MTHFR & HOMOCYSTEINE TEST

MTHFR is a gene that encodes for the methylenetetrahydrofolate reductase enzyme. This enzyme plays a critical role in vitamin-B12 and folate metabolism. Genetic variations in the MTHFR gene cause the enzyme to work less efficiently and can lead to elevated homocysteine, which is a potentially toxic amino acid. High levels of homocysteine are related to increased risk of developing cardiovascular disorders, psychiatric disorders, cancer, osteoporosis, and pregnancy complications. Castle performs MTHFR genotyping with OraCollect buccal swab samples. We also test for homocysteine levels in urine and blood samples.

Purpose
This test is especially relevant for people who have a personal or family history of premature cardiovascular disease or thrombosis. It may also be ordered when a close relative has MTHFR gene mutations. Prospective mothers with a history of miscarriages are also advised to take this test.

Methodology
MTHFR genotyping is performed using TaqMan fluorescence probe based assays in a realtime polymerase chain reaction (qPCR). The homocysteine test is performed using liquid chromatography-mass spectrometry (LC-MS).

Alleles tested
Two important MTHFR variants, rs1801133 (C677T) and rs1801131 (A1298C), are tested. The table below shows the percentage of MTHFR enzyme activity in different MTHFR gene variants.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>677CC</th>
<th>677CT</th>
<th>677TT</th>
</tr>
</thead>
<tbody>
<tr>
<td>1298AA</td>
<td>100%</td>
<td>67%</td>
<td>25%</td>
</tr>
<tr>
<td>1298AC</td>
<td>83%</td>
<td>47%</td>
<td>-</td>
</tr>
<tr>
<td>1298CC</td>
<td>61%</td>
<td>-</td>
<td>-</td>
</tr>
</tbody>
</table>

Specimen
OraCollect (OC-100) swab for MTHFR test.
Urine/Blood for homocysteine test.

Storage
For buccal swab and urine samples room temperature, out of intense light. Do not freeze or refrigerate specimens.
Blood samples should be stored in refrigerator (4ºC - 8ºC).

Instructions
For the buccal swab, please follow the instructions provided with the saliva collection kit.
For urine/blood collection, fasting for at least 12 hours is recommended.

Cause of Rejection
Insufficient/Contaminated sample