Why almost everybody has a MTHFR mutation.

A lot of guff is written about how mutations in the gene for the MTHFR enzyme (also called MTHFR). One would think that anybody with a MTHFR mutation must be lucky to be alive. But you almost certainly have at least one mutation in that gene.

The genetic analysis program Promethease lists 16 SNPs (Single Nucleotide Polymorphisms – the posh name for places that mutations can happen) for the MTHFR gene. Of those it lists the frequencies with which those mutations occur for 15. We can use that to calculate the probability of not having any of those mutations. I've used the probabilities for those with Northern and Western European Ancestry.

The probability of not having a mutation is location rs1801133 is 46.9% (or 0.469). The probability of not having a mutation at location rs4846049 is 38.5% (0.385) Of the 46.9% with no mutation at rs1801133, only 38.5% will also have no mutation at rs4846049. That means that 0.469 x 0.385 = 0.180 (18%) will not have a mutation at either location. Already we have 82% having a mutation at one of those two locations.

If we continue the sum for the other 13 locations we get.

0.469 x 0.385 x 0.619 x 0.487 x 0.460 x 0.496 x 0.690 x 0.903 x 0.690 x 0.761 x 0.905 x 0.892 x 0.885 x 1.00 x 0.434 = 0.00126 (0.126%)

That means that 99.874% of people do have at least one mutation on those 15 locations. That is just 8 people in a thousand who do not have a mutation.

How has the human race survived with all those people carrying such terrible mutations? Obviously those mutations are not so terrible. Indeed, most mutations do little or no harm. There is just one mutation that might possibly be harmful, according to the testing company 23andMe, and that is homozygous for the C677T mutation, carried by 9% of the population.