Introducing the Universal Genetic Test

Comprehensive

The New Standard of Care

- Test for cystic fibrosis, spinal muscular atrophy (SMA), Tay-Sachs disease, sickle cell disease and more than 100 other single gene disorders with a single saliva-based test.
- Individually rare, collectively common, these single gene disorders account for more than 10% of pediatric deaths and 80% occur in children with no known family history of the disorder.

Convenient

Simple, Non-Invasive, and Cost-Effective

- This single saliva-based assay is applicable to all ethnicities and encompasses major standard of care carrier tests.
- Counsyl provides a clear, easy-to-digest report that can be shared and discussed with the patient.
- The test quickly identifies IVF patients who can benefit from preimplantation genetic diagnosis (PGD).

Reliable

Clinically Valid, Grounded In Well-Established Science

- Designed by scientists from Stanford and Harvard, Counsyl's assay is 99.9% accurate for all targeted mutations and more thorough than the single gene tests for many disorders.
- Counsyl's CLIA-certified lab adheres to industry best practices in order to ensure the highest possible levels of accuracy and patient privacy.

"I'm excited about offering the Counsyl test in my practice because it's non-invasive, easy-to-use, comprehensive and cost-effective."

— Dr. Mohit Khera,
Baylor College of Medicine

"Every adult of reproductive age needs the Counsyl test."

— Professor Pasquale Patrizio, MD
Director, Yale Fertility Center

About Counsyl Counsyl is a medical genomics company founded by social entrepreneurs and philanthropists with the audacious belief that every child deserves a chance in life. With the Universal Genetic Test, many previously surprising and incurable childhood diseases can now be prevented.

Counsyl
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Disease List

The following diseases are covered on Counsyl's Universal Genetic Test, listed here with the number of mutations tested.

ACOG = Testing for this disease recommended to be offered by ACOG
ACMG = Testing for this disease recommended to be offered by ACMG

CAUCASIAN

- ACOG ACMG Cystic Fibrosis (109)
- ACMG Spinal Muscular Atrophy (1)
- Autosomal Recessive Polycystic Kidney Disease (5)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (7)
- Phenylalanine Hydroxylase Deficiency (11)

ASHKENAZI JEWISH

- ACOG ACMG Tay-Sachs Disease (4)
- ACOG ACMG Canavan Disease (4)
- ACOG ACMG Cystic Fibrosis (109)
- ACOG ACMG Familial Dysautonomia (3)
- ACMG Bloom Syndrome (2)
- ACMG Gaucher Disease (9)
- ACMG Fanconi Anemia Type C (4)
- ACMG Mucolipidosis IV (2)
- ACMG Niemann Pick Disease Type A (4)
- ACMG Spinal Muscular Atrophy (1)
- Glycogen Storage Disease Type 1a (21)
- Factor XI Deficiency (4)

AFRICAN

- ACOG ACMG Cystic Fibrosis (109)
- ACOG Sickle Cell Disease (37)
- ACMG Spinal Muscular Atrophy (1)

ASIAN

- ACOG ACMG Cystic Fibrosis (109)
- ACOG Beta Thalassemia (35)
- ACMG Spinal Muscular Atrophy (1)
- Glucose-6-Phosphate Dehydrogenase Deficiency (5)
- Phenylalanine Hydroxylase Deficiency (11)
- Wilson Disease (5)

HISPANIC

- ACOG ACMG Cystic Fibrosis (109)
- ACMG Spinal Muscular Atrophy (1)
- Sickle Cell Disease (37)
- Phenylalanine Hydroxylase Deficiency (11)

MIDDLE EASTERN

- ACOG ACMG Cystic Fibrosis (109)
- ACMG Spinal Muscular Atrophy (1)
- Beta Thalassemia (35)
- Familial Mediterranean Fever (13)
- Phenylalanine Hydroxylase Deficiency (11)

ADDITIONAL DISEASES

- ABCC8-Related Hyperinsulinism (3)
- Achromatopsia (6)
- Alkaptonuria (7)
- Alpha-1 Antitrypsin Deficiency (2)
- Andermann Syndrome (2)
- ARSACS (2)
- Aspartylglycosaminuria (2)
- Ataxia With Vitamin E Deficiency (1)
- Ataxia-Telangiectasia (1)
- Bardet-Biedl Syndrome (2)
- Biotinidase Deficiency (7)
- Carnitine Palmitoyltransferase IA Deficiency (2)
- Carnitine Palmitoyltransferase II Deficiency (13)
- Cartilage-Hair Hypoplasia (2)
- Choroideremia (1)
- Congenital Disorder Of Glycosylation (3)
  Types Ia and Ib
- Congenital Finnish Nephrosis (2)
- Cystinosis (4)
- Factor V Leiden Thrombophilia (3)
- Fumarase Deficiency (1)
- Galactosemia (10)
- GJB2-Related DFNB 1 Nonsyndromic Hearing Loss And Deafness (11)
- Glutaric Acidemia Type 1 (2)
- Glycogen Storage Disease (21)
  Types Ib, III, V
- GRACILE Syndrome (1)
- Hereditary Fructose Intolerance (4)
- Hereditary Thymine-Uraciluria (1)
- Herlitz Junctional Epidermolysis Bullosa (7)
- Hexosaminidase A Deficiency (8)
- HFE-Associated Hereditary Hemochromatosis (11)
- Homocystinuria Caused By Cystathionine Beta-Synthase Deficiency (2)
- Hurler Syndrome (2)
- Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (1)
- Hypophosphatasia, Autosomal Recessive (5)
- Inclusion Body Myopathy 2 (1)
- Infantile Refsum Disease (1)
- Isovaleric Acidemia (1)
- Krabbe Disease (3)
- Leigh Syndrome, French-Canadian Type (1)
- Limb-Girdle Muscular Dystrophy Type 2E (1)
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (2)
- Maple Syrup Urine Disease Types 1B and 3 (5)
- Metachromatic Leukodystrophy (4)
- Muscle-Eye-Brain Disease (1)
- MYH-Associated Polyposis (1)
- Neuronal Ceroid Lipofuscinosis (10)
  CLN5-, CLN8-, PPT1- and TPP1-related
- Niemann-Pick Disease Type C (4)
- Nijmegen Breakage Syndrome (1)
- Pendred Syndrome (3)
- Polyglandular Autoimmune Syndrome Type 1 (1)
- Primary Hyperoxaluria (4)
  Types 1 and 2
- Pycnodysostosis (1)
- Rhizomelic Chondrodysplasia Punctata Type 1 (2)
- Salla Disease (2)
- Segawa Syndrome (1)
- Short Chain Acyl-CoA Dehydrogenase Deficiency (2)
- Sjogren-Larsson Syndrome (1)
- Smith-Lemli-Opitz Syndrome (11)
- Sulfate Transporter-Related Oseochondrodysplasia (5)
  Achondrogenesis Type 1B
  Diastrophic Dysplasia
  Recessive Multiple Epiphyseal Dysplasia
- Tyrosinemia Type I (5)
- Usher Syndrome (2)
  Types 1F and 3
- X-Linked Juvenile Retinoschisis (3)