NutraHacker

Imputed Lactation Report for Customer: 55ffb33f-2a2f-45d3-88b1-a0d59bf766b6

Disclaimer: This report is offered by Nutrahacker, LLC. It is not intended to diagnose, treat, or prevent any disease. It is for informative and educational purposes only. Please follow the advice of your healthcare provider.

This is a genetic report that identifies single nucleotide polymorphisms (SNP) that have an influence on lactation or the breastfed infant. These genes relate to the mother and do not analyze genetics of the child.

SNPs starred with an asterisk (*) have been calculated using impute software.

Imputed alleles are phased, meaning it can be discerned which gene copy caries a particular allele. Phasing separates maternally and paternally inherited copies of alleles so that haplotypes can be determined.

Conventionally, major/minor allele is selected as the predominant allele versus less common allele in a particular population, and this can vary depending on the population assessed.

In this report, the majority of "expected" alleles come from the major allele and variant is minor allele, however in certain circumstances it was found that the minor allele provides a benefit and is selected as expected or "beneficial" allele. We refer to these as "expected" and "variant" allele.

This report is provided for informational purposes, speak with your doctor if you would like further understanding or guidance. This report should not be used to make any changes to diet or breastfeeding without consulting under the care of a licensed physician or nutritionist.

Helpful excerpts from various publications:

Secretor: The activity of alpha-[1,2]-fucosyltransferase, encoded by the FUT2 gene, determines "secretor" versus "nonsecretor" status and HBGA expression. Secretors can express 2-fucosylated oligosaccharides in breast milk and H-type 1 antigens on mucosal surfaces. Homozygotes for inactivating alleles in FUT2 are characterized as nonsecretors and lack this ability but seem to be protected against symptomatic RV infections.

Maternal Secretor Status Affects Oral Rotavirus Vaccine Response in Breastfed Infants in Bangladesh - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8561252/

Fucosylated oligosaccharides are decoy receptors for many pathogenic bacteria, including members of enterobacteria; hence, they have a potential to reduce their adhesion to the gut, thus protecting the infant. In addition, fucosylated HMOs bind enterotoxin produced by E. coli.

Fucosylated oligosaccharides in mother's milk alleviate the effects of caesarean birth on infant gut microbiota - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6137148/

What are the benefits of human milk oligosaccharides?

Several studies have reported the health benefits of HMOs, which include modulation of the intestinal microbiota, anti-adhesive effect against pathogens, modulation of the intestinal epithelial cell response, and development of the immune system.

The Role of Two Human Milk Oligosaccharides, 2'-Fucosyllactose and Lacto-N-Neotetraose, in Infant Nutrition - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6629589/

The results indicate that maternal secretor status may be particularly influential in infants with compromised microbiota development, and that these infants could benefit from corrective supplementation.

The combination of caesarean birth and lack of milk 2'FL appear to profoundly alter the infant's microbiota. The FUT3 enzyme determines the Lewis type (Le), resulting in strong reduction of alpha-1,3 and absence of alpha-1,4 fucosylated HMOs in milk. Lewis and Secretor type thus allow grouping of human milk in four main milk groups, also sometimes named 'lactotypes'.

Biology of human milk oligosaccharides: From basic science to clinical evidence - https://onlinelibrary.wiley.com/doi/full/10.1111/jhn.12990

The importance of functional Se and Le genes in infant development is an area of active research. One study showed that maternal secretor status appeared to be important for preventing diarrhea, as although the gut microbiota measured through 16S rRNA sequencing did not differ between infants of Se+ and Se- mothers, the prevalence of diarrhea was higher among infants of Se- mothers (18). Moreover, when these infants were provided iron supplements, infants of Se- mothers were more likely to experience a decrease in the abundance of Bifidobacterium and an increase in pathogens compared to infants of Se+ mothers (18). However, supplementation with galactooligosaccharides appeared to ameliorate the impact of iron supplementation (18).

In terms of the oligosaccharides and their metabolites, 3'galactosyllactose, 3'SL, fucose, and LNnT were between 2- and 10-fold lower in milk samples from Se-Le- and Se-Le+ compared to Se+ mothers. Galactose was 6 and 1 times higher in milk samples from Se-Le- and Se-Le+ mothers, respectively, compared to samples from Se+ mothers. For metabolites associated with energy metabolism, samples from Se-Le- milk were approximately 4 times higher in creatine phosphate, 12 times higher in creatine, 4 times higher in creatinine, 5 times higher in citrate, 6 times higher in pyruvate, and 10 times higher in succinate compared to Se+ milk, while these metabolites were similar in concentration between milk from Se-Le+ and Se+ mothers.

The Milk Metabolome of Non-secretor and Lewis Negative Mothers - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7901958/

Helpful reading:

Genetic and Physiological Factors Affecting Human Milk Production and Composition - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7284811/
Review of Infant Feeding: Key Features of Breast Milk and Infant Formula - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4882692/
Human milk oligosaccharides: Every baby needs a sugar mama - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3406618/
The Milk Metabolome of Non-secretor and Lewis Negative Mothers - https://www.frontiersin.org/articles/10.3389/fnut.2020.576966/full
Human Milk Oligosaccharides and Lewis Blood Group: Individual High-Throughput Sample Profiling to Enhance Conclusions From Functional Studies - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3649481/
The Impact of Dietary Fucosylated Oligosaccharides and Glycoproteins of Human Milk on Infant Well-Being - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7230487/
Influence of Gestational Age, Secretor, and Lewis Blood Group Status on the Oligosaccharide Content of Human Milk - https://pubmed.ncbi.nlm.nih.gov/27602704/
Innate protection conferred by fucosylated oligosaccharides of human milk against diarrhea in breastfed infants - https://academic.oup.com/glycob/article/14/3/253/642175
The Impact of Dietary Fucosylated Oligosaccharides and Glycoproteins of Human Milk on Infant Well-Being - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7230487/

Total number of expected homozygous mutations: 30
Total number of variant homozygous mutations: 8
Total number of heterozygous mutations: 11
Total number of SNPs imputed: 40

Gender of customer: Male

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs602662*		G258S	G	A	GG: 2/2
missense variant G739A (rs602662) and were designated as nonsecretors					
https://pubmed.ncbi.nlm.nih.gov/31763671/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs199854112	ABCG2	T421A		A	N/A
Mothers harboring the c.421C > A polymorphism in ABCG2, secreted threefold more nifedipine to human milk					
https://pubmed.ncbi.nlm.nih.gov/30659932/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3733890*	BHMT	Arg239Gln	G	A	GG: 2/2
When evaluated independently, the BHMT variant allele was associated with non-significantly decreased turnover of choline to betaine, and increased turnover of choline to CDP-PC as well as a (non-significantly, p = 0.07) lower betaine-d9/PC-d9 enrichment ratio. Together, these results indicate that the variant favors the use of dietary choline for CDP-PC synthesis at the expense of betaine synthesis.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs12676*	CHDH		CC	A	CC: 2/2
altered the use of choline as a methyl donor, altered the distribution of dietary choline between the CDP-choline and phosphatidylethanolamine N-methyltransferase (PEMT) denovo pathway.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs9001*	CHDH		TT	G	TT: 2/2
The rs9001 variant may relatively favor CDP-PC synthesis over PEMT-PC synthesis. This relatively greater use of choline for CDP-PC synthesis as compared to PEMT synthesis among CHDH rs9001 variants may conserve choline stores.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs10791957	CHKA		CC	A	CA: 1/2
altered the use of choline as a methyl donor, altered the distribution of dietary choline between the CDP-choline and phosphatidylethanolamine N-methyltransferase (PEMT) denovo pathway.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3798719*	ELOVL2		T	C	CT: 1/2
Regarding elongases rs953413-A and rs3798719-T in ELOVL2 gene were associated with higher EPA levels.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs953413*	ELOVL2		A	G	GA: 1/2
Regarding elongases rs953413-A and rs3798719-T in ELOVL2 gene were associated with higher EPA levels.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs12207094*	ELOVL5		T	A	AA: 2/2
Regarding the ELOVL5 gene, we found that children of mothers carrying the rs17544159-C allele (associated with higher EPA/AA ratio in colostrums) or carrying the rs12207094-T allele (associated with both higher EPA/AA and DHA/AA ratios in colostrum) had higher cognition scores related to children of mothers homozygote for the major allele.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs17544159*	ELOVL5		C	A	AA: 2/2
Regarding the ELOVL5 gene, we found that children of mothers carrying the rs17544159-C allele (associated with higher EPA/AA ratio in colostrums) or carrying the rs12207094-T allele (associated with both higher EPA/AA and DHA/AA ratios in colostrum) had higher cognition scores related to children of mothers homozygote for the major allele.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2397142*	ELOVL5		C	T	GC: 1/2
Not being breastfed resulted in a disadvantage in cognition (5 to 8 points) among children CC homozygote for rs23971 42 (low ELOVL5 activity), but not among those carrying the G allele. In ELOVL5, the rs2397142-C allele was nominally associated with lower capacity to metabolize EPA to DPA. Among children CC homozygotes for rs2397142 (low ELOVL5 index), exposure to high levels of EPA, and high EPA/AA and DHA/AA ratios were associated with a 6.7-, 8.6- and 10.1-points advantage in scores, respectively, relative to children exposed to low levels					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174464*	FADS1		G	A	GG: 2/2
Related to lower levels of AA (Table S2). The rs174602-G, and 174464-T in the FADS cluster were associated with lower levels of DHA.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174468*	FADS1		A	G	AG: 1/2
Not being breastfed conferred an 8- to 9-point disadvantage in cognition among children GG homozygote for rs174468 (low FADS1 activity) but not among those with the A allele.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174553*	FADS1		A	G	AA: 2/2
The minor allele homozygotes of rs174553 (GG), rs99780 (TT), and rs174583 (TT) were associated with significantly lower 14:0, arachidonic (ARA, 20:4) and eicosapentanoic acid (EPA, 20:5), but higher 20:2 (omega 6) fatty acid in human milk [66]. Mothers carrying the minor homozygous allele G/G of rs174575, showed lower ARA, EPA, and docosahexanoic acids (DHA, 22:6 (omega 3)) and 22:5 (omega 3) levels in human milk [66]. Mothers carrying FADS1 rs174561, FADS2 rs174575, and intergenic rs3834458 minor alleles were found to have lower proportions of DHA in human milk. Mothers carrying genetic variants associated with lower FADS1 activity (regulating AA and EPA synthesis), higher FADS2 activity (regulating DHA synthesis), and with higher EPA/AA and DHA/AA ratios in colostrum showed a significant advantage in cognition at 14 months.					
https://academic.oup.com/jn/article/138/11/2222/4670097?login=false					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174556*	FADS1		C	T	CC: 2/2
Together with a recently published study that found that mothers carrying the minor allele of FADS1 rs174556 had lower proportions of ARA [108]					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7284811/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174561*	FADS1		T	C	TT: 2/2
DHA proportions in milk increased only with fish and fish-oil intake in the major-allele carriers.					
https://pubmed.ncbi.nlm.nih.gov/20335541/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs99780*	FADS1		C	T	CC: 2/2
The minor allele homozygotes of rs174553 (GG), rs99780 (TT), and rs174583 (TT) were associated with significantly lower 14:0, arachidonic (ARA, 20:4) and eicosapentanoic acid (EPA, 20:5), but higher 20:2 (omega 6) fatty acid in human milk [66]. Mothers carrying the minor homozygous allele G/G of rs174575, showed lower ARA, EPA, and docosahexanoic acids (DHA, 22:6 (omega 3)) and 22:5 (omega 3) levels in human milk [66]. Mothers carrying FADS1 rs174561, FADS2 rs174575, and intergenic rs3834458 minor alleles were found to have lower proportions of DHA in human milk.					
https://academic.oup.com/jn/article/138/11/2222/4670097?login=false					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1535*	FADS2		A	G	AA: 2/2
GG homozygotes of rs1535 are disadvantageous for tissue DHA [15,17,24], meaning the G allele (major allele in this study, but minor in others) tended to decrease milk DHA proportions. Homozygous carriers of the minor allele of rs1535 had a DHA increase of 1.8 FA% (P = 0.001) relative to those with the wild-type allele, whereas minor allele carriers of rs174448 and rs174575 had a decrease of 1.1 FA% (P = 0.005) and 2.0 FA% (P = 0.001), respectively. Each 10-g increment in fish intake was associated with an increased DHA status of 0.3 FA%. At 3 y, fish intake was the only significant determinant of DHA status (0.2 FA%/10 g).					
https://pubmed.ncbi.nlm.nih.gov/32093185/					
https://pubmed.ncbi.nlm.nih.gov/23636240/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174448*	FADS2		A	G	AA: 2/2
Based on multiple linear regression of data from 164 mothers that completed this study, there was 0.28% (FA%) reduction in milk DHA in high versus low genetic risk (stratified by whether minor allele numbers were greater than or equal to 3 in rs1535 and rs174448) and 0.45% reduction in low versus high intake (stratified by whether DHA intake reached 200 mg/d).					
https://pubmed.ncbi.nlm.nih.gov/32093185/					
https://pubmed.ncbi.nlm.nih.gov/23636240/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174575*	FADS2		C	G	CC: 2/2
DHA proportions in milk increased only with fish and fish-oil intake in the major-allele carriers.					
https://pubmed.ncbi.nlm.nih.gov/20335541/					
https://www.pnas.org/doi/full/10.1073/pnas.0704292104					
https://academic.oup.com/ajcn/article/97/6/1403/4576927					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174583*	FADS2		C	T	CC: 2/2
The minor allele homozygotes of rs174553 (GG), rs99780 (TT), and rs174583 (TT) were associated with significantly lower 14:0, arachidonic (ARA, 20:4) and eicosapentanoic acid (EPA, 20:5), but higher 20:2 (omega 6) fatty acid in human milk [66]. Mothers carrying the minor homozygous allele G/G of rs174575, showed lower ARA, EPA, and docosahexanoic acids (DHA, 22:6 (omega 3)) and 22:5 (omega 3) levels in human milk [66]. Mothers carrying FADS1 rs174561, FADS2 rs174575, and intergenic rs3834458 minor alleles were found to have lower proportions of DHA in human milk.					
https://academic.oup.com/jn/article/138/11/2222/4670097?login=false					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174602*	FADS2		T	C	TT: 2/2
Moreover, the rs174602-G allele was associated with lower FADS2 index (capacity to synthesize DHA from EPA).					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs174627*	FADS2		T	G	GG: 2/2
Children of mothers carrying at least one copy of the minor allele (T) for this variant had higher cognition scores compared to children of mothers homozygotes for the major allele					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044172/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1000778	FADS3		G	A	GG: 2/2
Pairwise comparison showed that individuals major homozygous for the SNP rs1000778 in the FADS3 gene had lower concentrations of ALA and linoleic acid (LA) in their breast milk.					
https://www.sciencedirect.com/science/article/abs/pii/S095232781530034X					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2266782	FMO3	Glu158Lys	G	A	GA: 1/2
Converts TMA to TMAO, increased trimethylaminuria					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs9939609	FTO		T	A	TA: 1/2
Rs9939609 AA individuals were more susceptible than AT and TT individuals to both short and long breastfeeding durations, which is consistent with the differential susceptibility hypothesis.					
https://pubmed.ncbi.nlm.nih.gov/35127123/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1047781*	FUT2	A385T	A	T	AA: 2/2
Missense mutation, non-secretor					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6306508/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs200157007*	FUT2		C	T	CC: 2/2
One novel missense FUT2 SNP, rs200157007-TT and the earlier established rs601338-AA SNP were shown to be causing non-secretor status, with these SNPs being associated with symptomatic but not asymptomatic ETEC infection.					
https://www.nature.com/articles/s41598-017-10854-5					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs492602*	FUT2		G	A	AA: 2/2
Women homozygous for the rs492602[G] allele had higher B(12) levels. This allele is in strong linkage disequilibrium with the FUT2 nonsecretor variant encoding W143X, suggesting a plausible mechanism for altered B(12) absorption and plasma levels.					
https://pubmed.ncbi.nlm.nih.gov/18776911/					
https://pubmed.ncbi.nlm.nih.gov/23402911/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs601338*	FUT2	W143X	G	A	GG: 2/2
Individuals with a secretor phenotype were either homozygous (GG) or heterozygous (GA) at the position. Nonsense mutation W143X that introduces a premature stop codon in the FUT2 gene (rs601338) abolished the ability to synthesize alpha (1-2)-fucosylated HMOs (non-secretor status). Non-secretors where found to express less HMOs compared to mothers with secretor status (active FUT2) [67,68,69,70]. In addition, maternal secretor status was shown to be associated with the human milk microbiota composition [71]. Infants fed by non-secretor mothers, were delayed in the establishment of their gut microbiota, specifically bifidobacterial-laden (G- secretor, A- non-secretor). FUT2 SNP rs601338 was found to predominantly define the Secretor status Se-: 11.8% and it was highly correlated with 2'-fucosyllactose (2'FL, $p < 0.001$) and lacto-N-fucosylpentaose-I (LNFP-I, $p < 0.001$). Individuals with the wild type G/G genotype have high concentrations of 2'FL in their milk. All cases with vitamin B12 deficit carried the FUT2 rs601338 secretor variant.					
https://pubmed.ncbi.nlm.nih.gov/33195368/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs28362459*	FUT3	T59G	A		AA: 2/2
Milk samples derived from mothers with inactive FUT3 due to genetic variations are referred to as Lewis negative, as opposed to Lewis positive when FUT3 is active					
https://pubmed.ncbi.nlm.nih.gov/33195368/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3745635*	FUT3	G508A	C	T	CC: 2/2
Milk samples derived from mothers with inactive FUT3 due to genetic variations are referred to as Lewis negative, as opposed to Lewis positive when FUT3 is active					
https://pubmed.ncbi.nlm.nih.gov/33195368/					
https://onlinelibrary.wiley.com/doi/abs/10.1111/vox.13251					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3894326*	FUT3	I356K	A		AA: 2/2
Milk samples derived from mothers with inactive FUT3 due to genetic variations are referred to as Lewis negative, as opposed to Lewis positive when FUT3 is active					
https://pubmed.ncbi.nlm.nih.gov/33195368/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs778986	FUT3	C314T	G	A	GG: 2/2
Milk samples derived from mothers with inactive FUT3 due to genetic variations are referred to as Lewis negative, as opposed to Lewis positive when FUT3 is active					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7658960/					
https://www.sciencedirect.com/?ref=pdf_download&fr=RR-11&rr=71ef7bd8fbe3356b					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs812936	FUT3	T202C	A	G	AA: 2/2
Milk samples derived from mothers with inactive FUT3 due to genetic variations are referred to as Lewis negative, as opposed to Lewis positive when FUT3 is active					
https://pubmed.ncbi.nlm.nih.gov/33195368/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3834458	intergenic		C	CT	N/A
This meta-analysis indicates that minor allele of rs3834458 (insertion) in FADS2 may result in lower activity of delta-6 desaturase leading to higher ALA and lower EPA, DPA and DHA in blood. . For minor allele homozygotes, higher DHA intake compensated for lower DHA in women's plasma phospholipids but not in their milk.					
https://pubmed.ncbi.nlm.nih.gov/32093185/					
https://pubmed.ncbi.nlm.nih.gov/20335541/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2271714*	MFGE8		G	A	GG: 2/2
Maternal genotype for the rs2271714 variant within milk fat globule EGF and factor V/VIII domain containing gene (MFGE8) was associated with PIMS status (p = 0.009, adjusted p = 0.09, likelihood ratio = 9.33) and duration of exclusive breastfeeding (p = 0.009, adjusted p = 0.09, chi-square = 9.39).					
https://pubmed.ncbi.nlm.nih.gov/34939829/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1076991*	MTHFD1		C	T	TT: 2/2
rs1076991, rs2983733, rs2987981, rs8003379, and rs17824591 SNPs in the methylene tetrahydrofolate dehydrogenase 1 (MTHFD1) gene were found to be associated with very high human milk choline concentrations in three subjects					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2904035/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs17824591*	MTHFD1		G	A	GG: 2/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2236225	MTHFD1	Arg653Gln	G	A	AA: 2/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2983733*	MTHFD1		A	G	GG: 2/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2904035/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs2987981*	MTHFD1	C421A	G	T	AA: 0/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs8003379*	MTHFD1		A	C	AA: 2/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1801133*	MTHFR	C677T	G	A	GG: 2/2
The MTHFR 677C > T SNP was associated with higher levels of human milk unmetabolized folic acid (UMFA)					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6669053/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1805087*	MTR	Asp919Gly	A	G	AA: 2/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1801394	MTRR	Ile22Met	A	G	GA: 1/2
Overall, our findings indicate that loss-of-function variants in folate-metabolizing enzymes strain cellular PC production, possibly via impaired folate-dependent phosphatidylethanolamine-N-methyltransferase (PEMT)-PC synthesis, and suggest that women with these risk genotypes may benefit from choline intakes exceeding current recommendations					
https://pubmed.ncbi.nlm.nih.gov/27342765/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs53576	OXTR		G	A	AG: 1/2
Women with the minor allele OXTR rs53576 reported 8.18-fold higher breast and nipple pain severity over time					
https://pubmed.ncbi.nlm.nih.gov/33303340/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs4646343*	PEMT		T	G	GG: 2/2
Previous work has found a 92% overlap of the intronic PEMT rs4646343 SNP with the functional rs12325817 SNP, which is located in the promoter region of the PEMT gene, near the estrogen response element, and impedes its estrogen-mediated up-regulation [33]. This impairment leads to an increased susceptibility to organ dysfunction in variant individuals [10]. Although we did not observe direct indications of decreased PEMT activity, the observed decreased PEMT-PC/CDP-PC in rs4646343 variant individuals is consistent with decreased PEMT activity and an impaired estrogen response among variant individuals. Decreased PEMT-PC/CDP-PC was also observed in PEMT rs7946 variant lactating women.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs7946	PEMT	Val175met	C	T	TC: 1/2
Overall, these data suggest a relatively decreased contribution of PEMT-PC relative to CDP-PC in PC pools with both PEMT rs4646343 and rs7946 variants. Impaired PEMT activity may compromise PC-DHA supply to extra-hepatic tissue including vital reproductive organs during pregnancy and lactation. The PEMT rs7946 variant encodes a valine to methionine substitution at amino acid 175, which results in decreased enzymatic activity in vitro and may increase susceptibility to non-alcoholic fatty liver disease (NAFLD)					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1355400106	SLC30A2	S296L	G	A	N/A
Associated with transient neonatal zinc deficiency. We identified two novel missense mutations in the SLC30A2/ZnT2 gene in a Japanese mother with low milk zinc concentrations (>90% reduction) whose infant developed severe zinc deficiency; a T to C transition (c.454T>C) at exon 4, which substitutes a tryptophan residue with an arginine residue (W152R), and a C to T transition (c.887C>T) at exon 7, which substitutes a serine residue with a leucine residue (S296L).					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1477109267	SLC30A2	T312M	G	A	N/A
Associated with transient neonatal zinc deficiency.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4919441/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs1485358639	SLC30A2	663delC	C		N/A
Associated with transient neonatal zinc deficiency. We present a 4-month-old girl with TNZD due to a new autosomal dominant mutation (663delC) in the maternal SLC30A2 gene not previously described in the literature.					
https://pubmed.ncbi.nlm.nih.gov/24456035/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs151175941	SLC30A2	G299W	C	A	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs185398527	SLC30A2	G87R	C	T	N/A
Associated with transient neonatal zinc deficiency. Heterozygous mutations of human ZnT2 (hZnT2), including H54R and G87R, in mothers result in low (>75% reduction) secretion of zinc into the breast milk, and infants fed on the milk develop transient neonatal zinc deficiency. Dominant negative mutation, low zinc breast milk.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3436150/					
https://pubmed.ncbi.nlm.nih.gov/23741301/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs201084300	SLC30A2	G233D	C	T	N/A
Associated with transient neonatal zinc deficiency. Low zinc breast milk.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs35235055*	SLC30A2	L23P	A	G	AA: 2/2
Associated with transient neonatal zinc deficiency. Two reported SNPs resulting in L23P and R340C substitutions in hZnT2 may compromise mammary cell functions such as zinc secretion into the milk by changing the subcellular localization of hZnT2, as suggested in the transfection studies.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs377192955	SLC30A2	E355Q	G	A	N/A
Associated with transient neonatal zinc deficiency.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4919441/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs587776926	SLC30A2	H54R	T	C	N/A
Associated with transient neonatal zinc deficiency. Heterozygous mutations of human ZnT2 (hZnT2), including H54R and G87R, in mothers result in low (>75% reduction) secretion of zinc into the breast milk, and infants fed on the milk develop transient neonatal zinc deficiency.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3669329/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs746722376	SLC30A2	E279K	C	T	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs761084393	SLC30A2	V300L	C	A	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs765967769	SLC30A2	P245R	G	C	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs767112739	SLC30A2	G233R	C	G	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs769222243	SLC30A2	H106Y	G	A	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs772899336	SLC30A2	R165W	G	A	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs774477332	SLC30A2	N214K	G	T	N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs779896941	SLC30A2	G175W	C		N/A
Associated with transient neonatal zinc deficiency.					
https://onlinelibrary.wiley.com/doi/full/10.1111/jcmm.13982					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs3199966*	SLC4A1	Ser644Ala	T	G	TT: 2/2
Specifically, for both SNPs, variant (but not non-variant) individuals exhibited greater turnover of betaine ? methionine in the higher choline intake group as compared to the lower, suggesting that additional choline was used for methyl donation.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs7873937*	SLC4A1		G	C	CG: 1/2
altered the use of choline as a methyl donor, altered the distribution of dietary choline between the CDP-choline and phosphatidylethanolamine N-methyltransferase (PEMT) denovo pathway.					
https://pubmed.ncbi.nlm.nih.gov/28134761/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909174	SLC5A5	T354P	A	C	N/A
Mother carrying homozygous T354P mutation in the NIS transporter was reported to produce iodine-deficient milk., Mother supplementation with 50 mg potassium iodide tablet daily starting on the fifth day postpartum to increase iodine concentration in human milk.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7284811/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909175	SLCA5	C272X	C	A	N/A
This nonsense mutation produces a truncated NIS with undetectable I- transport activity when expressed into COS-7 cells. While the homozygous mutant NIS-272X causes congenital hypothyroidism, expression of one normal allele in the heterozygote (C272X) is sufficient to maintain active thyroidal I- uptake and function.					
https://pubmed.ncbi.nlm.nih.gov/9388506/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909176	SLCA5	Q267E	C	G	N/A
The Q267E mutation in the sodium/iodide symporter (NIS) causes congenital iodide transport defect (ITD) by decreasing the NIS turnover number.					
https://pubmed.ncbi.nlm.nih.gov/14734652/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909177	SLCA5	Y531X	C	T	N/A
Producing a stop codon as well as a downstream cryptic 3' splice acceptor site in exon 13, resulting in a 67 nucleotide deletion, frameshift, and premature stop (p.Tyr531X (Y531X)).					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC508654/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909178	SLCA5	G93R	G	C	N/A
Congenital I(-) transport defect-causing NIS mutant G93R					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5739519/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909179	SLCA5	G543E	G	A	N/A
Molecular analysis of a congenital iodide transport defect: G543E impairs maturation and trafficking of the Na+/I- symporter. Here, we show that in contrast to these mutants, G543E NIS matures only partially and is retained intracellularly; thus, it is not targeted properly to the cell surface, apparently because of faulty folding.					
https://pubmed.ncbi.nlm.nih.gov/15976004/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs121909180	SLCA5	G395R	G		N/A
Thyroid dyshormonogenesis I (also known as iodide transport defect). Active iodide uptake in the thyroid is mediated by the Na(+)/I(-) symporter (NIS), a key plasma membrane glycoprotein. Several NIS mutations have been shown to cause I(-) transport defect, a condition that, if untreated, can lead to congenital hypothyroidism and, ultimately, cretinism. The study of I(-) transport defect-causing NIS mutations provides valuable insights into the structure-function and mechanistic properties of NIS. Here we report the thorough analysis of the G395R NIS mutation. We observed no I(-) uptake activity at saturating or even supersaturating external I(-) concentrations in COS-7 cells transiently transfected with G395R NIS cDNA, even though we demonstrated normal expression of G395R NIS and proper targeting to the plasma membrane.					
https://pubmed.ncbi.nlm.nih.gov/12145342/					

RSID	Gene	Variant	Expected	Variant Allele	Your Genotype
rs200597118	SLCA5	R124H	G	A	N/A
Absence of NIS activity in R124H NIS-expressing cells.					
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3730242/					