Preconception Genetic Screening Intent Form

Patient Name: ____________________________ Date of Birth: ____________________________

☐ I would like to have genetic screening for the following disorders:

UCRM recommends the following testing (please see the following pages for more information):

For all ethnic groups (including Caucasian):
☐ Spinal Muscular Atrophy (SMA)
☐ Cystic Fibrosis (CF)

For Ashkenazi Jewish:
☐ Ashkenazi Jewish Panel

For French Canadian/Cajun:
☐ Tay Sachs disease

For African American
☐ West African
☐ Hispanic
☐ Mediterranean
☐ South-East Asian
☐ Western Pacific region
☐ Middle Eastern
☐ Caribbean
☐ South American
☐ Other, please specify: _________________________

☐ Hemoglobinopathies (please circle your ethnic group(s) above, so that we may order the correct test)

Other options:
☐ Counsyl (tests for 100+ conditions, including all of the above)
☐ Other, please specify: _________________________

If you selected any of these options, our office will contact you with the appropriate paper work and consent forms.

☐ I would like to talk more with my doctor or genetic counselor before making a decision.

You may wish to select this option if you checked “yes” to any of the questions on the “Personal Medical History/Genetic Questionnaire,” are of an ethnic background not listed above, or have any questions not addressed in this packet. If you selected this option, please contact our office to arrange a consultation. Or, you may discuss testing options with your OB or local genetic counselor.

☐ I do not want genetic screening.

I have been provided an information packet explaining genetic screening. I understand that a baby I have could be at risk for a genetic disorder. Genetic screening could help us determine what that risk is. There may be reproductive options that would help us reduce our risk of having a child with a genetic disorder.

_________________________  _________________________
Signature                                      Date
Preconception Genetic Screening - FAQ

Who is at risk of having a baby with a genetic disorder?

Everyone. Humans have over 40,000 genes. The average person has about three “broken” genes that may give rise to a child with a genetic disorder. Even if no one in your family has a genetic disorder, you could still have a baby with one.

What causes genetic disorders?

Many genetic disorders are recessive disorders. In order for a recessive genetic disorder to occur, a baby must inherit a broken gene from both parents. The parents usually do not know that they are carriers for the disorder because they only have one broken gene. People with only one broken gene (a carrier) typically do not have symptoms.

What is my chance of being a carrier?

The chance that you carry a broken gene that can cause a recessive genetic disorder is related to your ethnic background. The following table lists some of the more common diseases and carrier frequencies in different ethnic groups. If you have no one in your family affected with these condition, this table lists your chance of being a carrier. If you have someone your family affected with one of these conditions, your chance of being a carrier is higher. For descriptions of these conditions, please see the attached “Summary of Common Genetic Disorders.”

<table>
<thead>
<tr>
<th>Ethnic Group</th>
<th>Disease</th>
<th>Chance you are a carrier (i.e. have one broken gene)</th>
</tr>
</thead>
<tbody>
<tr>
<td>All ethnic groups</td>
<td>Cystic Fibrosis</td>
<td>1 in 25 – 90 (1 – 4%)</td>
</tr>
<tr>
<td></td>
<td>SMA</td>
<td>1 in 40 – 60 (2.5 – 1.5%)</td>
</tr>
<tr>
<td>Ashkenazi Jewish see “Ashkenazi Jewish Panel” in the Summary of Common Genetic Disorders. The panel includes all of these conditions except Cystic Fibrosis.</td>
<td>Gaucher Disease</td>
<td>1 in 15 (7%)</td>
</tr>
<tr>
<td></td>
<td>Tay Sachs Disease</td>
<td>1 in 30 (3%)</td>
</tr>
<tr>
<td></td>
<td>Familial Dysautonomia</td>
<td>1 in 30 (3%)</td>
</tr>
<tr>
<td></td>
<td>Canavan Disease</td>
<td>1 in 40 (2.5%)</td>
</tr>
<tr>
<td></td>
<td>Fanconi Anemia Group C</td>
<td>1 in 89 (1%)</td>
</tr>
<tr>
<td></td>
<td>Niemann-Pick Type-A</td>
<td>1 in 90 (1%)</td>
</tr>
<tr>
<td></td>
<td>Bloom</td>
<td>1 in 100 (1%)</td>
</tr>
<tr>
<td></td>
<td>Mucolipidosis IV</td>
<td>1 in 127 (0.8%)</td>
</tr>
<tr>
<td>African American/West African</td>
<td>Hemoglobinopathy</td>
<td>1 in 6 – 75 (1 – 16%)</td>
</tr>
<tr>
<td>Hispanic</td>
<td>Hemoglobinopathy</td>
<td>1 in 30 – 50 (2 – 3%)</td>
</tr>
<tr>
<td>Mediterranean(^1) /Asian (Western Pacific Region)(^2)/Middle Eastern(^3)</td>
<td>Hemoglobinopathy</td>
<td>1 in 20 – 40 (2.5 – 5%)</td>
</tr>
<tr>
<td>South East Asian(^4)</td>
<td>Hemoglobinopathy</td>
<td>1 in 4 – 30 (3 – 25%)</td>
</tr>
<tr>
<td>French/Cajun</td>
<td>Tay Sachs Disease</td>
<td>1 in 30 (3%)</td>
</tr>
</tbody>
</table>

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\(^1\) Mediterranean: e.g., Sardinia, Corsica, Sicily, Italy, Spain, Portugal, Greece, Cyprus, Turkey, Egypt, Algeria, Libya, Tunisia, Morocco, Malta

\(^2\) Asian (Western Pacific region): e.g., China, Vietnam, Philippines, Malaysia, Cambodia, Laos

\(^3\) Middle Eastern: e.g., Iran, Iraq, Syria, Jordan, Saudi Arabia and other Arabian peninsula countries, Qatar, Lebanon, Palestine, Israel (both Arabs and Sephardic Jews affected), Kuwait

\(^4\) South-East Asian: e.g., India, Afghanistan, Pakistan, Indonesia, Bangladesh, Thailand, Myanmar
If I’m a carrier, will my baby have a genetic disorder?

For most of these disorders, if both you and your partner are carriers, you have a 1 in 4 (25%) chance of having a baby with the disorder. However, for some disorders, a person with a broken gene has up to a 1 in 2 (50%) chance of having a baby with the disorder. Your doctor or a genetic counselor can help you determine your risk of having a baby with a specific condition.

Can I find out if I am a carrier?

Genetic screening tests can be done to tell us if you are a carrier for the more common genetic disorders and are at a higher risk for having a baby with one of these genetic disorders. You can either be tested for each condition individually or opt for a panel of tests. The genetic panels can test for several disorders to over 100 disorders, depending on the panel.

If I am a carrier for a genetic disorder, is there anything we can do?

First, for recessive conditions (where the baby needs to get a broken gene from both parents) we would test your partner to see if your partner also has the same broken gene. If only one of you is found to have the broken gene, the risk to the baby is greatly reduced.

Second, if the baby is at higher risk for a genetic disorder, there are reproductive options available. Your doctor will be able to help you choose the one that is right for you and your family.

Will my insurance company cover genetic screening?

Maybe. You can contact your insurance company and ask them if they cover genetic testing. Attached to this packet is a list of “CPT codes.” These are the codes that insurance companies use to describe certain tests. You can give them the CPT code and ask if it is covered. If they ask for a diagnosis code or an “ICD-9” code, it depends on who is being tested. If a female partner is being screened, tell them it is “v26.31.” If a male partner is being screened, tell them it is “v26.34.”

If genetic screening isn’t covered by my insurance, can you tell me how much it costs?

Usually, no. The laboratory that does the testing charges a certain price. However, the place where you have your blood drawn will charge you a different price that covers their costs. They also may have contracts with your insurance company that specifies what you will be charged. Depending on your insurance, that price will vary.

What are these genetic disorders?

Please see the attached sheet: Summary of common genetic disorders.

What is Counsyl?

Counsyl is one option for genetic screening. Counsyl is a private company that provides genetic screening for over 100 conditions. Some of these conditions are very rare and screening for them is not recommended by any medical organization. However, they do screen for all of the recommended conditions, except Fragile X. You can obtain a kit from our office. You spit into the kit’s tube, seal it, and mail it back to Counsyl. Your results are available online within 2 to 3 weeks. You can find their website at http://www.counsyl.com. Your insurance company may cover Counsyl.
CPT Codes

If you would like to check with your insurance to see if they cover genetic screening, these are the CPT codes that your insurance company will ask for. If they ask for a diagnosis code or an “ICD-9” code, it depends on who is being tested. If a female partner is being screened, tell them it is “v26.31.” If a male partner is being screened, tell them it is “v26.34.” If you are interested in screening for a disorder not found on this list, please contact us and we will give you the appropriate CPT code. If your insurance company won’t cover these CBT codes or needs pre-authorization, please contact us for further options.

Jewish Panel -- does not include Cystic Fibrosis

Includes:
- Gaucher Disease
- Tay Sachs Disease
- Familial Dysautonomia
- Canavan Disease
- Fanconi Anemia Group C
- Niemann-Pick Type-A
- Bloom
- Mucolipidosis IV

CPT Codes: 83891, 83892, 83900, 83901, 83914, 83909, 83912

Cystic Fibrosis

CPT Codes: 83891, 83900, 83901, 83914, 83909, 83912

Tay-Sachs

CPT Code: 83080

Hemoglobinopathies

Includes:
- Sickle Cell
- Thalassemia (beta & alpha)
- Other hemoglobin diseases (such as Hemoglobin E or Hemoglobin C)

CPT Codes: 83891, 83898, 83896, 83912, 83021, 83900, 83901, 83894, 83904, 83909, 85025, 83020, 85660

Spinal Muscular Atrophy

CPT Codes: 83891, 83892, 83898, 83912

Counsyl

Contact Counsyl directly for insurance coverage questions.