Telephone Disclosure Visual Aid Toolkit

This is your visual aid toolkit that will be used during your disclosure appointment.

Included in this packet are definitions and descriptions of things that will be discussed during your disclosure.

PASSWORD (For Telephone Disclosures Only): _______

Please remember, if you will be receiving your results by telephone, you will be asked for your password at the start of your telephone disclosure.

Please remember to have your visual aids available IF you are receiving your results by telephone.

It is very important that you meet with your genetics team at your in-person visit to discuss this further and receive more detailed medical recommendations.

Version Date: 12 10 2014
Genetic Testing Options (Discussed During Pre-Test Counseling Session)

**Single Syndrome Testing**
- Test 1-5 syndrome specific genes for mutations
- Tests for targeted hereditary cancer syndrome(s)

**Gene Panel Testing**
- Test multiple genes at the same time for mutations
- Tests for multiple, different hereditary cancer syndromes

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What will be discussed during your disclosure:

- If you are randomized to receive your genetic test results by telephone, your genetic counselor will ask you the following:
  - To confirm your identity using a phrase that you can select.
  - If you can hear them alright
  - If you have a pen and paper available to take notes.
  - If you have this packet available as they might refer to it during your disclosure.

- For both in-person and telephone disclosures, after verifying that you want your results, your genetic counselor will discuss the following:
  - Genetic Test Results
  - Interpretation of Results
  - Implication(s) of Results
  - Implication(s) of Results for your Family
  - Ask you to relay back to them you're understanding of your results
  - Provide you with any referrals you might need
  - Remind you that you’ll be receiving another survey after you’ve been disclosed.
Possible Results

• Positive for a mutation in a cancer gene:
  • May be associated with a **high risk** for one or more cancers
  • May be associated with a **moderate risk** for one or more cancers
  • May be associated with an **undetermined risk** for one or more cancers

• No mutation detected
  • **True Negative**: A *mutation* has already been found in another family member, and the person being tested has not inherited that *mutation*.
  • **Uninformative Negative**: No *mutation* has been found in. This is different from a True Negative result because no other family member has been tested and found to have a *mutation*.

• Variant of uncertain significance (VUS)
  • A genetic change is found that may or may not increase the risk for cancer
  • This result is **not positive or negative**
  • More information is needed before this information can help guide care
Possible Results Continued: Result Interpretation

- **No Mutation (Negative)**
- **VUS-Likely Benign**
- **Uncertain Significance (VUS)**
- **VUS-Likely Pathogenic**
- **Pathogenic (Positive)**

**Medical management based on personal and family history. Uncertain results do not influence recommendations for care.**

**Medical management based on cancer risks linked with gene where mutation found.**

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# Understanding a Positive Test Result: Cancer Risk

<table>
<thead>
<tr>
<th>Lifetime Risk for</th>
<th>High Risk</th>
<th>Moderate Risk</th>
<th>Unclear Risk</th>
<th>Average Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast Cancer</td>
<td>&gt;30%</td>
<td>15-30%</td>
<td>Studies suggest an increased risk, but exact risk is unclear</td>
<td>10-12%</td>
</tr>
<tr>
<td>Ovarian Cancer</td>
<td>&gt;10%</td>
<td>2-10%</td>
<td>Studies suggest an increased risk, but exact risk is unclear</td>
<td>~1.5%</td>
</tr>
<tr>
<td>Uterine Cancer</td>
<td>&gt;15%</td>
<td>NA</td>
<td>Studies suggest an increased risk, but exact risk is unclear</td>
<td>~2.7%</td>
</tr>
<tr>
<td>Colon Cancer</td>
<td>&gt;30%</td>
<td>NA</td>
<td>Studies suggest an increased risk, but exact risk is unclear</td>
<td>5-6%</td>
</tr>
<tr>
<td>Pancreatic Cancer</td>
<td>&gt;5%</td>
<td>NA</td>
<td>Studies suggest an increased risk, but exact risk is unclear</td>
<td>&lt;1%</td>
</tr>
</tbody>
</table>

**NOTES:**

- Different research studies may report different cancer risks, depending on how the study was conducted. For this reason, it is often most helpful to think about cancer risk as a range, which may vary from family to family.
- Percentages may change over time as more studies are conducted.
Understanding a Positive Result
Continued

• This result means that your chances of developing certain cancers may be higher than the average person.

• In general, management of this result will depend on the gene and cancer risk. Recommendations may include:
  • Screening/Surveillance
    • More frequent screening tests
    • Different kinds of screening tests
  • Medications (Chemoprevention)
  • Risk Reducing (Prophylactic) Surgeries

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Understanding a Positive Result
Continued

When to consider these options may be based on age, family history, and/or genetic testing results.

In some cases there is strong evidence and clear management recommendations, in others there may be lack of consensus or limited information.

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Understanding an Uninformative Negative Test Result

- No mutation(s) were found in the genes which were tested as part of your testing.

- Further testing of additional family members may help clarify results.

- People with this type of result are given recommendations based on their personal and family history of cancer.

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Understanding a True Negative Test Result

- Individuals with a True Negative test result have not inherited a mutation that was previously found in a relative.

- Because there is no mutation, an individual with a True Negative test result has a risk of developing cancer similar to that of the general population.

- Most often, individuals who test True Negative are recommended to continue using the same cancer screening recommendations that are given to the general population.
Understanding Variant of Uncertain Significance Test Result

A Variant of Uncertain Significance (VUS) is an alteration that cannot be interpreted at this time.

- There is not enough known about a VUS to classify it as benign (harmless) or as mutation (associated with cancer risk)
  - May or may not cause increased cancer risk

- Because the meaning of a VUS is not fully understood, this result should not be used to test relatives for cancer risk
  - Depending on the type of VUS result, relatives may be asked for blood and/or tumor samples to help further understand and study the VUS

- Over time, more information may be learned about this VUS.
  - It is recommended that you check back with your genetics team periodically to learn whether this result has been updated

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DEFINITIONS

- **Alteration** - A change in the usual DNA sequence of a particular gene. These can be harmful, beneficial, or neutral in their effect on cell function.

- **Cancer Screening** - Checking for disease when there are no symptoms. Since screening may find diseases at an early stage, there may be a better chance of curing the disease. Examples of cancer screening tests are the mammogram (breast), colonoscopy (colon), and the Pap test and HPV test (cervix). Screening can also include checking for a person's risk of developing an inherited disease by doing a genetic test.

- **Chemoprevention** - The use of drugs, vitamins, or other agents to try to reduce the risk of, or delay the development or recurrence of, cancer.

- **Disclosure** - Another term used to describe the appointment when you receive your test results.

- **First Degree Blood Relatives** - Mother, Father, Sister, Brother, Children

- **Genes** - The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.

- **Inherited** - Transmitted through genes that have been passed from parents to their offspring (children).

- **Mutation** - Any change in the DNA of a cell. Mutations may be caused by mistakes during cell division, or they may be caused by exposure to DNA-damaging agents in the environment. Mutations can be harmful, beneficial, or have no effect. If they occur in cells that make eggs or sperm, they can be inherited; if mutations occur in other types of cells, they are not inherited. Certain mutations may lead to cancer or other diseases.

- **Pedigree** - A diagram that shows relationships among family members. In medicine, a pedigree may also show the pattern of certain genes or diseases within a family.

- **Prophylactic** – Something that helps prevent or protect against certain types of cancers developing.