



# Personalized One Carbon panel

(extended Yasko methylation panel)



BiONuMeRi tools 2016/2017

Paziente

:

**Lilly**

**Mendel**

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rs id	Nome Gene	Rischio	G1	G2	Risultato	Breve descrizione
rs1801131	MTHFR A1298	G	G	T	(+/-)	Defect in folate metabolism, impacting BH4 levels
rs1801133	MTHFR C677T	A	G	G	(-/-)	Influence homocysteine levels and lowers folate levels. In homozygous mutation, severe psychomotor development and thrombotic issues
rs585800	BHMT 1	T	x	x	not tested	Convert homocysteine to methionine. Together BHMT4 play a role in the gut
rs567754	BHMT 2	T	C	T	(+/-)	Convert homocysteine to methionine. Together BHMT4 play a role in the gut
rs617219	BHMT 4	C	x	x	not tested	Convert homocysteine to methionine. Together BHMT4 play a role in the gut
rs651852	BHMT 8	T	T	T	(+/+)	Convert homocysteine to methionine, BHMT8 is related to the impact that psychological stress has on attention levels
rs234706	CBS C699T	A	G	G	(-/-)	Together A360A and N212N convert homocysteine into cystathionine, when mutated lowers homocysteine level, raise ammonia, increase taurine, increase toxic sulfur products and reduce folic acid effect
rs1801181	CBS A360A	A	A	G	(+/-)	Together C699T and N212N convert homocysteine into cystathionine, when mutated lowers homocysteine level, raise ammonia, increase taurine, increase toxic sulfur products and reduce folic acid effect
rs2298758	CBS N212N	G	x	x	not tested	Together A360A and C699T convert homocysteine into cystathionine, when mutated lowers homocysteine level, raise ammonia, increase taurine, increase toxic sulfur products and reduce folic acid effect
rs773115	SUOX S370S	C	x	x	not tested	Detoxify from sulfites

rs6323	MAO A	T	T	T	(+/+)	Degrades serotonin, melatonin, epinephrine and norepinephrine. It can lower the tryptophan levels. Together a COMT V158M mutation can give mood swings. Neutralizes tyramine (+/+ reduced activity -/- increased activity)
rs1799836	MAO B	T	T	T	(+/+)	Degrades dopamine. It contributes to Parkinson's disease
rs4680	COMT V158M	A	A	G	(+/-)	Degrades dopamine. With A allele you have anxiety so high dopamine, low pain threshold, Stress vulnerability, greater cognitive performance. With G allele you are wrestler then you have low dopamine, high pain threshold, resistance to stress and lower cognitive performance. (+/+) Poorly tolerate to methyl donors.
rs4633	COMT H62H	T	C	T	(+/-)	Degrades dopamine. Susceptibility to schizophrenia
rs769224	COMT P199P	A	G	G	(-/-)	Degrades dopamine.
rs1805087	MTR A2756G	G	A	A	(-/-)	Work with MTRR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.
rs1801394	MTRR A66G	G	A	G	(+/-)	Work with MTR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.
rs10380	MTRR H595Y	T	x	x	not tested	Work with MTR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.
rs162036	MTRR K350A	G	A	A	(-/-)	Work with MTR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.
rs2287780	MTRR R415T	T	x	x	not tested	Work with MTR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.

rs2303080	MTRR S257T	T	x	x	not tested	Work with MTR to regenerate and use the B12 helping to convert homocysteine to methionine. The mutation can lower B12 levels. Converts MTHF in THF adding the methyl group to B12.
rs1802059	MTRR 11	A	G	G	(-/-)	(see MTR/MTRR) Generally people with MTRR11 mutated are good heavy metal excretor
rs819147	AHCY 1	C	C	T	(+/-)	The AHCY mutations limit the activity of homocysteine conversion to methionine. They can mitigate the effects of CBS mutations.
rs819134	AHCY 2	G	x	x	not tested	The AHCY mutations limit the activity of homocysteine conversion to methionine. They can mitigate the effects of CBS mutations.
rs819171	AHCY 19	C	C	T	(+/-)	The AHCY mutations limit the activity of homocysteine conversion to methionine. They can mitigate the effects of CBS mutations.
rs1979277	SHMT C1420T	A	x	x	not tested	It helps to convert folic acid into 5MTHF. It transfers a methyl group from serine 5,10-methylene THF and glycine
rs1799983	NOS3 D298E	T	x	x	not tested	Detoxify from ammonia
rs4880	SOD2 A16V	G	A	G	(+/-)	mitochondrial SOD (WARNING very difficult to identify, refer to <a href="http://www.snpedia.com/index.php/Rs4880">http://www.snpedia.com/index.php/Rs4880</a> for alzheimer G risk allele)
rs1799895	SOD3 C760G	G	C	C	(-/-)	extracellular SOD
rs1695	GSTP1 I105V	G	G	G	(+/+)	Enzyme that promotes detoxification (Glutathione), a mutation causes a loss of function of the enzyme and less detoxification
rs1138272	GSTP1 A114V	T	C	T	(+/-)	see GSTP1 I105V
rs366631	GSTM1 -	A	A	A	(+/+)	(+/+) Double deletion results in a loss of functionality of the enzyme and less detoxification
rs731236	VDR Taq	A	A	G	(+/-)	Affects dopamine levels
rs10735810	VDR Fok	A	x	x	not tested	Affects blood sugar control and pancreatic function (possible adoption of a low-carbohydrate diet)

rs1544410	VDR Bsm	T	C	T	(+/-)	Affects dopamine levels, related to bone density. When mutated low vitamin D3 and D2 values.
rs2060793	CYP2R1 -	A	G	G	(-/-)	Activate D vitamin
rs12272004	APOA5 -	A	C	C	(-/-)	When mutated low values of Tocopherol (Vitamin E)
rs602662	FUT2 -	G	A	G	(+/-)	When mutated low vitamin B12 values.
rs4654748	NBPF3 -	C	T	T	(-/-)	When mutated low vitamin B6 values in the blood.
rs10483639	GCH1 -	C	G	G	(-/-)	Together with two other GCH1, when mutated low values of tetrabiopterine (BH4)
rs3783641	GCH1 -	A	T	T	(-/-)	Together with two other GCH1, when mutated low values of tetrabiopterine (BH4)
rs8007267	GCH1 -	T	C	C	(-/-)	Together with two other GCH1, when mutated low values of tetrabiopterine (BH4)
rs104894460	PNP -	T	C	C	(-/-)	When mutated, purine deficiency. T-cell immune deficiency
rs3741049	ACAT -	A	A	G	(+/-)	It contributes to the cholesterol synthesis and lipid balance of the cell membrane. Mediates the accumulation of oxalates. It contributes to energy production.
rs5963409	OTC -	A	A	A	(+/+)	degrades ammonia into urea, may cause high blood ammonia, Increase Alzheimer's Risk.
rs3749034	GAD1 -	G	G	G	(+/+)	Influences the GABA production, increases the risk of schizophrenia, (+/+) gaba significantly decreased, with comt v158m (+/-) (-/-) reduced cortical thickness
rs3828275	GAD1 -	T	C	T	(+/-)	(+/+) associated with post-traumatic seizures and high glutamate levels, (+/-) average glutamate levels
rs1978340	GAD1 -	G	A	G	(+/-)	Affects GABA levels. Complex attention! (+/-) Low levels, (+ / +) average levels, (- / -) high GABA levels
rs3791878	GAD1 -	G	G	T	(+/-)	Affects GABA levels. (+/+) high Glutamate, (+/-) intermediate levels
rs72554331	OTC -	A	G	G	(-/-)	degrades ammonia into urea, if mutated high ammonia levels

rs67960011	OTC -	G	C	C	(-/-)	degrades ammonia into urea, if mutated high ammonia levels
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NB: Il presente pannello e' estratto elaborando il file di esoma 23andme.com fornito dall'utente, pertanto non si certifica la veridicit  dei dati pervenuti. Il risultato del pannello non costituisce in alcun modo consiglio medico. Ogni commento di ogni singolo snp non costituisce assoluta certezza ma deve essere vagliato da un medico e verificato da indagini mirate o da prove cliniche. In questo pannello non viene presa in considerazione l'espressione genetica (epigenetica) ed i vari snp possono interferire tra loro enfatizzando o annullando determinati errori metabolici. Per maggiori dettagli e la bibliografia consultare la pagina <http://www.bionumeri.org/esami/onecarbon.html>

