

#ret #incomplete

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## 1 | SNP. Project. Write-up.

Resources: [\[\[KBxSNPPCR\]\]](#) Instructions

### 1.0.1 | Part One

#### Outline

- basics
- function and regulation
- SNP effect

#### Writing!

The *COMT* gene, or catechol-O-methyltransferase, encodes the *COMT* enzyme which is responsible for breaking down neurotransmitters in the brain's prefrontal cortex. More specifically, it acts as a catalyst for the transfer of a methyl group from S-adenosylmethionine to dopamine, epinephrine, and norepinephrine. This process, called O-methylation, leads to the degradation of the aforementioned neurotransmitters. The *COMT* enzyme also effects the metabolism of exogenous substances, but that is irrelevant for the mutation at hand. The *COMT* gene itself is 27.22kb long and located on chromosome 22q11.2. It has ubiquitous expression in 27 tissues, including the placenta, the adrenal, and the lung. Val158Met, also known as rs4680, is a common missense mutation swapping a guanine for an adenine. It has the frequency  $G=0.510915$ , and thus,  $A=0.489085$ . Val158Met causes the *COMT* enzyme to be roughly 25% as effective compared to the wild type. Expression levels in mRNA, however, are not effected by Val158Met.