COUNSYL FAMILY PREP SCREEN DISEASE LIST

UNIVERSAL PANEL

ABCC8-Related Hyperinsulinism (3)
Achromatopsia (3)
Alkaptonuria (11)
Alpha-1 Antitrypsin Deficiency (1)
Alpha-Mannosidosis (1)
Andermann Syndrome (2)
ARSACS (2)
Aspartylglycosaminuria (1)
Ataxia with Vitamin E Deficiency (1)
Ataxia-Telangiectasia (8)
Autosomal Recessive Polycystic Kidney Disease (4)
Bardet-Biedl Syndrome
- BBS1-Related (1)
- BBS10-Related (1)

Biotinidase Deficiency (4)
Bloom Syndrome (1) ACMG
Canavan Disease (4) ACOG ACMG
Carnitine Palmitoyltransferase IA Deficiency (1)
Carnitine Palmitoyltransferase II Deficiency (3)
Cartilage-Hair Hypoplasia (1)
Choroideremia (1)
Cohen Syndrome (1)
Citrullinemia Type 1 (2)
Congenital Disorder of Glycosylation
- Type 1a (4)
- Type 1b (1)

Congenital Finnish Nephrosis (2)
Costeff Optic Atrophy Syndrome (1)
Cystic Fibrosis (100) ACOG ACMG
Cystinosis (4)
D-Bifunctional Protein Deficiency (2)
Factor XI Deficiency (4)
Familial Dysautonomia (2) ACOG ACMG
Familial Mediterranean Fever (4)
Fanconi Anemia Type C (3) ACMG
Fragile X Syndrome (female specimens only) (1)
Galactosemia (8)
Gaucher Disease (10) ACMG
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness (7)
Glutaric Acidemia Type 1 (1)
Glycogen Storage Disease
- Type Ia (7)
- Type Ib (2)
- Type III (3)
- Type V (4)

GRACILE Syndrome (1)
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (28) ACOG
Hereditary Fructose Intolerance (3)
Hereditary Thymine-Uraciluria (1)
Herlitz Junctional Epidermolysis Bullosa
- LAMA3-Related (1)
- LAMB3-Related (3)
- LAMC2-Related (1)

Hexosaminidase A Deficiency (including Tay-Sachs Disease) (9) ACOG ACMG
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency (1)
Hurler Syndrome (2)
Hypophosphatasia, Autosomal Recessive (4)
Inclusion Body Myopathy 2 (2)
Isovaleric Acidemia (1)
Joubert Syndrome 2 (1)
Krabbe Disease (2)
Limb-Girdle Muscular Dystrophy
- Type 2D (1)
- Type 2E (1)

Lipoamide Dehydrogenase Deficiency (2)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (1)
Maple Syrup Urine Disease Type 1B (3)
Medium Chain Acyl-CoA Dehydrogenase Deficiency (2)
Megalencephalic Leukoencephalopathy with Subcortical Cysts (4)
Metachromatic Leukodystrophy (5)
Mucolipidosis IV (2) ACMG
Muscle-Eye-Brain Disease (1)
NEB-Related Nemaline Myopathy (1)
Neuronal Ceroid Lipofuscinosis
- CLN3-related (1)
- CLN5-related (1)
- PPT1-related (3)
- TPP1-related (3)

Niemann-Pick Disease
- SMPD1-Associated (4) ACMG
- Type C (1)

Nijmegen Breakage Syndrome (1)
Northern Epilepsy (1)
Pendred Syndrome (5)
PEX1-Related Zellweger Syndrome Spectrum (2)
Phenylalanine Hydroxylase Deficiency (13)
Polyglandular Autoimmune Syndrome Type 1 (2)
Pompe Disease (4)
Primary Carnitine Deficiency (1)
Primary Hyperoxaluria
- Type 1 (2)
- Type 2 (2)

PROP1-Related Combined Pituitary Hormone Deficiency (1)
Pseudocholinesterase Deficiency (1)
Pycnodysostosis (1)
Rhizomelic Chondrodysplasia Punctata Type 1 (4)
Salla Disease (1)
Segawa Syndrome (1)
Short Chain Acyl-CoA Dehydrogenase Deficiency (1)
Sjogren-Larsson Syndrome (1)
Smith-Lemli-Opitz Syndrome (13)
Spinal Muscular Atrophy (1) ACMG
Steroid-Resistant Nephrotic Syndrome (2)
Sulfate Transporter-Related Osteochondrodysplasia (4)
Tyrosinemia Type I (6)
Usher Syndrome
- Type 1F (1)
- Type 3 (1)

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (1)
Wilson Disease (2)
X-Linked Juvenile Retinoschisis (3)

ACOG
Indicates testing recommended by ACOG

ACMG
Indicates testing recommended by ACMG

Additional Information: Counsyl screening identifies but does not eliminate risk. Results are based on probabilities, and as such, cannot give 100% definitive conclusions, and cannot predict all disease. In addition to the Family Prep Screen, further testing options may be recommended to your patients. If only one member of a couple is of Ashkenazi Jewish background, a biochemical assay for Tay-Sachs disease can be performed.[1] Individuals of African, Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and should also be offered carrier testing by CBC and hemoglobin electrophoresis or HPLC.[2]

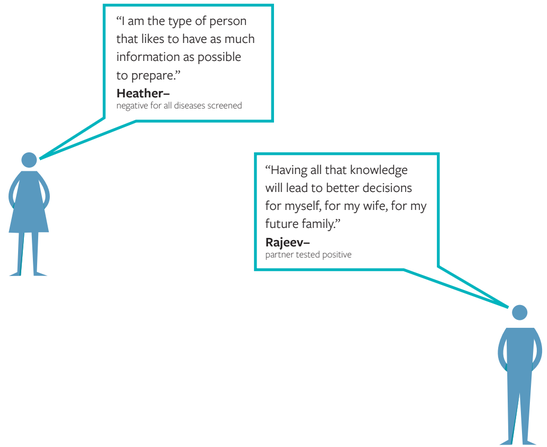
1. S Gross, BA Pletcher, KG Monaghan. Carrier screening in individuals of Ashkenazi Jewish descent. *Genetics in Medicine* (2008) 10. 54–56.
2. ACOG, Hemoglobinopathies in pregnancy. ACOG Practice Bulletin No. 78. (2007), 1–9.

It's Good to Know

You can take the Family Prep Screen before or during pregnancy

It's normal to be a carrier – what you really want to know is if both partners are carriers of the same disease

Most carriers have no family history of the disease within their family



LifeLabs Genetics™

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CSYL002 | V.1 | MAR 2015 PHYSICIAN DETAIL AID

COUNSYL FAMILY PREP SCREEN

Knowing whether you are a carrier of genetic conditions provides valuable health information when planning a family

Counsyl | Your partners in patient care | LifeLabs Genetics™

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TEST OPTIONS

Family Prep Screen 1.0
Quick turnaround

Targeted carrier screening ensures that patients receive valuable health information quickly with clear results from a pre-selected mutation panel

102	400	~2
Genes tested	Mutations tested	Week turnaround

Family Prep Screen 2.0
Thorough analysis

Next-generation sequencing provides the most comprehensive analysis for detection of genetic mutations

102	FULL	~3
Genes tested	Gene sequencing	Week turnaround

The Counsyl Family Prep Screen covers many types of diseases:

80
impact life expectancy and quality of life

- Cystic fibrosis
  40 years
- Tay-Sachs disease
  3-5 years
- Spinal muscular atrophy
  less than 2 years

59
have treatment options available

- MCADD
  normal life quality expected with treatment
- Wilson Disease
  most symptoms prevented with early treatment
- Beta thalassemia
  can extend beyond 40 years with treatment

45
carry a significant risk for intellectual disability

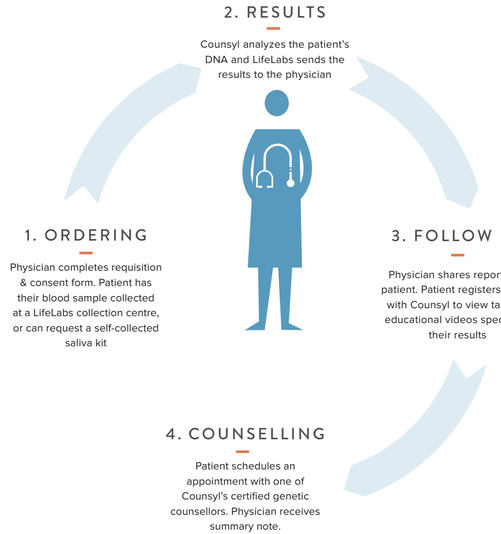
- Fragile X syndrome
- Smith-Lemli-Opitz syndrome
- Costeff optic atrophy syndrome

1/400 births are affected by a disease on the Counsyl Family Prep Screen.

ABOUT THE CATEGORIES

The information presented in this document is meant as a quick reference to diseases screened by Counsyl Family Prep Screen and is not meant to be a comprehensive guide. Individual diseases can have widely varying phenotypes not captured here. For specific disease information, please refer to counsyl.com/diseases or www.lifelabsgenetics.com.

PATIENT FLOW



OUR TEAM OF GENETIC COUNSELLORS ARE BY YOUR SIDE

SAMPLE REPORT

Full sample reports are available from www.lifelabsgenetics.com

Family Prep Screen — POSITIVE: CARRIER

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

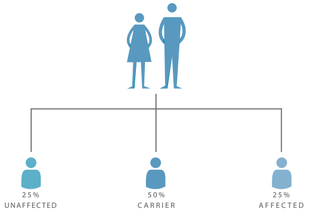
PANEL DETAILS
Universal Panel with Fragile X Syndrome **(102 diseases tested)**

VERSION
JANE DOE **(Family Prep Screen 1.0)**

RESULTS SUMMARY

Risk Details	JANE DOE	Partner
POSITIVE: CARRIER Smith-Lemli-Opitz Syndrome Reproductive Risk: 1 in 200 Inheritance: Autosomal Recessive	CARRIER* NM_001360.2(DHCR7):c.964-1G>C (aka IVS8-1G>C) heterozygote	N/A Carrier testing should be considered.

*Carriers generally do not experience symptoms.



WHAT IS CARRIER SCREENING?

A carrier screen analyzes a person's genes in order to determine if that person is a recessive genetic disease carrier. A screen is able to detect many, but not all, carriers of a disease.

WHAT IS A RECESSIVE DISEASE AND WHAT IS A CARRIER?

Recessive diseases are caused by changes (called mutations) in a person's genes. Every person has two copies of most genes, one inherited from each parent. A recessive disease occurs when both copies of the same gene have a mutation.

A carrier is someone who has only one gene with a mutation and one gene that is unaffected. Carriers are typically symptom-free and do not know that they carry a mutation.

Some of the diseases on the Family Prep Screen are inherited differently — only the female needs to be a carrier to have a baby at risk. Fragile X syndrome is an example of this.

WHAT DOES IT MEAN TO BE A CARRIER?

When two parents are carriers of the same genetic disease, each child has a 1 in 4 (or 25%) chance of having that disease. For certain diseases, such as Fragile X syndrome, only the mother needs to be carrier for the child to have a high risk. Knowing their carrier status before or early in a pregnancy gives individuals time to learn about the disorder and prepare.

WHAT IF AN INDIVIDUAL IS NOT A CARRIER?

Generally, no follow-up testing is suggested for the diseases screened. It is important to understand that no screen is able to identify every carrier of every disease. You should also know that while the Family Prep Screen covers a lot of information, we cannot screen for all possible birth defects and genetic diseases. Family history or other factors should also be considered.

24%
Percentage of individuals who test positive for at least one disorder on the Counsyl Family Prep Screen 1.0.

1.3%
Percentage of couples that tested positive for the same disorder on the Counsyl Family Prep Screen 1.0.

IT'S GOOD TO KNOW

Patients may want to share their results with close relatives, especially those who are planning on having children in the near future