result?

| | Targeted genotyping (97 mutations) | Foresight Carrier Screen |
|---|---|---|
| Detection rate | 78%[10] | >99% |
| Carrier couples missed | 1 in 5 | ~0 in 5 |
| | **Additional testing to validate negative result (and increased patient anxiety)** | **Trust in negative results** |

When one partner is a carrier, a 78% detection rate means you could miss 2 out of 10 at-risk couples.

By comparison, the Foresight Carrier Screen offers >99% detection rates for the vast majority of the genes on the panel, providing utmost confidence in every result.



# FORESIGHT CARRIER SCREEN DISEASE LIST

The Myriad Foresight Carrier Screen focuses on serious, clinically-actionable, and prevalent conditions to ensure you are providing meaningful information to your patients.

## Fundamental panel

Cystic Fibrosis *(CFTR)* ACOG, ACMG

Spinal Muscular Atrophy *(SMN1)** ACOG ACMG

Fragile X Syndrome *(FMR1)** X-linked

## Fundamental Plus panel

Alpha Thalassemia *(HBA1/HBA2)** ACOG ACMG

Bloom Syndrome *(BLM)* ACMG

Canavan Disease *(ASPA)* ACOG ACMG

Cystic Fibrosis *(CFTR)* ACOG ACMG

Familial Dysautonomia *(IKBKAP)* ACOG ACMG

Fanconi Anemia, Type C *(FANCC)* ACMG

Gaucher Disease *(GBA)** ACMG

Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) *(HBB)* ACOG

Hexosaminidase A Deficiency (including Tay-Sachs Disease) *(HEXA)* ACOG ACMG

Mucolipidosis IV *(MCOLN1)* ACMG

Niemann-Pick Disease, SMPD1-Associated *(SMPD1)* ACMG

Spinal Muscular Atrophy *(SMN1)** ACOG ACMG

Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy) *(DMD)* X-linked

Fragile X Syndrome *(FMR1)** X-linked

## Universal panel

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (*CYP11B1*)

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (*PTS*)

ABCC8-Related Familial Hyperinsulinism (*ABCC8*)

Adenosine Deaminase Deficiency (*ADA*)

Adrenoleukodystrophy: X-Linked (*ABCD1*) X-linked

Alpha Thalassemia (*HBA1/HBA2*)* ACOG ACMG

Alpha-Mannosidosis *(MAN2B1)*

Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) (*SGCA*)

Alport Syndrome, X-Linked (*COL4A5*) X-linked

Alstrom Syndrome (*ALMS1*)

AMT-Related Glycine Encephalopathy (*AMT*)

Andermann Syndrome (*SLC12A6*)

Argininemia (*ARG1*)

Argininosuccinic Aciduria (*ASL*)

Aspartylglycosaminuria (*AGA*)

Ataxia with Vitamin E Deficiency (*TTPA*)

Ataxia-Telangiectasia (*ATM*)

ATP7A-Related Disorders (*ATP7A*) X-linked

Autoimmune Polyglandular Syndrome Type 1 (*AIRE*)

Autosomal Recessive Osteopetrosis, Type 1 (*TCIRG1*)

Autosomal Recessive Polycystic Kidney Disease, PKHD1-Related (*PKHD1*)

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay *(SACS)*

Bardet-Biedl Syndrome, BBS1-Related (*BBS1*)

Bardet-Biedl Syndrome, BBS10-Related (*BBS10*)

Bardet-Biedl Syndrome, BBS12-Related (*BBS12*)

Bardet-Biedl Syndrome, BBS2-Related (*BBS2*)

BCS1L-Related Disorders *(BCS1L)*

Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (*SGCB*)

Biotinidase Deficiency (*BTD*)

Bloom Syndrome (*BLM*) ACMG

Calpainopathy (*CAPN3*)

Canavan Disease (*ASPA*) ACOG ACMG

Carbamoylphosphate Synthetase I Deficiency (*CPS1*)

Carnitine Palmitoyltransferase IA Deficiency (*CPT1A*)

Carnitine Palmitoyltransferase II Deficiency (*CPT2*)

Cartilage-Hair Hypoplasia (*RMRP*)

Cerebrotendinous Xanthomatosis (*CYP27A1*)

Citrullinemia, Type 1 (*ASS1*)

CLN3-Related Neuronal Ceroid Lipofuscinosis (*CLN3*)

CLN5-Related Neuronal Ceroid Lipofuscinosis (*CLN5*)

CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (*CLN6*)

CLN8-Related Neuronal Ceroid Lipofuscinosis *(CLN8)*

Cohen Syndrome (*VPS13B*)

COL4A3-Related Alport Syndrome (*COL4A3*)

COL4A4-Related Alport Syndrome (COL4A4)

Combined Pituitary Hormone Deficiency, PROP1-Related (*PROP1*)

Congenital Adrenal Hyperplasia, CYP21A2-Related *(CYP21A2)**

Congenital Disorder of Glycosylation, MPI-Related *(MPI)*

Congenital Disorder of Glycosylation, Type Ia (*PMM2*)

## DISEASE LIST

Congenital Disorder of Glycosylation, Type Ic (*ALG6*)

Costeff Optic Atrophy Syndrome (*OPA3*)

Cystic Fibrosis (*CFTR*) ACOG ACMG

Cystinosis (*CTNS*)

D-Bifunctional Protein Deficiency (*HSD17B4*)

Delta-Sarcoglycanopathy (*SGCD*)

Dihydrolipoamide Dehydrogenase Deficiency (*DLD*)

Dysferlinopathy (*DYSF*)

Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)(*DMD*) X-linked

ERCC6-Related Disorders (*ERCC6*)

ERCC8-Related Disorders (*ERCC8*)

EVC-Related Ellis-Van Creveld Syndrome (*EVC*)

EVC2-Related Ellis-Van Creveld Syndrome (*EVC2*)

Fabry Disease (*GLA*) X-linked

Familial Dysautonomia (*IKBKAP*) ACOG ACMG

Familial Mediterranean Fever (*MEFV*)

Fanconi Anemia Complementation, Group A (*FANCA*)

Fanconi Anemia, FANCC-Related (*FANCC*) ACMG

FKRP-Related Disorders (*FKRP*)

FKTN-Related Disorders (including Walker-Warburg Syndrome) (*FKTN*)

Fragile X Syndrome (*FMR1*)* X-linked

Galactokinase Deficiency (*GALK1*)

Galactosemia (*GALT*)

Gamma-Sarcoglycanopathy (*SGCG*)

Gaucher Disease (*GBA*)* ACMG

GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (*GJB2*)

GLB1-Related Disorders (*GLB1*)

GLDC-Related Glycine Encephalopathy (*GLDC*)

Glutaric Acidemia, GCDH-Related (*GCDH*)

Glycogen Storage Disease, Type Ia (*G6PC*)

Glycogen Storage Disease, Type Ib (*SLC37A4*)

Glycogen Storage Disease, Type III (*AGL*)

GNE Myopathy *(GNE)*

GNPTAB-Related Disorders (*GNPTAB*)

HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (*HADHA*)

Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)(*HBB*) ACOG

Hereditary Fructose Intolerance (*ALDOB*)

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (*LAMB3*)

Hexosaminidase A Deficiency (including Tay-Sachs Disease) (*HEXA*) ACOG ACMG

HMG-CoA Lyase Deficiency (*HMGCL*)

Holocarboxylase Synthetase Deficiency (*HLCS*)

Homocystinuria, CBS-Related *(CBS)*

Hydrolethalus Syndrome (*HYLS1*)

Hypophosphatasia (*ALPL*)

Isovaleric Acidemia (*IVD*)

Joubert Syndrome 2 (*TMEM216*)

Junctional Epidermolysis Bullosa, LAMC2-Related *(LAMC2)*

Junctional Epidermolysis Bullosa, LAMA3-Related *(LAMA3)*

KCNJ11-Related Familial Hyperinsulinism (*KCNJ11*)

Krabbe Disease (*GALC*)

LAMA2-Related Muscular Dystrophy (*LAMA2*)

Leigh Syndrome, French-Canadian Type (*LRPPRC*)

Lipoid Congenital Adrenal Hyperplasia (*STAR*)

Lysosomal Acid Lipase Deficiency (*LIPA*)

Maple Syrup Urine Disease, Type Ia (*BCKDHA*)

Maple Syrup Urine Disease, Type Ib *(BCKDHB)*

Maple Syrup Urine Disease, Type II (*DBT*)

Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*)

Megalencephalic Leukoencephalopathy with Subcortical Cysts (*MLC1*)

Metachromatic Leukodystrophy (*ARSA*)

Methylmalonic Acidemia, cblA Type (*MMAA*)

Methylmalonic Acidemia, cblB Type (*MMAB*)

Methylmalonic Aciduria and Homocystinuria, cblC Type (*MMACHC*)

MKS1-Related Disorders (*MKS1*)

Mucolipidosis III Gamma (*GNPTG*)

Mucolipidosis IV (*MCOLN1*) ACMG

Mucopolysaccharidosis, Type I (including Hurler Syndrome) (*IDUA*)

Mucopolysaccharidosis, Type II (*IDS*) X-linked

Mucopolysaccharidosis, Type IIIA (*SGSH*)

Mucopolysaccharidosis, Type IIIB (*NAGLU*)

Mucopolysaccharidosis, Type IIIC (*HGSNAT*)

MUT-Related Methylmalonic Acidemia (*MUT*)

MYO7A-Related Disorders (*MYO7A*)

NEB-Related Nemaline Myopathy (*NEB*)

Nephrotic Syndrome, NPHS1-Related *(NPHS1)*

Niemann-Pick Disease, SMPD1-Related *(SMPD1)* ACMG

Niemann-Pick Disease, Type C1 *(NPC1)*

Niemann-Pick Disease, Type C2 (*NPC2*)

Nijmegen Breakage Syndrome (*NBN*)

Ornithine Transcarbamylase Deficiency (*OTC*) X-linked

PCCA-Related Propionic Acidemia (*PCCA*)

PCCB-Related Propionic Acidemia (*PCCB*)

PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (*PCDH15*)

Pendred Syndrome (*SLC26A4*)

Peroxisome Biogenesis Disorder, Type 1 *(PEX1)*

Peroxisome Biogenesis Disorder, Type 3 (*PEX12*)

Peroxisome Biogenesis Disorder, Type 4 (*PEX6*)

Peroxisome Biogenesis Disorder, Type 5 (*PEX2*)

Peroxisome Biogenesis Disorder, Type 6 (*PEX10*)

Phenylalanine Hydroxylase Deficiency (*PAH*)

POMGNT-Related Disorders *(POMGNT1)*

Pompe Disease (*GAA*)

PPT1-Related Neuronal Ceroid Lipofuscinosis (*PPT1*)

Primary Carnitine Deficiency (*SLC22A5*)

Primary Hyperoxaluria, Type 1 (*AGXT*)

Primary Hyperoxaluria, Type 2 (*GRHPR*)

Primary Hyperoxaluria, Type 3 (*HOGA1*)

Pycnodysostosis (*CTSK*)

Pyruvate Carboxylase Deficiency (*PC*)

Rhizomelic Chondrodysplasia Punctata, Type 1 (*PEX7*)

RTEL1-Related Disorders (*RTEL1*)

Salla Disease (*SLC17A5*)

Sandhoff Disease (*HEXB*)

Short Chain Acyl-CoA Dehydrogenase Deficiency (*ACADS*)

Sjogren-Larsson Syndrome (*ALDH3A2*)

SLC26A2-Related Disorders *(SLC26A2)*

Smith-Lemli-Opitz Syndrome (*DHCR7*)

Spastic Paraplegia, Type 15 (*ZFYVE26*)

Spinal Muscular Atrophy (*SMN1*)* ACOG ACMG

Spondylothoracic Dysostosis (*MESP2*)

Steroid-Resistant Nephrotic Syndrome (*NPHS2*)

TGM1-Related Autosomal Recessive Congenital Ichthyosis (*TGM1*)

TPP1-Related Neuronal Ceroid Lipofuscinosis (*TPP1*)

Tyrosine Hydroxylase Deficiency *(TH)*

Tyrosinemia, Type I (*FAH*)

Tyrosinemia, Type II (*TAT*)

USH1C-Related Disorders (*USH1C*)

USH2A-Related Disorders (*USH2A*)

Usher Syndrome, Type 3 (*CLRN1*)

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*)

Wilson Disease (*ATP7B*)

X-Linked Congenital Adrenal Hypoplasia (*NR0B1*) X-linked

X-Linked Juvenile Retinoschisis (*RS1*) X-linked

X-Linked Myotubular Myopathy (*MTM1*) X-linked

X-Linked Severe Combined Immunodeficiency (*IL2RG*) X-linked

Xeroderma Pigmentosum, Group A (*XPA*)

Xeroderma Pigmentosum, Group C (*XPC*)

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ACOG
Indicates disease listed in ACOG guidelines

ACMG
Indicates disease listed in ACMG guidelines

X-linked
Indicates X-linked disorders

*Analyzed using custom assay

# MYRIAD COMPLETE™: YOUR PARTNER IN PATIENT CARE

## We make screening simple for your patients and for your practice

### EDUCATION

We provide resources to help you educate your patients about Myriad products.

- Patient education in multiple formats
- Comprehensive provider support
- Patient identification tools and tele-education

### ACCESS

We offer a comprehensive program to make our products accessible for more of your patients.

- Personalized cost estimates
- Broad in-network status with health plans
- Comprehensive financial assistance

### RESULTS

We deliver screening and testing results effectively and thoroughly so you can focus on care plans.

- Automated email and text notifications
- Online results portal and tracking
- Resources to assist with results comprehension

### CONSULTS

We offer consults with genetic counselors to ensure your patients understand their results.

- Consultation with genetic counselors
- Detailed report available for providers
- Provider access to genetic counselors

Visit **myriadwomenshealth.com/access** to learn more about how Myriad makes screening accessible



# EXPERIENCE THE MYRIAD ADVANTAGE

## Unparalleled performance

The Myriad Foresight Carrier Screen has been methodically designed to achieve the highest published at-risk couple detection for serious conditions and results that offer the utmost confidence in patient care.

## Complete practice support

From ordering to automated online results reporting to on-demand genetic counseling — Myriad Complete makes it easier for your practice to integrate and offer expanded carrier screening.

## About us

Myriad Women's Health is your premier genetic screening and testing partner when you need a result with actionable guidance to deliver superior patient care which empowers women and their families to make critical and timely healthcare decisions.

Our genetic products include:

**myRisk® Hereditary Cancer**
Help patients get ahead of cancer with our hereditary cancer test.

- For men and women
- 4mL blood or saliva sample
- Results in ~2 weeks

**Foresight® Carrier Screen**
Unmatched detection of at-risk couples for serious conditions.

- For men and women
- 4mL blood or saliva sample
- Results in ~2 weeks

**Prequel™ Prenatal Screen**
Reliable results, the first time with our non-invasive prenatal screen.

- For pregnant women
- 10mL blood sample
- Results in ~1 week

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