

MTHFR Genetic test result	Expected outcome and possible further treatment
<p><b>C677T heterozygote (CT)</b> This means one copy (allele) of the MTHFR C677T variant gene has the normal C allele and the other copy is the variant T allele.</p>	<p>C677T heterozygotes have approximately 50% enzyme activity<sup>7</sup> however this does not usually cause any health concerns if dietary folate intake is adequate.</p>
<p><b>A1298C heterozygote (AC)</b> This means one copy (allele) of the MTHFR A1298C variant gene has the normal A allele and the other copy is the variant C allele.</p>	<p>A1298C heterozygotes have approximately 60% enzyme activity<sup>7</sup>. This has not been associated with any health concerns.</p>
<p><b>C677T/A1298C compound heterozygote (CT/AC)</b> This means one copy (allele) of the MTHFR C677T variant gene has the normal C allele and the other copy is the variant T allele and the MTHFR A1298C variant gene has the normal A allele and the other copy is the variant C allele.</p>	<p>MTHFR compound heterozygotes have approximately 36% enzyme activity<sup>7</sup>. This result does not usually cause any health concerns if dietary folate intake is adequate.</p>
<p><b>A1298C homozygote (CC)</b> This means both copies (alleles) of the MTHFR A1298C variant gene have the variant C allele.</p>	<p>A1298 homozygotes have approximately 50% enzyme activity<sup>7</sup>. This result does not usually cause any health concerns.</p>
<p><b>C677T homozygote (TT)</b> This means both copies (alleles) of the MTHFR C677T variant gene have the variant T allele.</p>	<p>C677T homozygotes have approximately 22% enzyme activity<sup>7</sup>. Some people with this result may develop a mild to moderate increased blood homocysteine level or hyperhomocysteinaemia, which is a risk factor for cardiovascular disease.</p> <p>The thrombophilic tendency is minimised by an adequacy of folate, riboflavin, B6 and B12. Some clinicians advocate that all pregnant women should ensure adequate intake of these vitamins<sup>8</sup>.</p> <p>Further testing may be recommended, such as B12, RBC folate and homocysteine.</p>