Counsyl

Results Recipient	Reviewing Physician	Male Details	Female Details
Daniel MacArthur Report Date: 10/03/2009	Report reviewed and released by the Counsyl ordering physician.	Name: Daniel MacArthur [redacted] Ethnicity: Northern European Indication: Population Screening Sample Type: Saliva (OG-300) Date of Collection: 07/02/2009 (est.)	Name: Ilana Fisher [redacted] Ethnicity: Northern European Indication: Population Screening Sample Type: Saliva (OG-300) Date of Collection: 07/02/2009 (est.)

Universal Genetic Test

The Universal Genetic Test uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual or couple for 100+ Mendelian diseases. Counsyl reports which mutations, if any, were detected for each disease. The risk of conceiving a child affected with a disease is presented below and calculated using the test results as well as published data on each disease. The child risk summary is provided as an aid to genetic counseling. *

Daniel MacArthur

Daniel MacArthur's DNA test shows that he is a carrier of adenosine monophosphate deaminase deficiency.

Ilana Fisher

Ilana Fisher's DNA test shows that she is a carrier of cystic fibrosis and MTHFR deficiency.

Child Risk Summary

Based on your DNA test results and ethnicities, your child is at increased risk to inherit the disease below. The following pages contain detailed information about your results as well as next steps to take.

Cystic Fibrosis
Child Risk: 1 in 1,400. Risk before testing: 1 in 3,100.

The following diseases were identified as potential risks based on your ethnicities, but your DNA test indicates the actual risk of occurrence is low.

- Autosomal Recessive Polycystic Kidney Disease
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Phenylalanine Hydroxylase Deficiency
- Spinal Muscular Atrophy

* **Limitations:** Interpretation is given as a probability due to the inheritance pattern of these diseases and because only targeted mutations are detected. Other nearby genetic variants may interfere with this detection. Inaccurate reporting of ethnicity or clinical information may cause errors in risk calculation.

Mild Disease Summary

The chart below shows Daniel MacArthur and Ilana Fisher's carrier status for 5 mild diseases. The conditions on this list have been highlighted because they are extremely common in the general population and usually do not cause major health problems. In many cases, individuals with these mild conditions remain asymptomatic. For this reason, the results in this section of the report are unlikely to influence reproductive choices. However for those who do show symptoms, knowledge of one's genetic status for these conditions can be helpful to recognize the disease and direct treatment.

Mild Disease	Daniel MacArthur	Ilana Fisher
Adenosine Monophosphate Deaminase Deficiency	AMPD1:p.Gln12X (Q12X) heterozygote.	No disease-causing mutations detected.
Factor V Leiden Thrombophilia	No disease-causing mutations detected.	No disease-causing mutations detected.
Glucose-6-Phosphate Dehydrogenase Deficiency	No disease-causing mutations detected.	No disease-causing mutations detected.
HFE-Associated Hereditary Hemochromatosis	No disease-causing mutations detected.	No disease-causing mutations detected.
MTHFR Deficiency	No disease-causing mutations detected.	MTHFR:p.Glu429Ala (E429A) heterozygote.

For details on adenosine monophosphate deaminase deficiency, see page 6. For details on MTHFR deficiency, see page 8.

Male	Female
Name: Daniel MacArthur [redacted]	Name: Ilana Fisher

Positive Report: Cystic Fibrosis

This disease report is included due to increased child risk.

Increased risk	Your child's risk: 1 in 1,400	Risk before testing: 1 in 3,100

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	CFTR:p.Asp1152His (D1152H) heterozygote.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 340.	This individual is a carrier of cystic fibrosis. The D1152H mutation is associated with a broad clinical spectrum that can range from asymptomatic to severe.

Daniel MacArthur's detection rate: 91.7%. **Ilana Fisher's detection rate:** 91.7%.
Variants on the Counsyl panel: 109
Gene: CFTR. **Variants:** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, G91R, E92X, R117C, Y122X, G178R, L206W, G330X, T338I, R347H, R352Q, S364P, G480C, Q493X, V520F, C524X, S549I, S549N, S549R, Q552X, A559T, P574H, G622D, R709X, K710X, Q890X, R1066C, R1070Q, W1089X, Y1092X, M1101K, D1152H, R1158X, S1196X, W1204X, S1235R, Q1238X, S1251N, S1255X, R1283M, dele2-3 21kb, 3199del6, F311del, 394delTT, 574delA, 663delT, 935delA, 936delTA, 1078delT, 1161delC, 1609delCA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 3171delC, 3667del4, 3821delT, 3876delA, 1288insTA, 2184insA, 2307insA, 2869insG, 3905insT, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1811+1.6kbA>G, 1812-1G>A, 1898+1G>T, 1898+5G>T, 3272-26A>G, 3120G>A, 457TAT>G, 2183AA>G, S549R, W1204X, IVS8-5T, I148T, I506V, F508C.

Child Risk

Affected	1 in 1,400
Carrier	1 in 2
Unaffected	1 in 2

What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition characterized by the production of abnormally thick, sticky mucus, particularly in the lungs and digestive system. While it is normal to have mucus lining the organs of the respiratory, digestive, and reproductive systems in order to lubricate and protect them, in people with CF this mucus is thick and sticky. This abnormal mucus results in the clogging and obstructing of various systems in the body. CF is a chronic condition that worsens over time.

Most people with CF experience breathing problems and frequent lung infections that lead to permanent lung damage such as scarring (fibrosis) and sac-like growths (cysts). The pancreas, an organ that produces insulin and digestive enzymes, is often affected by CF. The sticky mucus caused by CF can block ducts which ferry enzymes from the pancreas to the rest of the body, resulting in problems

such as diarrhea, malnutrition, and poor growth. Infertility, particularly in men, and delayed puberty are also common among people with cystic fibrosis.

The severity of symptoms varies from person to person. Most cases of CF are diagnosed in early childhood. Those who are diagnosed after the age of 18 typically have a milder form of the condition.

How common is cystic fibrosis?

According to the National Institutes of Health, CF is the most common deadly inherited condition among Caucasians in the United States. Disease-causing mutations in the CFTR gene are more common in some ethnic populations than others.

Ethnic Group	Carrier Rate	Affected Rate
Caucasian	1 in 28	1 in 3,000
Ashkenazi Jewish	1 in 28	1 in 3,000
Hispanic	1 in 46	1 in 8,300
African American	1 in 66	1 in 17,000
Asian	1 in 87	1 in 30,000

How is cystic fibrosis treated?

There is no treatment that addresses the cause of CF, but there are many options to treat the symptoms it produces. Because thick mucus can build up in the respiratory system, it is important to keep the person's airways open in order to ease breathing and prevent infection. This can be accomplished with various prescription drugs as well as by physically loosening mucus by pounding on the person's back in a prescribed way. This treatment, known as "postural drainage and chest percussion" must be performed by someone other than the affected person, and is typically done at least once daily. As respiratory infections occur, physicians typically prescribe antibiotics.

Physicians will also monitor the digestive system to ensure that the person is getting proper nutrition. Enzymes or vitamin supplements may be prescribed. Both the respiratory and digestive systems of a person with CF must be monitored regularly by his or her medical team.

Surgery may be needed to correct certain problems caused by CF. Lung transplants are an option for some people.

What is the prognosis for someone with cystic fibrosis?

Thanks to improved treatments and a better understanding of the condition, the average life expectancy for people with CF who live to adulthood is 35 years. Children born with CF today who receive early treatment may live even longer.

What next steps could you take?

The Universal Genetic Test indicates that Ilana Fisher is a carrier of cystic fibrosis but Daniel MacArthur is not. Based on the results of this DNA test, as well as the couple's ethnicities, any child they have together would have a 1 in 1,400 chance of developing cystic fibrosis.

There is no requirement to take any further action, however some people choose to take the following actions:

Consult With a Physician or Genetic Counselor

There is an increased risk of Daniel MacArthur and Ilana Fisher's future children inheriting the disease. We strongly suggest the couple speak with a physician or genetic counselor about this finding. These medical professionals may be able to suggest actions the couple can take to lower the risk of their children developing cystic fibrosis.

Genetic testing can be technical and confusing—patients don't have to navigate the process alone. If they have any concerns or questions about the test results, we encourage them to talk to their physician or visit a genetic counselor. These licensed medical professionals can help clarify and explain the results in detail. This is particularly important given Ilana Fisher's positive result.

It is perfectly normal to experience a range of emotions after receiving the results of a genetic test: anxiety, confusion, relief, anger, even denial. Genetic counselors are trained in both genetics and psychosocial counseling to help patients sort through their test results and use them to come to a decision with which they feel comfortable. They will not pressure a patient to make a particular choice, but rather will explain all the options and help him or her find the best one based on his or her values and wishes.

To find a genetic counselor in their area, patients or physicians can visit the National Society of Genetic Counselors website—www.ngsc.org—and click on "Find a Counselor."

Mild Disease Positive Report: Adenosine Monophosphate Deaminase Deficiency

This disease report is included due to positive result for Daniel MacArthur.

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	AMPD1:p.Gln12X (Q12X) heterozygote.	No disease-causing mutations detected.
Interpretation:	This individual is a carrier of adenosine monophosphate deaminase deficiency. Carriers generally do not experience symptoms.	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 5,600.

Daniel MacArthur's non-disease-causing mutations: P48L. **Ilana Fisher's non-disease-causing mutations:** None.
Variants on the Counsyl panel: 2
Gene: AMPD1. **Variants:** Q12X, P48L.

What is adenosine monophosphate deaminase deficiency?

Adenosine monophosphate (AMP) deaminase deficiency is a common inherited condition that can cause muscle pain, cramps, or weakness after exercise. It is important to note that the vast majority people with the disease **do not** develop any symptoms. The reason for this is unknown. Some researchers hypothesize there may be other factors involved, such as mutations in other genes.

People with the condition who do develop symptoms can develop muscle weakness in childhood or early adulthood. After exercise they may be more tired than would be expected and stay tired for a longer period of time. The symptoms do not worsen over time.

As the name indicates, people with AMP deaminase deficiency do not make enough of the enzyme AMP deaminase. This enzyme helps muscle cells produce energy.

How common is adenosine monophosphate deaminase deficiency?

AMP deaminase deficiency is surprisingly common. Roughly 10% to 20% of Caucasians and African Americans are carriers of mutations in the gene that cause this deficiency.

The vast majority of people with AMP deaminase deficiency will show no symptoms.

How is adenosine monophosphate deaminase deficiency treated?

Because most people with AMP deaminase deficiency do not have symptoms, no treatment is required. For those who do experience muscle pain or weakness following exercise, an oral dose of the sugar ribose can help to relieve those symptoms. This ribose is only effective while in the bloodstream, so for sustained relief of symptoms, it must be taken regularly.

What is the prognosis for someone with adenosine monophosphate deaminase deficiency?

The prognosis for a person with AMP deaminase deficiency is very good. Most people will never experience any symptoms of the disease and will live completely normal lives. The small percentage who do experience symptoms can be treated with medication and will also live normal lives.

What next steps could you take?

The Universal Genetic Test has indicated that Daniel MacArthur is a carrier of adenosine monophosphate (AMP) deaminase deficiency.

Carriers of AMP deaminase deficiency are extremely common. This finding is not cause for alarm. Carriers of the condition do not have any symptoms, nor do the vast majority of people who actually have AMP deaminase deficiency. Those affected by the condition typically have mild symptoms.

Consult With a Physician or Genetic Counselor

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It is perfectly normal to experience a range of emotions after receiving the results of a genetic test: anxiety, confusion, relief, anger, even denial. Genetic counselors are trained in both genetics and psychosocial counseling to help patients sort through their test results and use them to come to a decision with which they feel comfortable. They will not pressure a patient to make a particular choice, but rather will explain all the options and help him or her find the best one based on his or her values and wishes.

To find a genetic counselor in their area, patients or physicians can visit the National Society of Genetic Counselors website—www.ngsc.org—and click on "Find a Counselor."

Mild Disease Positive Report: MTHFR Deficiency

This disease report is included due to positive result for Ilana Fisher.

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	MTHFR:p.Glu429Ala (E429A) heterozygote.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 320.	This individual is a carrier of MTHFR deficiency. Carriers generally do not experience symptoms.

Variants on the Counsyl panel: 2
Gene: MTHFR. **Variants:** A222V, E429A.

What is MTHFR deficiency?

MTHFR deficiency is a mild condition associated with a slightly higher risk of neural tube defects and pregnancy loss. Roughly 40% of Americans are carriers of MTHFR deficiency, while 10% have the condition. For the vast majority, it causes no problems with their health or the health of their children. The mild MTHFR deficiency for which Counsyl tests should not be confused with severe MTHFR deficiency.

The MTHFR enzyme is involved in the conversion of the amino acid homocysteine to another amino acid, methionine. People with mild MTHFR deficiency have a decreased ability to perform this conversion, and as a result they have higher levels of homocysteine in their body and lower levels of the vitamin folate.

There are two common mutations found in the MTHFR gene, A222V and E429A (also known as C677T and A1298C respectively). Having one copy of either mutation (being a carrier) is not thought to have any impact on one's health or that of one's children.

The following gene combinations may be significant. Note that they are only significant if they raise the level of homocysteine in the blood, and this does not happen in everyone:

A222V/A222V

For women, two copies of the A222V mutation has been associated with a 2 to 3-fold higher risk of having a child with severe neural tube defects such as spina bifida. This type of birth defect normally affects 1 in 1,000 births, so women with two A222V mutations would face a 2 to 3 in 1,000 risk, which is still low.

Taking folate supplements has been shown to reduce neural tube defects by 75% and may be doing so by normalizing levels of homocysteine in the body.

Some studies have shown that mild MTHFR deficiency may be beneficial in lowering the risks for colorectal cancer or boosting immunity to certain pathogens. These findings are tentative, however.

A222V/E429A

This mutation pairing is thought to share the same risks as described above for the A222V/A222V mutation pairing.

E429A/E429A

This mutation pairing is NOT associated with any elevated risks for health problems.

How common is MTHFR deficiency?

Mild MTHFR deficiency is very common. The table below shows the results of numerous studies conducted worldwide looking at the A222V mutation. The first number represents the percentage of the population who is a carrier and the second number represents the percentage of the population thought to be affected.

Ethnic Group	Carrier Rate	Affected Rate
Hispanic American	48%	15%
Caucasian American	45%	12%
Japanese	45%	12%
German	37%	6%
Asian	29%	3%
African American	24%	2%
Sub-Saharan African	12%	1%

The E429A mutation is less well-studied, but is also thought to be quite common. In three studies, the A222V/E429A mutation pairing was found in 17% of Americans, 15% of Canadians, and 20% of Dutch people.

How is MTHFR deficiency treated?

Most people with mild MTHFR deficiency require no treatment. Because food in the United States is often fortified with vitamins, most people eat sufficient amounts of folate to compensate for higher levels of homocysteine.

Pregnant women with the condition—and all pregnant women—are advised to take folate supplements (folic acid) before and during pregnancy to reduce the risk of birth defects by as much as 75 to 85%. These vitamins are particularly important for women with MTHFR deficiency.

What is the prognosis for someone with MTHFR deficiency?

Based on current scientific knowledge, most people with MTHFR deficiency will be totally unaffected by it. Women face a slightly elevated risk of having a child with neural tube defects, however the risk is still low.

What next steps could you take?

The Universal Genetic Test has indicated that Ilana Fisher is a carrier of MTHFR deficiency. Because MTHFR deficiency is a mild condition in most people and carriers do not typically have any symptoms, there is no cause for concern.

Women who are carriers of MTHFR are not known to be at elevated risk for having children with neural tube defects, however this information can serve as a reminder to take folic acid supplements, which dramatically reduce the incidence of these serious birth

defects. The U.S. Public Health Service and the Centers for Disease Control recommend that all women between the ages of 15 and 45 take 400 micrograms of folic acid daily.

Women who actually have MTHFR deficiency—as opposed to those who are only carriers—can have lower folate levels in the body, making it all the more important to take folic acid supplements prior to and during their pregnancies. Low levels of folic acid in a mother's body have been shown to increase the risk of neural tube defects in her children.

Male carriers of the condition need not take any further steps.

Consult With a Physician or Genetic Counselor

Genetic testing can be technical and confusing—patients don't have to navigate the process alone. If they have any concerns or questions about the test results, we encourage them to talk to their physician or visit a genetic counselor. These licensed medical professionals can help clarify and explain the results in detail.

It is perfectly normal to experience a range of emotions after receiving the results of a genetic test: anxiety, confusion, relief, anger, even denial. Genetic counselors are trained in both genetics and psychosocial counseling to help patients sort through their test results and use them to come to a decision with which they feel comfortable. They will not pressure a patient to make a particular choice, but rather will explain all the options and help him or her find the best one based on his or her values and wishes.

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Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher
[redacted]	

Negative Report: Autosomal Recessive Polycystic Kidney Disease

This disease report is included due to Daniel MacArthur and Ilana Fisher's ethnic backgrounds.

Reduced risk

Your child's risk: **1 in 20,000**

Risk before testing: **1 in 15,000**

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	No disease-causing mutations detected.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72.	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72.

Daniel MacArthur's detection rate: 14.3%. **Ilana Fisher's detection rate:** 14.3%.
Variants on the Counsyl panel: 5
Gene: PKHD1. **Variants:** Leu1965fs, 9689delA, T36M, R496X, V3471G.

Child Risk

Affected	1 in 20,000
Carrier	1 in 72
Unaffected	98%

What is autosomal recessive polycystic kidney disease?

Autosomal recessive polycystic kidney disease (ARPKD) is an inherited disease in which clusters of fluid-filled sacs (cysts) form in the kidneys, often leading to kidney failure by the age of 10 and a reduced lifespan. According to studies, between 23 and 30% of infants with ARPKD die hours or days after birth due to breathing difficulties.

The majority of infants with ARPKD show enlarged, cyst-filled kidneys within the first month of life. These cysts will impair the kidneys' ability to filter waste from the body. About 50% of infants with the disease will also have an enlarged liver. These anomalies are often detectable through ultrasound before the child is born. More than half of children will develop kidney failure by the age of 10. Without dialysis or transplantation, the disease is often fatal.

A minority of people with ARPKD do not show symptoms of the disease until later in childhood or early in adulthood, with liver disease being the dominant symptom. In these people, the kidney disease is often mild.

Extremely high blood pressure is common in people with ARPKD. They are also prone to urinary tract infections, frequent urination, low blood cell counts, pain in the back or the sides, varicose veins, and hemorrhoids. Many are also smaller than normal in stature.

How common is autosomal recessive polycystic kidney disease?

ARPKD affects 1 in every 15,000 to 40,000 infants. However, the disease may actually be more common since people with milder forms of the disease may not be diagnosed without genetic testing. About 1 in 70 U.S. adults is thought to be a carrier of ARPKD.

How is autosomal recessive polycystic kidney disease treated?

The initial concern with infants who have ARPKD is to protect their ability to breathe. Eating a nutritious diet can help the child's growth, and in some cases, growth hormones are recommended. Infants and children may require feeding tubes in order to ensure proper growth.

If faced with kidney failure, people with ARPKD frequently undergo dialysis (a "cleansing" of the blood through a machine that remove wastes) or kidney transplants. If the liver is extremely damaged, transplantation of this organ may also be recommended. Some people with ARPKD must undergo dialysis or a kidney transplant while they are still in infancy.

In all people with ARPKD, medications can lower blood pressure and clear up urinary tract infections.

What is the prognosis for someone with autosomal recessive polycystic kidney disease?

Between 20 and 30% of infants with ARPKD die hours or days after birth due to breathing difficulties. Of those who survive infancy, about 85% survive their first year of life, 82% survive to age 10, and 73% live past the age of 15. In one study, 42% of those who survived their first year lived to the age of 20.

As transplantation methods improve, it is expected that people with ARPKD will live longer lives.

Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher

Negative Report: Medium Chain Acyl-CoA Dehydrogenase Deficiency

This disease report is included due to Daniel MacArthur and Ilana Fisher's ethnic backgrounds.

Reduced risk	Your child's risk: 1 in 280,000	Risk before testing: 1 in 14,000

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	No disease-causing mutations detected.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270.	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270.

Daniel MacArthur's detection rate: 77.5%. **Ilana Fisher's detection rate:** 77.5%.
Variants on the Counsyl panel: 7
Gene: ACADM. **Variants:** L59F, G170R, G242R, Y42H, K304E, R181C, R181H.

Child Risk

Affected	1 in 280,000
Carrier	1 in 270
Unaffected	Nearly 100%

What is medium chain acyl-CoA dehydrogenase deficiency?

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency is a treatable inherited disease in which the body cannot turn certain fatty acids into energy due to a deficient enzyme. As a result, partially metabolized fatty acids can accumulate in body tissues, causing damage to the brain, liver, and other organs. If treated early and consistently, people with MCAD deficiency can live normal lives.

Children with untreated MCAD deficiency are prone to quick-developing, life-threatening health problems including seizures, breathing problems, brain damage, coma, and death. The liver may also be enlarged. It is thought that a small percentage of sudden infant death syndrome is due to undiagnosed MCAD deficiency.

The first symptoms of the disease usually appear in infancy or early childhood. These include vomiting, lack of energy, and low blood sugar. Rarely, these symptoms do not appear until adulthood. Often the episodes of metabolic crisis can be triggered by long periods without eating or by illness.

Women whose fetuses have MCAD deficiency are more prone to certain pregnancy complications and should speak with their physician.

How common is medium chain acyl-CoA dehydrogenase deficiency?

MCAD deficiency is most common in Caucasians from Northern Europe. In the United States, the disease affects approximately 1 in 17,000 people. Affected Americans are often of Northern European ancestry. The disease is rare among Hispanics, African Americans, Asians, and Native Americans in the United States.

Studies have found high rates of MCAD deficiency in Northern Germany (1 in 4,900) and Southern Germany (1 in 8,500). One study found that Germans and Turks are equally affected.

How is medium chain acyl-CoA dehydrogenase deficiency treated?

The key to treatment for people with MCAD deficiency is to avoid fasting, or long periods without eating. Infants will need to be fed frequently with a special diet low in fat. Consuming cornstarch can provide a sustained release of energy and allow for longer gaps between meals. Certain types of fat should be avoided while high amounts of carbohydrates can be beneficial. If the person is unable to consume food, intravenous glucose may be necessary. People with MCAD deficiency should speak with their medical team to devise a specialized diet.

What is the prognosis for someone with medium chain acyl-CoA dehydrogenase deficiency?

If a person affected by MCAD deficiency is diagnosed early and treated promptly, the prognosis is good. He or she can lead a normal or near-normal life.

Because the metabolic crises caused by the disease can quickly progress from first symptom to death, it is possible for people who remain undiagnosed to die during their first episode.

Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher

Negative Report: Phenylalanine Hydroxylase Deficiency

This disease report is included due to Daniel MacArthur and Ilana Fisher's ethnic backgrounds.

Reduced risk	Your child's risk: 1 in 23,000	Risk before testing: 1 in 10,000

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	No disease-causing mutations detected.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 77.	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 77.

Daniel MacArthur's detection rate: 34.4%. **Ilana Fisher's detection rate:** 34.4%.
Variants on the Counsyl panel: 11
Gene: PAH. **Variants:** IVS-10int-546, IVS12+1G>A, L48S, I65T, R158Q, R252W, R261Q, G272X, R408Q, R408W, Y414C.

Child Risk

Affected	1 in 23,000
Carrier	1 in 77
Unaffected	98%

What is phenylalanine hydroxylase deficiency?

Phenylalanine hydroxylase deficiency is a treatable inherited disease in which the body cannot properly process the amino acid phenylalanine due to a deficient enzyme called phenylalanine hydroxylase. If severe forms of the disease go untreated, the buildup of phenylalanine can be toxic to the brain, causing impaired development and leading to severe and irreversible mental retardation. If treated early and consistently however, people with phenylalanine hydroxylase deficiency can lead completely normal lives.

The disease can be divided into several categories based on the amount of enzyme deficiency: Classic phenylketonuria (PKU), variant PKU, and non-PKU hyperphenylalaninemia (non-PKU HPA). Since the mid-1960s, it has been standard for hospitals in North America to screen newborns for phenylalanine hydroxylase deficiency using a drop of blood obtained from a heel prick. This is now a routine practice in most developed countries.

It can be difficult to predict how severely affected a child will be based on the particular genetic mutations they carry. Children with any form phenylalanine hydroxylase deficiency should be evaluated by a specialist immediately after birth.

Classic PKU

Classic PKU is the most common and severe form, resulting from an absence or near absence of the phenylalanine hydroxylase enzyme.

If PKU is not promptly diagnosed and treated with a special diet, mental retardation will occur, along with a number of other symptoms including a small head, seizures, behavior problems, a "mousy" or "musty" odor, abnormal gait, low bone density, and eczema (a skin condition). These are all avoidable if the proper diet is instituted shortly after birth.

Variant PKU

Variant PKU is an intermediate form of the disease, less severe than classic PKU but more severe than non-PKU HPA. A child with variant PKU is at risk for developing the symptoms associated with classic PKU. Though the symptoms may be milder, there is still a risk for impaired mental development if the child's intake of phenylalanine is not monitored.

Non-PKU Hyperphenylalaninemia

Non-PKU HPA is the mildest form of phenylalanine hydroxylase deficiency. People with non-PKU HPA have a higher level of phenylalanine hydroxylase than do people with classic or variant PKU and are consequently at lower risk for developing brain damage. Some people with non-PKU HPA are able to tolerate a normal diet and do not require treatment. This will vary from person to person and must be determined by a medical professional based on the levels of phenylalanine in the person's blood.

How common is phenylalanine hydroxylase deficiency?

The frequency of carriers and affected individuals in select populations is listed below.

Ethnic Group	Carrier Rate	Affected Rate
Turkish	1 in 26	1 in 2,600
Irish	1 in 33	1 in 4,500
Caucasian American	1 in 50	1 in 10,000
East Asian	1 in 51	1 in 10,000
Finnish	1 in 200	1 in 160,000
Japanese	1 in 200	1 in 160,000
Ashkenazi Jewish	1 in 225	1 in 200,000

How is phenylalanine hydroxylase deficiency treated?

The degree of enzyme deficiency varies among people with phenylalanine hydroxylase deficiency, and therefore the treatment must also be individualized based on the levels of phenylalanine in the blood. An infant with any form of phenylalanine hydroxylase deficiency should be evaluated immediately after birth to determine whether or not he or she requires treatment. A blood test can reveal the amount of functioning phenylalanine hydroxylase in the body and this will indicate the amount of phenylalanine the person can safely consume.

While people with classic PKU must adhere to a strict low-phenylalanine diet, others with milder form of the disease are able to safely consume small amounts of the amino acid. For people with non-PKU HPA, treatment may not even be necessary.

Generally speaking, a diet low in protein and free from phenylalanine is important in preserving mental function in a person with classic PKU. Phenylalanine-free formulas are available for infants. Maintaining appropriate levels of phenylalanine in the brain can be achieved through blood testing and diet adjustment. This must be closely supervised by medical professionals. In most cases, this special diet must be maintained for life.

People with any form of phenylalanine hydroxylase deficiency should be conscious to avoid consuming aspartame, an artificial sweetener that contains phenylalanine.

Women with phenylalanine hydroxylase deficiency who become pregnant must be particularly careful to maintain safe levels of phenylalanine in their own bodies in order to avoid birth defects in their children. Ideally this begins prior to conception.

In late 2007, the medication sapropterin dihydrochloride (brand name: Kuvan) was approved by the FDA for use in people with phenylalanine hydroxylase deficiency. In some, it can enhance the activity of the deficient enzyme and lower levels of phenylalanine in the body, allowing for a relaxation of the dietary restrictions. Some people with the disease do not respond to the drug, however. The people who do respond to this treatment usually have milder forms of the disease.

What is the prognosis for someone with phenylalanine hydroxylase deficiency?

If a person with classic or variant PKU is treated early and consistently for the disease, the prognosis can be excellent. Many people with PKU have gone on to lead normal lives with normal intelligence and lifespan.

If treatment is not begun early or adequately maintained, a person with a more severe form of PKU is at risk for severe and irreversible brain damage.

The prognosis is good for a person with non-PKU HPA. He or she may lead a normal life without treatment.

Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher
[redacted]	

Negative Report: Spinal Muscular Atrophy

This disease report is included due to Daniel MacArthur and Ilana Fisher's ethnic backgrounds.

Reduced risk

Your child's risk:
Less than 1 in 1,000,000

Risk before testing:
1 in 4,800

Patient Results

	Daniel MacArthur	Ilana Fisher
Result:	No disease-causing mutations detected.	No disease-causing mutations detected.
Interpretation:	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 700.	This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 700.

Daniel MacArthur's detection rate: 95.0%. **Ilana Fisher's detection rate:** 95.0%.
Variants on the Counsyl panel: 1
Gene: SMN1. **Variants:** Exon 7 deletion.

Child Risk

Affected	Less than 1 in 1,000,000
Carrier	1 in 700
Unaffected	Nearly 100%

What is spinal muscular atrophy?

Spinal muscular atrophy (SMA) is a disease in which certain nerves in the brain and spinal cord die, impairing the person's ability to move. Called motor neurons, these nerves control our ability to sit up, crawl, and walk. In severe cases, a person will not be able to sit up independently and their breathing and swallowing may also be impaired. In the mildest cases, symptoms begin in adulthood and make independent movement such as walking more difficult, but still possible. There are five main subtypes of spinal muscular atrophy, each described below. It is not always possible to predict which type of SMA a child could have based on the genetic mutation he or she inherits. This is true of the mutation (exon 7 deletion) for which Counsyl tests.

Type 0

Type 0 is the most severe form of SMA. Symptoms can often be seen in the later stages of pregnancy as the fetus is less active than expected. Once born, the infant will have little ability to move and may not be able to breathe and swallow independently. Infants with type 0 SMA often die before the age of 6 months.

Type I - also called Werdnig-Hoffmann disease

Type I is another severe form of the disease. Symptoms develop within the first six months of life. Infants with SMA type I often have trouble breathing and swallowing. Their muscle tone and strength are extremely poor; they cannot sit up without support and will not meet any motor skills milestones. Their intellect, however, is normal. Most children with type I SMA will die before the age of two.

Type II – also called Dubowitz disease

In children with type II SMA, muscle weakness becomes apparent between the ages of 6 and 12 months. When placed in a sitting position, people with type II SMA can usually maintain the position without support, however they often lose this ability by their mid-teens. People with SMA type II cannot stand or walk without assistance. They have poor muscle tone and strength and their fingers usually tremble uncontrollably. Their intelligence is typically normal or above average.

Type III – also called Kugelberg-Welander disease

Type III SMA is a milder form of the disease. Its symptoms begin sometime between the age of one year and early adulthood. As young children, they may fall repeatedly and have trouble walking down stairs. While their muscles are weaker than normal, people with type III SMA can usually stand and walk without assistance, although they may lose this ability later in life. The legs are often more severely affected than the arms.

Type IV

Type IV is the mildest form of spinal muscular atrophy. With this form of the disease, muscle weakness does not begin until one's 20s or 30s, or even later. This weakness is often mild to moderate, and the person can still walk and move independently. These individuals may experience mild to moderate tremors and/or twitching. The disease typically does not diminish lifespan. With all types of SMA, there can be difficulties in sleeping and gaining weight. Frequent pneumonia is common. A curvature of the spine and stiff joints are also common. Women with milder forms the disease have been known to give birth to healthy children, although many of the pregnancies had complications.The disease is caused by a shortage in SMN protein, which helps preserve motor neurons. Without it, the neurons cannot pass messages from the brain to the muscles of the body.

How common is spinal muscular atrophy?

In the United States, 1 in every 6,000 to 10,000 people develop spinal muscular atrophy and 1 in 50 is a carrier of the disease. It has been found in people of every race, but is most common in Caucasians, of whom 1 in 35 is a carrier. Carrier rates for other populations include: Ashkenazi Jews (1 in 41 to 62), Asians (1 in 53), African Americans (1 in 66), and Hispanics (1 in 117). Studies done in specific populations have found carrier rates of 1 in 50 in Germany, 1 in 57 in Italy, and 1 in 62 in China.

How is spinal muscular atrophy treated?

There is no cure for spinal muscular atrophy, however some of its symptoms can be addressed.

For children with the more severe forms of spinal muscular atrophy, mechanical breathing aids may prolong lifespan. In some cases, breathing is more difficult at night, leading to a lack of sleep. In those cases, certain types of respiratory assistance may be helpful. If getting enough nutrition is an issue, some people with SMA have turned to feeding tubes.

Those with milder forms of the disease sometimes choose to have surgery to correct curvature of the spine (scoliosis) or joint problems. In forms of the diseases that are fatal in early childhood, these surgeries are often not done.

What is the prognosis for someone with spinal muscular atrophy?

The prognosis for a person with SMA varies greatly depending on which type of the disease he or she has.

Type 0

The disease is typically fatal between 2 and 6 months of age. These infants do not develop any motor skills expected of infants their age.

Type I

This type of SMA is usually fatal within two years. With mechanical breathing aids, children with type I SMA may live longer. There are a few known cases of SMA type I in which the child survived to adolescence or adulthood.

Type II

With type II SMA, 75% of those affected live to the age of 25. They are often able to sit independently when placed in a sitting position, but lose this ability by their mid-teens.

Type III

People with type III SMA may live a normal lifespan. Many learn to walk independently, though most lose the ability to do so by their 30s or 40s.

Type IV

A normal lifespan is possible for people with type IV SMA. They do not develop symptoms until their 20s or 30s and usually retain the ability to walk independently.

Full Results

Below are the full test results for all diseases on the panel. Noted are the specific genetic mutations for which the patients tested positive or negative. If there was insufficient data to determine the genotype for any variant, this will be noted as "no call." Also listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that their future children could inherit each disease.

ABCC8-Related Hyperinsulinism	Reduced risk	Your child's risk: 1 in 52,000	Risk before testing: 1 in 50,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 110. 1.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 110. 1.5% detection rate.

Variants on the Counsyl panel: 3
Gene: ABCC8. **Variants:** F1388del, V187D, 3992-9G>A.

Achromatopsia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 30,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 670. 87.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 670. 87.0% detection rate.

Variants on the Counsyl panel: 6
Gene: CNGB3. **Variants:** R403Q, E336X, IVS8-3T>G, 819_826del8, T383fs, 886-896del11insT.

Adenosine Monophosphate Deaminase Deficiency	MILD

Daniel MacArthur's results: Detected disease-causing mutations: AMPD1:p.Gln12X (Q12X) heterozygote. Detected non-disease-causing mutations: P48L. This individual is a carrier of adenosine monophosphate deaminase deficiency. Carriers generally do not experience symptoms.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 5,600.

Variants on the Counsyl panel: 2
Gene: AMPD1. **Variants:** Q12X, P48L.

Alkaptonuria	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 3,200. 84.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 3,200. 84.4% detection rate.

Variants on the Counsyl panel: 7
Gene: HGD. **Variants:** G161R, G270R, P230S, S47L, M368V, IVS1-1G>A, IVS5+1G>A.

Alpha-1 Antitrypsin Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 720

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 14,000. Nearly 100% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 14,000. Nearly 100% detection rate.

Variants on the Counsyl panel: 2
Gene: SERPINA1. **Variants:** S allele, Z allele.

Andermann Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 560. 10.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 560. 10.0% detection rate.

Variants on the Counsyl panel: 2
Gene: SLC12A6. **Variants:** Thr813fsX813, R675X.

ARSACS	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 10,000. 95.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 10,000. 95.0% detection rate.

Variants on the Counsyl panel: 2
Gene: SACS. **Variants:** 6594delT, 5254C>T.

Aspartylglycosaminuria	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 6.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 6.0% detection rate.

Variants on the Counsyl panel: 2
Gene: AGA. **Variants:** 199_200delGA, C163S.

Ataxia With Vitamin E Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: TTPA. **Variants:** 744delA.

Ataxia-Telangiectasia	Reduced risk	Your child's risk: 1 in 100,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 0.9% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 0.9% detection rate.

Variants on the Counsyl panel: 1
Gene: ATM. **Variants:** R35X.

Autosomal Recessive Polycystic Kidney Disease	Reduced risk	Your child's risk: 1 in 20,000	Risk before testing: 1 in 15,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72. 14.3% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72. 14.3% detection rate.

Variants on the Counsyl panel: 5
Gene: PKHD1. **Variants:** Leu1965fs, 9689delA, T36M, R496X, V3471G.

Bardet-Biedl Syndrome, BBS1-Related	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 790. 80.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 790. 80.0% detection rate.

Variants on the Counsyl panel: 1
Gene: BBS1. **Variants:** M390R.

Bardet-Biedl Syndrome, BBS10-Related	Reduced risk	Your child's risk: 1 in 340,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 46.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 46.0% detection rate.

Variants on the Counsyl panel: 1
Gene: BBS10. **Variants:** C91fs.

Beta Thalassemia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 250,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 3,000. 91.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 3,000. 91.5% detection rate.

Variants on the Counsyl panel: 35
Gene: HBB. **Variants:** Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, K17X, Q39X, 619 bp deletion, Pro5fs, Gly16fs, Glu6fs, Phe41fs, Lys8fs, Phe71fs, Ser9fs, IVS-II-654, IVS-II-705, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-II-844, IVS-I-1, IVS-I-1, IVS-II-849, IVS-II-849, Gly24 T>A, -30T>A, -88C>T, -28A>G, -29A>G, CAP+1 A>C, -87C>G, Hb C, Hb E, Hb O-Arab.

Biotinidase Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 55,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 730. 86.3% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 730. 86.3% detection rate.

Variants on the Counsyl panel: 7
Gene: BTD. **Variants:** G98:d7i3, A171T, D252G, F403V, Q456H, R538C, D444H.

Bloom Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 2.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 2.8% detection rate.

Variants on the Counsyl panel: 2
Gene: BLM. **Variants:** 2281del6ins7, 2407insT.

Canavan Disease	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,100. 53.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,100. 53.4% detection rate.

Variants on the Counsyl panel: 4
Gene: ASPA. **Variants:** E285A, Y231X, A305E, IVS2-2A>G.

Carnitine Palmitoyltransferase IA Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 6.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 6.8% detection rate.

Variants on the Counsyl panel: 2
Gene: CPT1A. **Variants:** P479L, G710E.

Carnitine Palmitoyltransferase II Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 110,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170,000. Nearly 100% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170,000. Nearly 100% detection rate.

Variants on the Counsyl panel: 13
Gene: CPT2. **Variants:** S38fs, Leu178_Ile186delinsPhe, Q413fs, P50H, S113L, R124X, P227L, R503C, G549D, Q550R, P604S, Y628S, R631C.

Cartilage-Hair Hypoplasia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 2.2% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 2.2% detection rate.

Variants on the Counsyl panel: 2
Gene: RMRP. **Variants:** 262G>T, g.70A>G.

Choroideremia	Reduced risk	Your child's risk: 1 in 200,000	Risk before testing: 1 in 200,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being affected by untested mutations. The post-test risk of being affected is 1 in 100,000. 0.9% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 50,000. 0.9% detection rate.

Variants on the Counsyl panel: 1
Gene: CHM. **Variants:** IVS13+2dupT.

CLN5-Related Neuronal Ceroid Lipofuscinosis	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: CLN5. **Variants:** 2467AT.

Congenital Disorder Of Glycosylation Type Ia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 820. 38.7% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 820. 38.7% detection rate.

Variants on the Counsyl panel: 2
Gene: PMM2. **Variants:** F119L, R141H.

Congenital Disorder Of Glycosylation Type Ib	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: MPI. **Variants:** R295H.

Congenital Finnish Nephrosis	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 1.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 1.6% detection rate.

Variants on the Counsyl panel: 2
Gene: NPHS1. **Variants:** 121_122del, R1109X.

Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher
[redacted]	

Cystic Fibrosis	Increased risk	Your child's risk: 1 in 1,400	Risk before testing: 1 in 3,100

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 340. 91.7% detection rate.

Ilana Fisher's results: Detected disease-causing mutations: CFTR:p.Asp1152His (D1152H) heterozygote. This individual is a carrier of cystic fibrosis. The D1152H mutation is associated with a broad clinical spectrum that can range from asymptomatic to severe. 91.7% detection rate.

Variants on the Counsyl panel: 109

Gene: CFTR. **Variants:** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, G91R, E92X, R117C, Y122X, G178R, L206W, G330X, T338I, R347H, R352Q, S364P, G480C, Q493X, V520F, C524X, S549I, S549N, S549R, Q552X, A559T, P574H, G622D, R709X, K710X, Q890X, R1066C, R1070Q, W1089X, Y1092X, M1101K, D1152H, R1158X, S1196X, W1204X, S1235R, Q1238X, S1251N, S1255X, R1283M, dele2-3 21kb, 3199del6, F311del, 394delTT, 574delA, 663delT, 935delA, 936delTA, 1078delT, 1161delC, 1609delCA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 3171delC, 3667del4, 3821delT, 3876delA, 1288insTA, 2184insA, 2307insA, 2869insG, 3905insT, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1811+1.6kbA>G, 1812-1G>A, 1898+1G>T, 1898+5G>T, 3272-26A>G, 3120G>A, 457TAT>G, 2183AA>G, S549R, W1204X, IVS8-5T, I148T, I506V, F508C.

Cystinosis	Reduced risk	Your child's risk: 1 in 290,000	Risk before testing: 1 in 200,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 16.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 16.6% detection rate.

Variants on the Counsyl panel: 4

Gene: CTNS. **Variants:** 537del21, W138X, L158P, D205N.

Factor V Leiden Thrombophilia	MILD

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 33,000.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 33,000.

Variants on the Counsyl panel: 3

Gene: F5. **Variants:** R506Q, H1299R, D2222G.

Factor XI Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 2.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 2.4% detection rate.

Variants on the Counsyl panel: 4
Gene: F11. **Variants:** E117X, F283L, IVS14+1G>A, IVS14del14.

Familial Dysautonomia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 590. 15.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 590. 15.0% detection rate.

Variants on the Counsyl panel: 3
Gene: IKBKAP. **Variants:** IVS20+6T>C, R696P, P914L.

Familial Mediterranean Fever	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,600. 68.7% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,600. 68.7% detection rate.

Variants on the Counsyl panel: 13
Gene: MEFV. **Variants:** I692del, T267I, F479L, R653H, M680I, M694I, M694V, K695R, V726A, A744S, R761H, P369S, R408Q.

Fanconi Anemia Type C	Reduced risk	Your child's risk: 1 in 620,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 400. 59.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 400. 59.8% detection rate.

Variants on the Counsyl panel: 4
Gene: FANCC. **Variants:** IVS4+4A>T, 322delG, Q13X, R548X.

Fumarase Deficiency	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 670. 25.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 670. 25.0% detection rate.

Variants on the Counsyl panel: 1
Gene: FH. **Variants:** 1431_1433dupAAA.

Galactosemia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 30,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 610. 85.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 610. 85.8% detection rate.

Variants on the Counsyl panel: 10
Gene: GALT. **Variants:** IVS2-2A>G, S135L, T138M, F171S, Q169K, Q188R, L195P, Y209C, K285N, X380R.

Gaucher Disease	Reduced risk	Your child's risk: 1 in 240,000	Risk before testing: 1 in 50,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 390. 70.8% detection rate.

Ilana Fisher's results: No mutations detected. No call for L444P and IVS2+1G>A. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 28.7% detection rate.

Variants on the Counsyl panel: 9
Gene: GBA. **Variants:** N370S, L444P, 1035insG, IVS2+1G>A, V394L, D409V, R463C, R463H, R496H.

GJB2-Related DFNB 1 Nonsyndromic Hearing Loss And Deafness	Reduced risk	Your child's risk: 1 in 24,000	Risk before testing: 1 in 7,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 79. 46.2% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 79. 46.2% detection rate.

Variants on the Counsyl panel: 11
Gene: GJB2. **Variants:** 35delG, 167delT, 313del14, E120del, W24X, V37I, W77R, W77X, Q124X, R184P, M34T.

Glucose-6-Phosphate Dehydrogenase Deficiency — MILD

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being affected by untested mutations. The post-test risk of being affected is 1 in 420.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 210.

Variants on the Counsyl panel: 5
Gene: G6PD. **Variants:** V68M, S188F, R459P, R459L, N126D.

Glutaric Acidemia Type 1	Reduced risk	Your child's risk: 1 in 53,000	Risk before testing: 1 in 40,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120. 13.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120. 13.4% detection rate.

Variants on the Counsyl panel: 2
Gene: GCDH. **Variants:** R402W, A421V.

Glycogen Storage Disease Type Ia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 130,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 730. 75.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 730. 75.8% detection rate.

Variants on the Counsyl panel: 10
Gene: G6PC. **Variants:** 727G>T, F327del, Q27fsdelC, 459insTA, R83H, R83C, G188R, Q242X, G270V, Q347X.

Glycogen Storage Disease Type Ib	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 500,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 690. 48.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 690. 48.4% detection rate.

Variants on the Counsyl panel: 4
Gene: G6PT1. **Variants:** 1211delCT, G339C, G339D, A367T.

Glycogen Storage Disease Type III	Reduced risk	Your child's risk: 1 in 120,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170. 8.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170. 8.4% detection rate.

Variants on the Counsyl panel: 3
Gene: AGL. **Variants:** Q6X, 17delAG, 1484delT.

Glycogen Storage Disease Type V	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 68.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 68.8% detection rate.

Variants on the Counsyl panel: 4
Gene: PYGM. **Variants:** R49X, G204S, K542T, K542X.

GRACILE Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: BCS1L. **Variants:** S78G.

Hereditary Fructose Intolerance	Reduced risk	Your child's risk: 1 in 250,000	Risk before testing: 1 in 26,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 67.9% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 67.9% detection rate.

Variants on the Counsyl panel: 4
Gene: ALDOB. **Variants:** Delta4E4, A149P, Y204X, N334K.

Hereditary Thymine-Uraciluria	Reduced risk	Your child's risk: 1 in 54,000	Risk before testing: 1 in 40,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120. 14.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120. 14.0% detection rate.

Variants on the Counsyl panel: 1
Gene: DPYD. **Variants:** IVS14+1G>A.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.6% detection rate.

Variants on the Counsyl panel: 1
Gene: LAMA3. **Variants:** R650X.

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,000. 51.7% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,000. 51.7% detection rate.

Variants on the Counsyl panel: 5
Gene: LAMB3. **Variants:** 3024delT, R42X, R144X, Q243X, R635X.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.3% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.3% detection rate.

Variants on the Counsyl panel: 1
Gene: LAMC2. **Variants:** R95X.

Hexosaminidase A Deficiency

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 330.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 330.

Variants on the Counsyl panel: 8
Gene: HEXA. **Variants:** 1278insTATC, IVS12+1G>C, R178C, R178H, G269S, IVS7+1G>A, IVS9+1G>A, R247W.

HFE-Associated Hereditary Hemochromatosis

MILD

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 340.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 340.

Variants on the Counsyl panel: 11
Gene: HFE. **Variants:** H63D, S65C, Q127H, E168Q, E168X, W169X, C282Y, Q283P, V53M, V59M, H63H.

Homocystinuria Caused By Cystathionine Beta-Synthase Deficiency

Reduced risk

Your child's risk: 1 in 160,000

Risk before testing: 1 in 63,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 38.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 38.0% detection rate.

Variants on the Counsyl panel: 2
Gene: CBS. **Variants:** I278T, G307S.

Hurler Syndrome

Reduced risk

Your child's risk: 1 in 280,000

Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 40.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 40.5% detection rate.

Variants on the Counsyl panel: 2
Gene: IDUA. **Variants:** A327P, W402X.

Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.5% detection rate.

Variants on the Counsyl panel: 1
Gene: SLC25A15. **Variants:** F188del.

Hypophosphatasia, Autosomal Recessive	Reduced risk	Your child's risk: 1 in 170,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 22.2% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 22.2% detection rate.

Variants on the Counsyl panel: 5
Gene: ALPL. **Variants:** 1559delT, F310L, D361V, E174K, G317D.

Inclusion Body Myopathy 2	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 1.8% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510. 1.8% detection rate.

Variants on the Counsyl panel: 1
Gene: GNE. **Variants:** M712T.

Infantile Refsum Disease	Reduced risk	Your child's risk: 1 in 160,000	Risk before testing: 1 in 50,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 43.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 43.1% detection rate.

Variants on the Counsyl panel: 1
Gene: PEX1. **Variants:** G843D.

Isovaleric Acidemia	Reduced risk	Your child's risk: 1 in 890,000	Risk before testing: 1 in 250,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 470. 47.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 470. 47.0% detection rate.

Variants on the Counsyl panel: 1
Gene: IVD. **Variants:** A311V.

Krabbe Disease	Reduced risk	Your child's risk: 1 in 89,000	Risk before testing: 1 in 25,000

Daniel MacArthur's results: Detected non-disease-causing mutations: R168C. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150. 46.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150. 46.5% detection rate.

Variants on the Counsyl panel: 3
Gene: GALC. **Variants:** Ex11-17del, G270D, R168C.

Leigh Syndrome, French-Canadian Type	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: LRPPRC. **Variants:** A354V.

Limb-Girdle Muscular Dystrophy Type 2E	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 550. 9.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 550. 9.1% detection rate.

Variants on the Counsyl panel: 1
Gene: SGCB. **Variants:** S114F.

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Reduced risk	Your child's risk: 1 in 110,000	Risk before testing: 1 in 90,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 8.3% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 8.3% detection rate.

Variants on the Counsyl panel: 2
Gene: HADHA. **Variants:** Q342X, E474Q.

Maple Syrup Urine Disease Type 1B	Reduced risk	Your child's risk: 1 in 700,000	Risk before testing: 1 in 250,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 420. 40.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 420. 40.1% detection rate.

Variants on the Counsyl panel: 3
Gene: BCKDHB. **Variants:** R183P, G278S, E322X.

Maple Syrup Urine Disease Type 3	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 560. 10.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 560. 10.0% detection rate.

Variants on the Counsyl panel: 2
Gene: DLD. **Variants:** 105insA, G229C.

Medium Chain Acyl-CoA Dehydrogenase Deficiency	Reduced risk	Your child's risk: 1 in 280,000	Risk before testing: 1 in 14,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 77.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 77.5% detection rate.

Variants on the Counsyl panel: 7
Gene: ACADM. **Variants:** L59F, G170R, G242R, Y42H, K304E, R181C, R181H.

Metachromatic Leukodystrophy	Reduced risk	Your child's risk: 1 in 130,000	Risk before testing: 1 in 40,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 180. 44.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 180. 44.0% detection rate.

Variants on the Counsyl panel: 4
Gene: ARSA. **Variants:** P377L, P426L, IVS2+1G>A, T274M.

MTHFR Deficiency	MILD

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 320.

Ilana Fisher's results: Detected disease-causing mutations: MTHFR:p.Glu429Ala (E429A) heterozygote. This individual is a carrier of MTHFR deficiency. Carriers generally do not experience symptoms.

Variants on the Counsyl panel: 2
Gene: MTHFR. **Variants:** A222V, E429A.

Mucolipidosis IV	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. No call for 511_6944del. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.5% detection rate.

Ilana Fisher's results: No mutations detected. No call for 511_6944del. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.5% detection rate.

Variants on the Counsyl panel: 2
Gene: MCOLN1. **Variants:** 511_6944del, IVS3-2A>G.

Muscle-Eye-Brain Disease	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 690. 27.7% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 690. 27.7% detection rate.

Variants on the Counsyl panel: 1
Gene: POMGNT1. **Variants:** IVS17+1G>A.

MYH-Associated Polyposis	Reduced risk	Your child's risk: 1 in 68,000	Risk before testing: 1 in 40,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 23.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 23.1% detection rate.

Variants on the Counsyl panel: 1
Gene: MUTYH. **Variants:** Y165C.

Niemann-Pick Disease Type A	Reduced risk	Your child's risk: 1 in 260,000	Risk before testing: 1 in 250,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 260. 2.7% detection rate.

Ilana Fisher's results: No mutations detected. No call for fsP330. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 260. 1.8% detection rate.

Variants on the Counsyl panel: 3
Gene: SMPD1. **Variants:** fsP330, L302P, R496L.

Niemann-Pick Disease Type C	Reduced risk	Your child's risk: 1 in 190,000	Risk before testing: 1 in 150,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 220. 10.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 220. 10.0% detection rate.

Variants on the Counsyl panel: 1
Gene: NPC1. **Variants:** I1061T.

Nijmegen Breakage Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 720. 78.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 720. 78.0% detection rate.

Variants on the Counsyl panel: 1
Gene: NBN. **Variants:** 657del5.

Northern Epilepsy	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: CLN8. **Variants:** R24G.

Pendred Syndrome	Reduced risk	Your child's risk: 1 in 64,000	Risk before testing: 1 in 20,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 44.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 44.0% detection rate.

Variants on the Counsyl panel: 3
Gene: SLC26A4. **Variants:** L236P, E384G, T416P.

Phenylalanine Hydroxylase Deficiency	Reduced risk	Your child's risk: 1 in 23,000	Risk before testing: 1 in 10,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 77. 34.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 77. 34.4% detection rate.

Variants on the Counsyl panel: 11
Gene: PAH. **Variants:** IVS-10int-546, IVS12+1G>A, L48S, I65T, R158Q, R252W, R261Q, G272X, R408Q, R408W, Y414C.

Polyglandular Autoimmune Syndrome Type 1	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 660. 23.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 660. 23.6% detection rate.

Variants on the Counsyl panel: 2
Gene: AIRE. **Variants:** Y85C, R257X.

PPT1-Related Neuronal Ceroid Lipofuscinosis	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,600. 69.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 1,600. 69.6% detection rate.

Variants on the Counsyl panel: 4
Gene: PPT1. **Variants:** L10X, T75P, R122W, R151X.

Primary Hyperoxaluria Type 1	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 500,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 34.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 34.0% detection rate.

Variants on the Counsyl panel: 3
Gene: AGXT. **Variants:** F152I, G170R, I244T.

Primary Hyperoxaluria Type 2	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 790. 37.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 790. 37.0% detection rate.

Variants on the Counsyl panel: 1
Gene: GRHPR. **Variants:** 103delG.

Pycnodysostosis	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 3.6% detection rate.

Variants on the Counsyl panel: 1
Gene: CTSK. **Variants:** X330W.

Rhizomelic Chondrodysplasia Punctata Type 1	Reduced risk	Your child's risk: 1 in 550,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 370. 57.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 370. 57.1% detection rate.

Variants on the Counsyl panel: 2
Gene: PEX7. **Variants:** G217R, L292X.

Salla Disease	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 6.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 540. 6.6% detection rate.

Variants on the Counsyl panel: 2
Gene: SLC17A5. **Variants:** Leu336fsX13, R39C.

Segawa Syndrome	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.2% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 520. 4.2% detection rate.

Variants on the Counsyl panel: 1
Gene: TH. **Variants:** R233H.

Short Chain Acyl-CoA Dehydrogenase Deficiency	Reduced risk	Your child's risk: 1 in 100,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: Detected non-disease-causing mutations: G185S. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 2.3% detection rate.

Ilana Fisher's results: Detected non-disease-causing mutations: G185S. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. 2.3% detection rate.

Variants on the Counsyl panel: 2
Gene: ACADS. **Variants:** R107C, G185S.

Sickle Cell Disease	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 20,000. 97.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 20,000. 97.5% detection rate.

Variants on the Counsyl panel: 37

Gene: HBB. **Variants:** Hb S, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, K17X, Q39X, 619 bp deletion, Pro5fs, Gly16fs, Glu6fs, Phe41fs, Lys8fs, Phe71fs, Ser9fs, IVS-II-654, IVS-II-705, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-II-844, IVS-I-1, IVS-I-1, IVS-II-849, IVS-II-849, Gly24 T>A, -30T>A, -88C>T, -28A>G, -29A>G, CAP+1 A>C, -87C>G, Hb C, Hb E, Hb D-Punjab, Hb O-Arab.

Sjogren-Larsson Syndrome	Reduced risk	Your child's risk: 1 in 430,000	Risk before testing: 1 in 250,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 330. 24.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 330. 24.0% detection rate.

Variants on the Counsyl panel: 1

Gene: ALDH3A2. **Variants:** P315S.

Smith-Lemli-Opitz Syndrome	Reduced risk	Your child's risk: 1 in 250,000	Risk before testing: 1 in 40,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 59.5% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 59.5% detection rate.

Variants on the Counsyl panel: 11

Gene: DHCR7. **Variants:** IVS8-1G>C, T93M, L109P, W151X, L157P, V326L, R352Q, R352W, C380Y, R404C, W151X.

Spinal Muscular Atrophy	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 4,800

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 700. 95.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 700. 95.0% detection rate.

Variants on the Counsyl panel: 1
Gene: SMN1. **Variants:** Exon 7 deletion.

Sulfate Transporter-Related Osteochondrodysplasia	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 3,600

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 46,000. Nearly 100% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 46,000. Nearly 100% detection rate.

Variants on the Counsyl panel: 5
Gene: SLC26A2. **Variants:** C653S, R178X, R279W, V340del, IVS1+2T>C.

Tay-Sachs Disease	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 360,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 570. 47.6% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 570. 47.6% detection rate.

Variants on the Counsyl panel: 4
Gene: HEXA. **Variants:** 1278insTATC, IVS12+1G>C, IVS7+1G>A, IVS9+1G>A.

TPP1-Related Neuronal Ceroid Lipofuscinosis	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: 1 in 350,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 800. 63.1% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 800. 63.1% detection rate.

Variants on the Counsyl panel: 4
Gene: TPP1. **Variants:** G284V, R208X, IVS5-1G>C, IVS5-1G>A.

Tyrosinemia Type I	Reduced risk	Your child's risk: 1 in 220,000	Risk before testing: 1 in 120,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 240. 26.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 240. 26.4% detection rate.

Variants on the Counsyl panel: 5
Gene: FAH. **Variants:** IVS8-1G>C, IVS12+5G>A, P261L, W262X, E357X.

Usher Syndrome Type 1F	Reduced risk	Your child's risk: 1 in 160,000	Risk before testing: 1 in 150,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 3.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 3.4% detection rate.

Variants on the Counsyl panel: 1
Gene: PCDH15. **Variants:** R245X.

Usher Syndrome Type 3	Reduced risk	Your child's risk: Less than 1 in 1,000,000	Risk before testing: Less than 1 in 1,000,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 530. 5.0% detection rate.

Variants on the Counsyl panel: 1
Gene: CLRN1. **Variants:** N48K.

Wilson Disease	Reduced risk	Your child's risk: 1 in 91,000	Risk before testing: 1 in 30,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150. 42.3% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150. 42.3% detection rate.

Variants on the Counsyl panel: 5
Gene: ATP7B. **Variants:** 1340del4, 2337delC, R778G, W779X, H1069Q.

Counsyl

Male	Female
Name: Daniel MacArthur	Name: Ilana Fisher
[redacted]	

X-Linked Juvenile Retinoschisis	Reduced risk	Your child's risk: 1 in 120,000	Risk before testing: 1 in 100,000

Daniel MacArthur's results: No mutations detected. This does not rule out the possibility of being affected by untested mutations. The post-test risk of being affected is 1 in 68,000. 14.4% detection rate.

Ilana Fisher's results: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 29,000. 14.4% detection rate.

Variants on the Counsyl panel: 3

Gene: RS1. **Variants:** E72K, G74V, G109R.