ExomeNext

A GUIDE TO EXOME SEQUENCING
What is DNA?

DNA (Deoxyribonucleic acid) is a molecule that carries genetic information in the form of a code. It is found in the nucleus of every cell. The code is composed of four types of nucleotide bases: adenine (A), cytosine (C), guanine (G), and thymine (T). These bases pair with each other in a specific way, forming the double-stranded structure of DNA.
What is the Exome?
Why is the Exome Important?

Watermelon = Genome

Slice = Exome 1-2%

Seeds = Genes
What Do Genes Do?

1. Inside our cells, we have genes.

2. Genes are like "recipes" that tell our bodies how to make proteins.

3. These proteins help our bodies work properly.

4. Mutations in our genes can change these proteins, which can affect any part of our bodies.

Recipe: A-G-C-T
What is a Mutation?

- Genetic Code: A-C-G-T
- Misspelled: A-C-T-G
- Misspelled: A-C-G-T
- Deleted: A-C--T
- Duplicated: A-C-C-G-T
How is ExomeNext Done?

1. Blood Sample
2. Extract DNA
3. Sequence DNA
4. Analysis
5. Results
Testing Family Members

- NOT AFFECTED
- NOT AFFECTED
- AFFECTED
- NOT AFFECTED
- NOT AFFECTED
Is ExomeNext Right For You?

ExomeNext might be something to consider if your or your family member's medical concern could be due to an underlying genetic change, but:

- Your previous genetic testing did not find an answer (or diagnosis)
- There is no specific genetic testing available for the genetic condition you may have
- Your medical concerns may be caused by changes in more than one gene
What Are the Benefits of ExomeNext?

- Quicker and more cost-effective since it studies multiple genes at once
- Can suggest changes to treatment or medical care
- Removes feelings of uncertainty related to having an undiagnosed disease
- Provides awareness about health concerns to your other family members
Things to Consider Before Testing

LIMITATIONS

PRIVACY

SECONDARY FINDINGS
Supplementary Information
<table>
<thead>
<tr>
<th>RESULT TYPE</th>
<th>OVERALL RESULT CATEGORY</th>
<th>RESULT MEANING</th>
</tr>
</thead>
<tbody>
<tr>
<td>Primary Result</td>
<td>Positive</td>
<td>A mutation was found in a gene known to be associated with your medical concerns</td>
</tr>
<tr>
<td></td>
<td>Likely Positive</td>
<td>A mutation was found in a gene that most likely causes your medical concerns</td>
</tr>
<tr>
<td></td>
<td>Uncertain</td>
<td>A genetic change was found, but it is unclear if it is the cause of your medical concerns</td>
</tr>
<tr>
<td></td>
<td>Negative</td>
<td>The underlying cause of your medical concerns has not been found among genes associated with genetic conditions</td>
</tr>
<tr>
<td>Candidate Genetic Etiologies</td>
<td>Uncertain</td>
<td>A genetic change was found, but based on information we know today, it is unclear this is the cause of your medical concerns</td>
</tr>
<tr>
<td></td>
<td>Negative</td>
<td>The underlying genetic cause of your medical concerns has not been found</td>
</tr>
<tr>
<td>Secondary Findings Result</td>
<td>Positive</td>
<td>Mutation(s) was found in a gene that is not associated with your medical concerns</td>
</tr>
<tr>
<td></td>
<td>Negative</td>
<td>No mutation(s) was found among a list of genes that are not associated with your medical concerns</td>
</tr>
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</table>
Characterized Genetic Etiologies: **Positive**

- **Result**: Mutation(s) was found in a gene known to be associated with your medical concerns.

- **Medical Management**: Your doctor or genetic counselor will discuss if or how this result changes your or your family’s medical care.

- **Genetic Testing for Family Members**: Other family members may consider testing for the specific mutation(s).

- **Future Testing**: Usually not recommended/necessary.
Characterized Genetic Etiologies: ** Likely Positive

**Result**
A mutation was found in a gene that most likely causes your medical concerns

**Medical Management**
Your doctor or genetic counselor will discuss if or how this result changes your or your family’s medical care

**Genetic Testing for Family Members**
Other family members may consider testing for the specific mutation(s)

**Future Testing**
Usually not recommended/necessary
Characterized Genetic Etiologies: **Uncertain**

**Result**
A genetic change was found, but it is unclear if it is the cause of your medical concerns.

**Medical Management**
Your doctor or genetic counselor will discuss if or how this result changes your or your family’s medical care.

**Genetic Testing for Family Members**
In some cases, testing other family members for this specific gene change(s) may help to better understand your results.

**Future Testing**
Studying your exome data again in the future may be helpful.
Characterized Genetic Etiologies: **Negative**

**Result**
The underlying cause of your medical concerns has not been found in genes associated with genetic conditions.

**Medical Management**
Your doctor or genetic counselor will discuss if or how this result changes your or your family’s medical care.

**Genetic Testing for Family Members**
Testing other family members is not recommended at this time.

**Future Testing**
Studying your exome data again in the future may be helpful.
Candidate Genetic Etiologies: **Uncertain**

**Result**
A change(s) in a gene was found, but based on the information we know today, it is not clear if this is the cause of your medical concerns.

**Medical Management**
Your doctor or genetic counselor will discuss if or how this result changes your or your family’s medical care.

**Genetic Testing for Family Members**
In some cases, testing other family members for this specific gene change(s) may help to better understand your results.

**Future Testing**
Studying your exome data again in the future may be helpful.
**Candidate Genetic Etiologies: Negative**

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<tr>
<th>Result</th>
<th>No relevant mutation(s) was identified among genes that could cause a genetic condition. The underlying cause of your medical concerns has not been found.</th>
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<td>Medical Management</td>
<td>No relevant mutation(s) was identified among genes that could cause a genetic condition. The underlying cause of your medical concerns has not been found.</td>
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