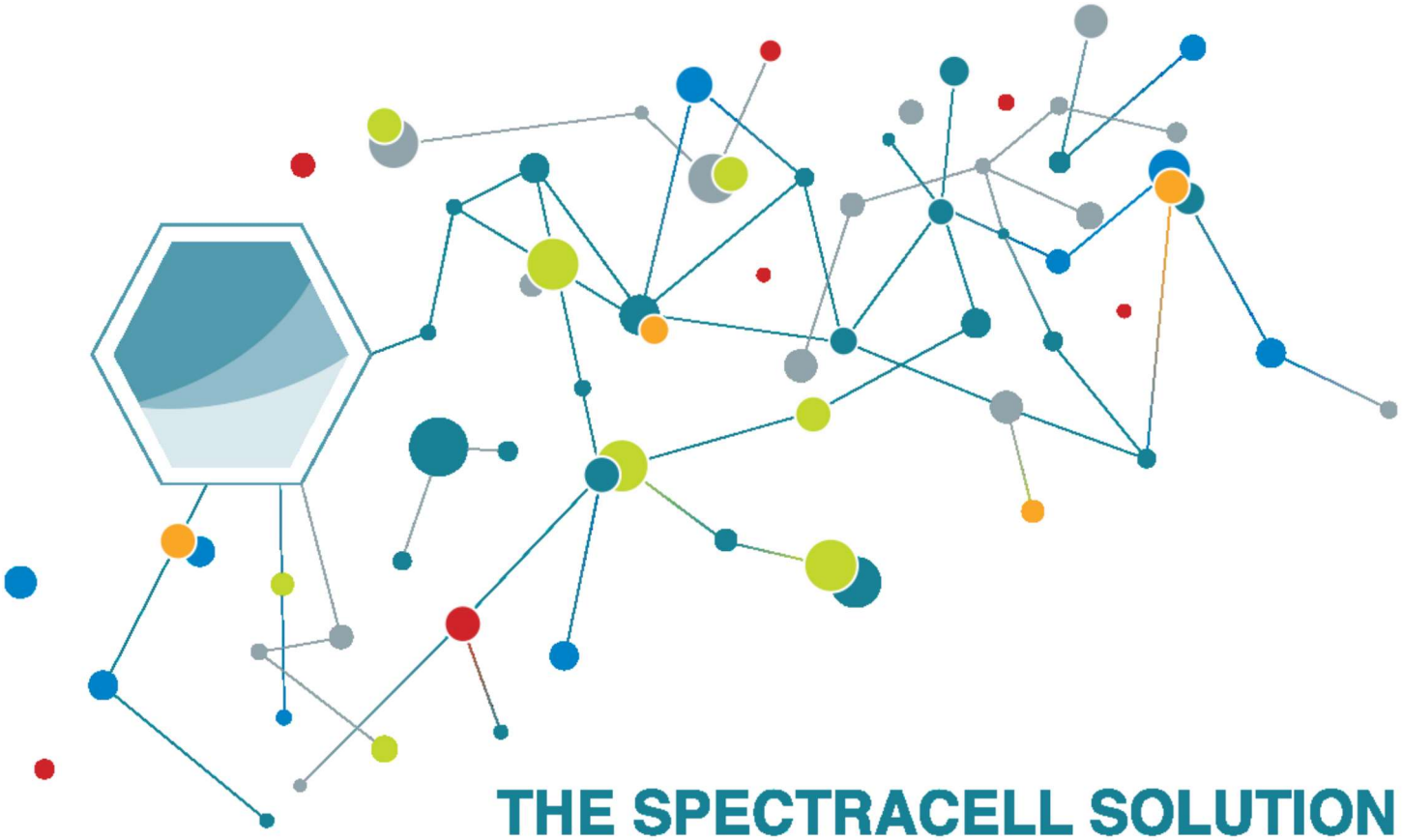

THE SPECTRACELL SOLUTION

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Patient: **Doe, Jon**

Accession ID: 0000000000

Provider: Sample Provider, MD

Order Status: Complete

MTHFR

10401 Town Park Drive, Houston, TX 77072
1(800) 227-5227 | support@spectracell.com | www.spectracell.com

SpectraCell Laboratories
Science + Health + Solutions

PATIENT

NAME	AGE
Doe, Jon	**25**
DOB	Gender
6/25/1993	**Male**
Patient ID	
00-000-00000	

SPECIMEN

ACCESSION ID	DATE COLLECTED
0000000000	**04/02/2019**
ORDER ID	DATE RECEIVED
0000-00000000000-000000	**04/03/2019**
	DATE REPORTED
	04/22/2019

PROVIDER

Account ID	CLIENT NAME
00000000	**Sample Provider, MD**

Address
123 S. Any Street
ANYWHERE, TX 77000

MTHFR

Tests	Results
C677T Mutation A1298C Mutation	Homozygous Negative

MTHFR Interpretation

This sample has two copies of the C677T mutation and is negative for the A1298C mutation.
* Is associated with decreased enzyme activity (approximately 30% of normal activity).
* Is associated with increased homocysteine levels.
* Is correlated with increased risk of cardiovascular disease or thrombosis.
* Is associated with potential methotrexate intolerance and patients may require dosage adjustments or discontinuation.

MTHFR Overview

MTHFR (methylenetetrahydrofolate reductase) is an enzyme involved in the metabolism of folate and homocysteine. It plays a role in maintaining cellular folate levels and is a cofactor needed to convert homocysteine (a potentially toxic amino acid) to methionine.

Certain common genetic point mutations have been characterized that reduce the function of the MTHFR enzyme. These are the C677T mutation (which is a change from cytosine to thymine at position 677 within the gene) and the A1298C mutation (which is a change from adenine to cytosine at position 1298 within the gene.) An MTHFR enzyme with reduced function can lead to elevated homocysteine levels, which is a known independent risk factor for development of cardiovascular disease and venous thrombosis. Reduced enzyme function can also affect folate status.

An additional area in which the function of MTHFR can have an effect is during methotrexate therapy. Methotrexate is a drug often used in treatment of certain cancers or autoimmune diseases. It is a structural analogue of folate and can interfere with folate metabolism. Defects in folate metabolism such as those potentially arising from mutations affecting MTHFR function can increase sensitivity to methotrexate and may lead to lower dosage requirements, increased side effects, or intolerance of the drug.

Testing Limitations

Only the C677T and A1298C mutations are analyzed in this assay. There may be other unknown non-genetic factors or genetic factors besides the tested mutations that can affect homocysteine levels, folate status, or drug sensitivities. Rare mutations in the primer binding sites used to detect the C677T and A1298C mutations may prevent detection. Specific dosing guidelines for methotrexate based on MTHFR genotype are not currently available.

MTHFR genotyping can provide useful information concerning risks of developing cardiovascular disease or thrombosis, or potential for increased sensitivity to methotrexate treatment. However, genotyping alone is not predictive of development of disease or complication and should not be used as the primary means of clinical diagnosis or treatment decision making. This information should be used by a physician in conjunction with additional clinical information to determine an appropriate treatment regimen.