ALPHA-TOCOPHEROL
BLOOD LEVELS

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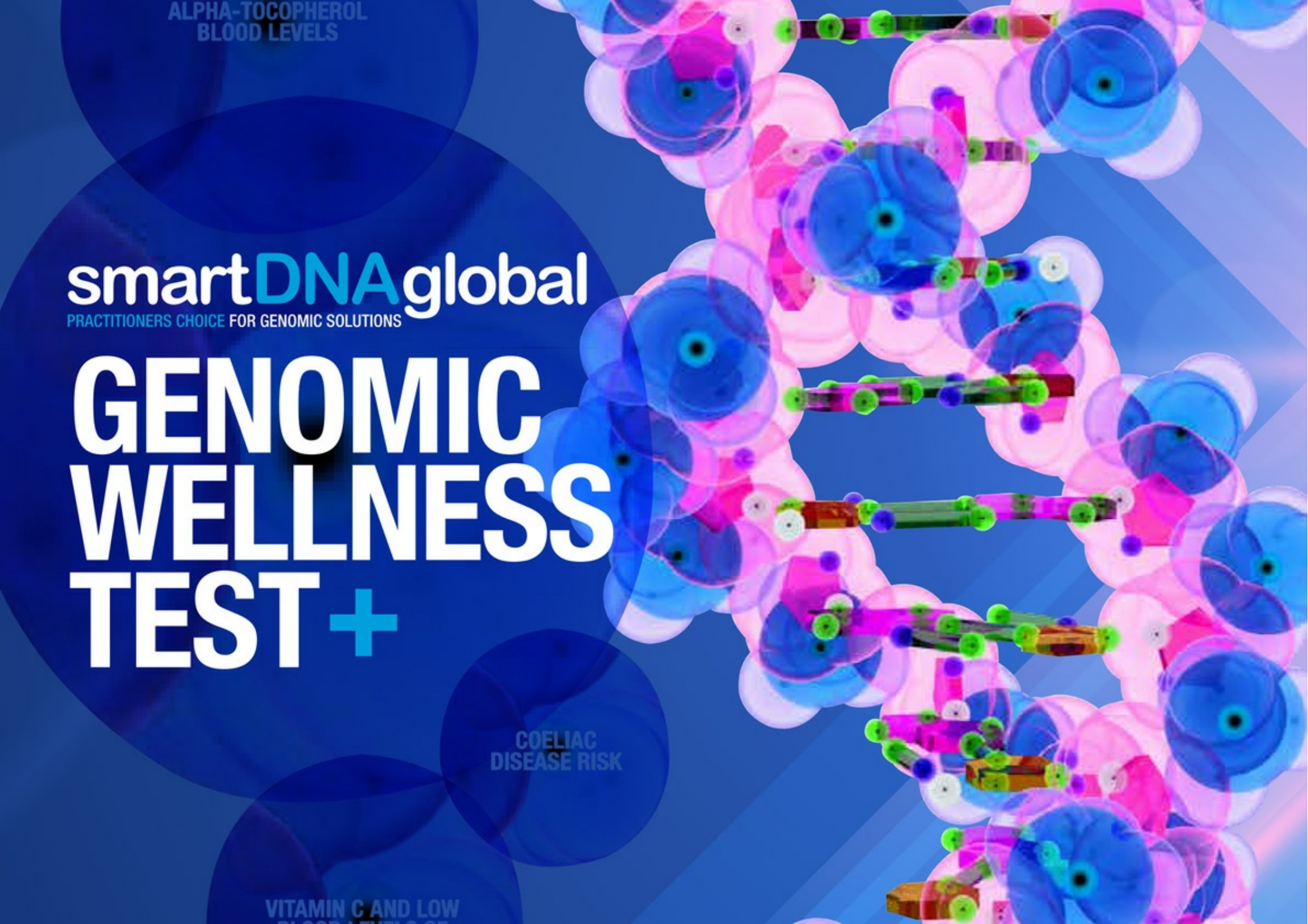
PRACTITIONERS CHOICE FOR GENOMIC SOLUTIONS

GENOMIC WELLNESS TEST+

COELIAC
DISEASE RISK

VITAMIN C AND LOW



Genetic Test Registration Information

Patient Identification

Patient Name:	Mrs Antonetta Calarco
Submission Number:	SNP 19-00303-50
Aliquot Number:	18730
Patient DOB:	15 December 1968
Patient ID Code:	41987
Patient Gender:	Female

Ordering Healthcare Professional

Requesting Practitioner:	Suzanne Ellis
Clinic Address:	NSW Australia

Laboratory Information

Specimen Source:	Saliva
Sample Collected:	5 February 2025
Sample Received:	12 February 2025
Sample Processed:	19 February 2025
Sample Reported:	5 March 2025

Lab Result Messages

No Messages

Test Performed / Method

Genotyping by sequenome based assay.

This report is solely intended as an educational tool and is not intended to treat or diagnose disease.
Each sample is run on arrays that have been analytically validated.

IMPORTANT NOTIFICATION FOR PRACTITIONERS:

The action steps contained in this report are provided as a guide for practitioners to discuss and review with their clients. The practitioner should consider the overall health status of their client before making recommendations.

Effect Legend

	No Action
	Practitioner Review/Action
	Practitioner Action
−/−	No risk allele has been inherited.
−/+	One risk allele has been inherited which has affected the enzyme activity or biological function in a pathway.
+/+	Homozygous: both risk alleles have been inherited with known effects on enzyme activity or biological function in a pathway.

Report Summary

Gene	Gene Variation	rs Number	Result		Effect	Link
Cardiovascular Disease Risk Section						
Homocysteine						
MTHFR	C677T	rs1801133	TT	+/+	🔴	View Result
MTHFR	A1298C	rs1801131	AA	−/−		
Hypertension						
ACE	G2328A	rs4343	GA	−/+	🟡	View Result
AGT	ATG>ACG	rs699	CT	−/+	🟡	View Result
Lipid Metabolism Section						
Lipid Transporter						
APOE	Cys130Arg	rs429358	TT	−/−	🟢	View Result
APOE	Arg176Cys	rs7412	CC	−/−		
HDL-C / Polyunsaturated Fat						
APOA1	5 Prime UTR Variant	rs670	GA	−/+	🟢	View Result
Plasma HDL-C Level						
CETP	Val422Ile	rs5882	AG	−/+	🟡	View Result
Plasma HDL-C - Alcohol Intake						
CETP	Intron Variant	rs708272	GG	+/+	🔴	View Result
Triglyceride Levels						
LPL	SNV	rs268	AA	−/−	🟢	View Result
APOCIII	Intron Variant	rs5128	CC	−/−	🟢	View Result
APOA5	Intron Variant	rs12286037	CT	−/+	🟡	View Result
APOA5	Intron Variant	rs662799	TT	−/−	🟢	View Result
NOS3	Asp298Glu	rs1799983	TT	+/+	🔴	View Result
LDL-C and Dietary Saturated Fat Intake						
APOB	T7673T	rs693	AG	−/+	🟡	View Result
APOB100	Intron Variant	rs754523	AA	−/−	🟢	View Result
LDL-R	Asn591Asn	rs688	TT	+/+	🔴	View Result
LDL Oxidation						
PON1	Gln192Arg	rs662	TT	−/−	🟢	View Result
LP(a)						
Lp(a)	Intron Variant	rs10455872	AA	−/−	🟢	View Result
Lp(a)	SNV	rs3798220	TT	−/−	🟢	View Result
HDL-C and Physical Activity Response						
LPL	Intron Variant	rs10096633	CT	−/+	🔴	View Result

Gene	Gene Variation	rs Number	Result		Effect	Link
Lipid Metabolism Section						
HDL-C and Physical Activity Response						
LIPC	Intron Variant	rs1800588	CC	+/+	🔴	View Result
CETP	Intron Variant	rs1532624	CC	+/+	🔴	View Result
APOA1 and Physical Activity Repsonse						
LPL	Intron Variant	rs10096633	CT	−/+	🟢	View Result
LIPC	Intron Variant	rs1800588	CC	+/+	🔴	View Result
CETP	Intron Variant	rs1532624	CC	+/+	🔴	View Result
Metabolic Syndrome and Diabetes Section						
LEPR	Lys656Asn	rs1805094	GC	−/+	🟡	View Result
ACSL1	Intron Variant	rs9997745	GG	+/+	🔴	View Result
ACC2	intron Variant	rs4766587	GG	−/−	🟢	View Result
TCF7L2	Intron Variant	rs7903146	TT	+/+	🔴	View Result
TCF7L2	Intron	rs12255372	GT	−/+	🟡	View Result
WFS1	Intron Variant	rs10010131	GA	−/+	🟡	View Result
SLC30A8	Arg325Trp	rs13266634	CT	−/+	🟡	View Result
IRS1	SNV	rs2943641	TC	−/+	🟡	View Result
HHEX	SNV	rs7923837	AA	−/−	🟢	View Result
HHEX	SNV	rs1111875	TT	−/−	🟢	View Result
KCNJ11	SNV	rs5219	CT	−/+	🟡	View Result
MTNR1B	SNV	rs10830963	CC	−/−	🟢	View Result
Fat Absorption, Fat Metabolism and Obesity Risk Section						
FTO	Intron Variant	rs9939609	TT	−/−	🟢	View Result
PPARG	Pro12Ala	rs1801282	CC	+/+	🔴	View Result
APOA2	Intron Variant	rs5082	TT	−/−	🟢	View Result
APOA5	Intron Variant	rs662799	TT	+/+	🔴	View Result
NOS3	Asp298Glu	rs1799983	TT	+/+	🔴	View Result
LIPC	Intron Variant	rs1800588	CC	−/−	🟢	View Result
FADS1	Intron Variant	rs174546	CC	−/−	🟢	View Result
FADS1	Intron Variant	rs174547	TT	−/−	🟢	View Result
FADS2	SNV	rs1535	AA	−/−	🟢	View Result
Carbohydrate Sensitivity Section						
AMY1	CNV	CNV	9		🟢	View Result

Report Summary

Gene	Gene Variation	rs Number	Result		Effect	Link
Nutrition and Low Grade Chronic Inflammation Section						
IL-6	Intron Variant	rs1800795	GC	−/+		View Result
TNFA	Intron Variant	rs1800629	AG	−/+		View Result
CRP	SNV	rs3093059	AA	−/−		View Result
Anti-inflammatory Cytokines Section						
IL-10	Intron Variant	rs1800871	CT	−/+		View Result
IL-10	Intron Variant	rs1800872	CA	−/+		View Result
IL-10	Intron Variant	rs1800896	AA	+/+		View Result
Pro-inflammatory Cytokines Section						
IL1-α	Ala114Ser	rs17561	GG	−/−		View Result
IL1-a2	Intron Variant	rs1800587	CC	−/−		View Result
IL1-β	Intron Variant	rs16944	CT	−/+		View Result
Periodontal Disease Risk/Inflammatory Disorders Section						
TNFA	Intron Variant	rs1800629	AG	−/+		View Result
IL1-α	Ala114Ser	rs17561	GG	−/−		View Result
IL1-β	Phe105Phe	rs1143634	CC	−/−		View Result
IL1-a2	Intron Variant	rs1800587	CC	−/−		View Result
IL-6	Intron Variant	rs1800795	GC	−/+		View Result
IL2	SNV	rs2069763	CC	−/−		View Result
1L-10	Intron Variant	rs1800896	AA	+/+		View Result
Food Responses Section						
Sodium Sensitivity						
AGT	ATG>ACG	rs699	CT	−/+		View Result
ACE	G2328A	rs4343	GA	−/+		View Result
Caffeine Metabolism						
CYP1A2	Intron Variant	rs762551	AA	−/−		View Result
Alcohol Flush						
ALDH2	SNV	rs671	GG	−/−		View Result
Gutbiome - Lactose, Coeliac, FodMap, Infections, Immune, Stress						
Lactose Intolerance						
MCM6	Intron Variant	rs4988235	TC	−/+		View Result

Gene	Gene Variation	rs Number	Result		Effect	Link
Coeliac Disease HLA 2.5/HLA DQ8/HLA DQ2.2						
DQ8	Single Nucleotide Variation	rs7454108	AA	−/−		
DQ2.5	Single Nucleotide Variation	rs2187668	CC	−/−		
HLA DQ2.2	Single Nucleotide Variation	rs2395182	TT	+/+		View Result
HLA DQ2.2	Single Nucleotide Variation	rs7775228	AG	−/+		
HLA DQ2.2	Single Nucleotide Variation	rs4713586	AA	+/+		
FUT2 Secretor Status						
FUT2	G428A	rs601338	AA	+/+		View Result
FUT2 / Gut Biome Bacterial Profile						
FUT2	G428A	rs601338	AA	+/+		View Result
FUT2 and Norovirus and Rotavirus Infection						
FUT2	G428A	rs601338	AA	−/−		View Result
Gut Biome and B12						
FUT2	SNV	rs492602	GG	−/−		View Result
Gut Microbiome Bifidobacterium						
APOA5	SNV	rs651821	TT	−/−		View Result
Gut Microbiome H. pylori						
TLR4	SNV	rs10759932	TT	+/+		View Result
Fodmap Diet Response						
SI	SNV	rs9290264	AC	−/+		View Result
IBS Immune Response						
TNFSF15	SNV	rs4263839	AG	−/+		View Result
Linolenic acid Metabolism Section						
FADS1	Intron Variant	rs174547	TT	−/−		View Result
Iron Status Section						
TMPRSS6	Asp512=	rs4820268	GA	−/+		View Result
TMPRSS6	SNV	rs855791	GA	−/+		View Result
TFR2	Intron Variant	rs7385804	CA	−/+		View Result
TF	Intron Variant	rs3811647	GA	−/+		View Result
TF	SNV	rs1799852	CC	−/−		View Result
Vitamins Section						
Vitamin A Metabolism						
BCMO1	Single Nucleotide Variant	rs11645428	GG	+/+		View Result
BCMO1	C1136T	rs7501331	CT	−/+		View Result

Report Summary

Gene	Gene Variation	rs Number	Result		Effect	Link
Vitamin B2 Metabolism						
MTHFR	C677T	rs1801133	TT	+/+		View Result
Vitamin B6 Metabolism						
NBPF3	Intron Variant	rs4654748	TT	−/−		View Result
Vitamin B12 Transport and Absorption						
TCN2	C766G	rs1801198	CC	−/−		View Result
FUT2	G772A	rs602662	AA	−/−		View Result
FUT2	G428A	rs601338	AA	−/−		View Result
Vitamin C Metabolism						
SLC23A1	G790A	rs33972313	GG	−/−		View Result
GSTT1	Insertion	CNV	Detected	PRESENT		View Result
GSTM1	Deletion	CNV	Not Detected	NULL		View Result
Vitamin E Metabolism						
INTERGENIC	Single Nucleotide Variant	rs12272004	AC	−/+		View Result
Vitamin D Metabolism						
DHCR7	Intron Variant	rs12785878	TT	−/−		View Result
GC	Intron Variant	rs2282679	CC	+/+		View Result
CYP2R1	UTR Variant	rs10741657	GG	+/+		View Result
VDR-FOK1	FOK1	rs2228570	TC	−/+		View Result
CYP24A1	Single Nucleotide Variant	rs6013897	TA	−/+		View Result
Hormones: Phase I Detoxification and Metabolism Section						
Phase I Metabolism of Estrogens						
CYP1A1	A2455G	rs1048943	AA	−/−		View Result
CYP1A1/1A2	Intergenic	rs2472297	CT	−/+		View Result
CYP1A1_M1	SNV	rs4646903	TC	−/+		View Result
Metabolism of estradiol to 4-OH-estradiol: Cytochrome P450 1B1						
CYP1B1	L432V	rs1056836	CC	−/−		View Result
CYP1B1	A10106G	rs1800440	AA	−/−		View Result
Phase I Steroidogenesis/ Androgen and Estrogen Metabolism						
CYP17A1	T-34C	rs743572	GG	+/+		View Result
CYP19A1	C19T	rs10046	CT	−/+		View Result
SRD5A2	G264C	rs523349	CC	+/+		View Result
UGT2B15	T253G	rs1902023	CA	−/+		View Result

Gene	Gene Variation	rs Number	Result		Effect	Link
Phase I Detoxification/Xenobiotic Metabolism						
CYP2C19	*17	rs12248560	CC	−/−		View Result
CYP3A4*1B	-392A>G	rs2740574	AA	−/−		View Result
CYP2A6	A>T	rs1801272	AA	−/−		View Result
CYP2D6	C>T	rs1065852	CT	−/+		View Result
CYP2D6	G>A	rs3892097	GG	−/−		
Hormones: Phase II Estrogen and Estrogen Metabolite Elimination Section						
COMT	Val158Met	rs4680	GA	−/+		View Result
GSTT1		CNV	Detected	PRESENT		View Result
GSTM1		CNV	Not Detected	NULL		View Result
GSTP1	Ile105Val	rs1695	AG	−/+		View Result
MnSOD	Val16Ala	rs4880	CC	−/−		View Result
Hormones: LHC, Progesterone and Thyroid Section						
LHCGR	A226096G	rs13405728	AA	−/−		View Result
PGR	Intronic	rs10895068	CC	−/−		View Result
FOXE1	Intronic	rs965513	GA	−/+		View Result
DIO1	Intronic	rs2235544	CA	−/+		View Result
DI01	UTR_variant	rs11206244	CT	−/+		View Result
DIO2	SNV	rs225014	TC	−/+		View Result
Hormones: Blood Pressure Section						
AGT	T803C	rs699	CT	−/+		View Result
ACE	G2328A	rs4343	GA	−/+		View Result
NOS3	G894T	rs1799983	TT	+/+		View Result
EDN1	SNV	rs5370	GG	−/−		View Result
Hormones: Coagulation Factors Section						
F5	R506Q	rs6025	GG	−/−		View Result
F2	G20210A	rs1799963	GG	−/−		View Result
Hormones: Fatty Acid and Choline Metabolism Section						
FADS1	G53A	rs174546	CC	−/−		View Result
FADS1	T61803311C	rs174547	TT	−/−		View Result
PEMT	523G>A	rs7946	AA	+/+		View Result
Phase II: Folate and Homocysteine Metabolism Section						
MTHFR	C677T	rs1801133	TT	+/+		View Result
MTHFR	A1298C	rs1801131	AA	−/−		

Report Summary

Gene	Gene Variation	rs Number	Result		Effect	Link
Phase II: Folate and Homocysteine Metabolism Section						
MTHFD1	C105T	rs1076991	CT	−/+		View Result
MTHFD1	G1958A	rs2236225	GG	−/−		View Result
SHMT1	C1420T	rs1979277	CC	−/−		View Result
Phase II: Homocysteine-Methionine Metabolism/Co-factors Section						
MTR	A2756G	rs1805087	AA	+/+		View Result
MTRR	A66G	rs1801394	AG	−/+		View Result
TCN2	C766G	rs1801198	CC	−/−		View Result
SLC19A1	Intron Variant	rs4819130	TT	−/−		View Result
Phase II: Homocysteine-Methionine Metabolism Section						
GNMT	SNV	rs11752813	GC	−/+		View Result
Phase II: Homocysteine Metabolism/Transulphuration Pathway Section						
CBS	C699T	rs234706	TT	+/+		View Result
Phase II: Anti-oxidant Enzymes Section						
MnSOD	Val16Ala	rs4880	CC	−/−		View Result
CAT	-262G>A	rs1001179	GG	−/−		View Result
GPX1	Pro200Leu	rs1050450	CC	−/−		View Result
PON1	Q192R	rs662	TT	−/−		View Result
SLC23A1	G790A	rs33972313	GG	−/−		View Result
HMOX-1	SNV	rs2071746	AA	−/−		View Result
NQO1	C609T	rs1800566	CT	−/+		View Result
Phase II Detoxification: Glutathione Enzymes Section						
GCLC	C53410037T	rs17883901	GG	−/−		View Result
GCLM	C590T	rs41303970	GG	−/−		View Result
GSTP1	Ile105Val	rs1695	AG	−/+		View Result
GSTP1	C341T	rs1138272	CC	−/−		View Result
GSTT1		CNV	Detected	PRESENT		View Result
GSTM1		CNV	Not Detected	NULL		View Result
Phase II Detoxification: N-acetyltransferase 2 (NAT2) Section						
NAT2	Tag SNP G>A	rs1495741	GA	−/+		View Result
Phase II: Amine Metabolism Section						
MAO-A	Intron Variant	rs909525	GA	−/+		View Result
Dopamine metabolism						
COMT	Val158Met	rs4680	GA	−/+		View Result

Gene	Gene Variation	rs Number	Result		Effect	Link
Histamine Metabolism						
AOC1/DAO	G4586T	rs2052129	CA	−/+		View Result
AOC1/DAO	His664Asp	rs1049793	CC	−/−		View Result
AOC1/DAO	Ser332Phe	rs1049742	CC	−/−		View Result
AOC1/DAO	Thr16Met	rs10156191	CT	−/+		View Result
Histamine Degradation						
HNMT	Thr105Ile	rs11558538	CC	−/−		View Result
HNMT	T939C	rs1050891	AA	+/+		View Result
Neurotransmitters, Mood, Cognition and Circadian Rhythm Section						
MAO-A	Intron Variant	rs909525	GA	−/+		View Result
MAO-A	R297R	rs6323	GT	−/+		View Result
COMT	Val158Met	rs4680	GA	−/+		View Result
DRD2/ANNK1	Taq1a	rs1800497	CC	−/−		View Result
TH	C824T	rs10770141	CT	−/+		View Result
BDNF	Val66Met	rs6265	AG	−/+		View Result
GAD1	147G>A	rs3749034	GA	−/+		View Result
Memory Performance						
KIBRA	Intron Variant	rs17070145	CC	+/+		View Result
Stress Reponses - HPA axis						
MR	Intron Variant	rs5522	AA	−/−		View Result
MR	Intron Variant	rs56149945	TT	−/−		View Result
Circadian Rhythm						
CLOCK	Intron Variant	rs1801260	TC	−/+		View Result
CLOCK	Intron Variant	rs2412646	GA	−/+		View Result
NPAS2	Intron Variant	rs6725296	GA	−/+		View Result
NPAS2	Intron Variant	rs2305160	AG	−/+		View Result
Weight Management Section						
PPARD	Intron Variant	rs2016520	GA	−/+		View Result
APOA2	Intron Variant	rs5082	TT	−/−		View Result
APOA5	Intron Variant	rs662799	TT	+/+		View Result
TAS2R38	Ala49Pro	rs713598	GG	−/−		View Result
FTO	Intron Variant	rs9939609	TT	−/−		View Result
MC4R	Intron Variant	rs17782313	TT	−/−		View Result
DRD2/ANNK1	Taq1a	rs1800497	CC	−/−		View Result

Report Summary

Gene	Gene Variation	rs Number	Result		Effect	Link
Weight Management Section						
SLC2A2	Intron Variant	rs5400	CC	−/−		View Result
ADIPOQ	Intron Variant	rs17366568	GG	−/−		View Result
ADRB2	Arg16Gly	rs1042713	AG	−/+		View Result
ADRB3	Trp64Arg	rs4994	TT	−/−		View Result
ADIPOQ	Intron Variant	rs17300539	GG	+/+		View Result
FTO	Intron Variant	rs1558902	AT	−/+		View Result
FTO	Intron Variant	rs1121980	CT	−/+		View Result
Sports and Exercise Section						
INSIG2	Intron Variant	rs7566605	GG	−/−		View Result
COL1A1	Intron Variant	rs1800012	GG	−/−		View Result
VDR-FOK	FOK1	rs2228570	TC	−/+		View Result
VEGFR2	Gln472His	rs1870377	AA	−/−		
ACE	Intron Variant	rs4341	CG	−/+		
ACE	Thr776=	rs4343	AG	−/+		View Result
ACTN3	Arg620Ter	rs1815739	CC	−/−		
HIF1	Pro583Ser	rs11549465	CT	−/+		
HIF1	Pro583Ser	rs11549465	CT	−/+		View Result
MCT-1	Asp490Glu	rs1049434	AT	−/+		View Result
eNOS3	Intron Variant	rs2070744	CC	+/+		View Result
ACE	Intron Variant	rs4341	CG	−/+		View Result
ACE	Thr776=	rs4343	AG	−/+		View Result
IL-6	Intron Variant	rs1800795	GC	−/+		View Result
COL5A1	Intron Variant	rs12722	CT	−/+		View Result
Concussion Recovery						
SLC17A7	SNV	rs74174284	CG	−/+		View Result

Cardiovascular Disease Risk Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Homocysteine

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin B9: Methylenetetrahydrofolate Reductase				
MTHFR rs1801133	C677T	TT +/+		The 'TT' genotype is associated with 70% reduced MTHFR enzyme activity and an increased risk of reduced folate metabolism and elevated homocysteine level. There is an increased risk of low serum folate and elevated homocysteine levels occurring if the dietary intake of folate and other B group vitamins is not optimal. Low vitamin B2 is associated with reduced MTHFR activity with the 677TT genotype. Please note that enzyme activity requires adequate intake of B vitamins and consideration of other factors such as alcohol intake and medications.
MTHFR rs1801131	A1298C	AA −/−		

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Hypertension

Gene SNP ID	Gene Variation	Result	Effect	Comment
Angiotensin-converting enzyme 2				
ACE rs4343	G2328A	GA −/+		The 'GA' genotype is associated with an increased risk of high blood pressure and heart disease when consuming a diet high saturated fat. Review dietary saturated fat intake.
Angiotensinogen				
AGT rs699	ATG>ACG	CT −/+		The 'CT' genotype is associated with an increased risk of developing hypertension. It has been reported that a healthy diet which includes vegetables, such as spinach, broccoli, carrots, fruits, such as apples, oranges, a fish, particularly those rich in omega-3 fatty acids, lean cuts of beef or pork, skinless chicken or turkey eggs and lower-fat dairy products, such as cheese and yogurt can help to lower blood pressure. Lifestyle contributions associated with reducing hypertension include reducing sodium intake, limiting alcohol, quitting smoking, a healthy sleep wake cycle and reducing stress.

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Table 1: LIPID TYPES, GENERAL DIETARY GUIDELINES AND EXERCISE

Cholesterol profiles ought to be used to monitor each individual with respect to HDL-C, LDL-C and triglyceride levels.

		APOE ε2 General Dietary Guidelines		APOE ε3 General Dietary Guidelines		APOE ε4 General Dietary Guidelines	
Lipid Metabolism Type		LMT A.1	LMT A.2	LMT B.1	LMT B.2	LMT C.1	LMT C.2
Genotype		ε2/ε2	ε2/ε3	ε3/ε3	ε2/ε4	ε3/ε4	ε4/ε4
Population Frequency		1%	10-15%	50-65%	2%	20-25%	2-5%
Soluble Fibre[1]		YES		YES		YES	
Fish Oils[2]		YES		YES		YES[*1]	
Energy Sources[3,4,6]	Fat	35%	30%	25%	25%	20%	20%
	Protein	15%	15%	20%	20%	25%	25%
	Carbohydrate	50%	55%	55%	55%	55%	55%
Moderate Alcohol[5,6]		HDL ↑ LDL ↓		HDL ↑		HDL ↓ LDL ↑	
		BENEFICIAL		**BENEFICIAL**		**Not Beneficial[*2]**	
Exercise[6]	Aerobic Based	55%	55%	50%	50%	75%	75%
	Strength Based	45%	45%	50%	50%	25%	25%

References: 1(a) Wolver et al. Am J Clin Nutr 66, 584-90 (1997) 1(b) Jenkins et al Metabolism 42, 585-93 (1993). 2(a) Varvel et al. www.hdlabinc.com/sciencebulletin 2(2) 2012 (b) Olano-Martin E Atherosclerosis 209; 104-110 (2010) 3. Masson LF et al. Am J clin Nutr 77:1098-111 (2003) 4. Moreno JA et al. 134:2517-2522 (2004) 5. a) Corella D et al. Am J clin Nutr 73:736-45 (2001) b) Marque-Vidal et al. Obes Res 11:1200-6 (2003) c) Mukamal KJ et al., Atherosclerosis 173:79-87 (2004) d) Bleich S et al. J Neural Trans 110:401-11 (2003) e) Lussier Cacan et al. Arterioscler Thromb Vasc Biol 1:22:824-31 (2002) 6. a) www.ApoegeneDiet.com b) Bernstein et al. Arterioscler Thromb Vasc Biol 22:133-140 (2002) [*1] Minihane et al Arterioscler Thromb Vasc Biol 20; 1990-1997 (2000) and Olano-Martin E et al. Atherosclerosis 209:10-4-110 (2010) reported genotype x treatment interaction in response to fish oil treatment. [*2] Males are more susceptible than females in this Geno-group to the effects of alcohol on HDL-C and LDL-C. The information for each geno-group does not mean that an individual should be treated equivocally.

Lipid Metabolism Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Lipid Transporter

Genetic variations detected in the Lipid Panel have been associated with inefficient lipid transportation, lipid absorption and lipid metabolism. Dietary changes in particular responses to polyunsaturated fats (PUFA), omega- 6 fatty acids and saturated fats and exercise may improve HDL-C, LDL-C, triglyceride level and fat absorption. Dietary fats are broken down by our digestive system into smaller molecules which are then absorbed into the blood stream. The measurable level of fats in the blood is due to a combination of the fats consumed from the diet and our genes. Review - Antioxidants, Inflammation and Folate Metabolism/Homocysteine sections of the report.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Apolipoprotein E				
APOE rs429358	Cys130Arg	TT −/−		The APO E3/E3 genotype is associated with neutral lipid metabolism and intermediate antioxidant capacity when compared to E2 and E4 genotypes. Other diet types suitable for this genotype are a Low Glycaemic diet, Intermittent Fasting, Paleo Diet. Avoid refined carbohydrates. Refer to Table 1 for additional information relating to this genotype.
APOE rs7412	Arg176Cys	CC −/−		

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HDL-C / Polyunsaturated Fat

The well-established inverse relationship between plasma HDL-C levels and the risk of coronary artery disease (CAD) has led to an extensive search for genetic factors influencing HDL-C concentrations. Environmental and metabolic factors that are commonly associated with low HDL-C concentrations include alcohol consumption, dietary saturated fat intake, decreased exercise, cigarette smoking, obesity, and diabetes. In addition to environmental factors, strong evidence also exists for the role of genetics in determination of HDL-C level.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Apolipoprotein A-I				
APOA1 rs670	5 Prime UTR Variant	GA −/+		The 'GA' genotype is associated with increased HDL-C levels. If HDL levels are sub-optimal then increase PUFA intake to 6-8% of calories has been reported to improve HDL-C. An improved response in females has been reported when compared to males. This result does not indicate that HDL-C levels are optimal.

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Plasma HDL-C Level

Gene SNP ID	Gene Variation	Result	Effect	Comment
Cholesterol Ester Transfer Protein				
CETP rs5882	Val422Ile	AG −/+		The 'AG' genotype is associated with reduced plasma CETP activity and increased HDL-C level however there is no benefit from plant sterols in lowering triglyceride levels. The CETP gene functions by removing cholesterol from HDL particles and transferring it to other lipid-carrying particles, such as LDL, in a process termed reverse cholesterol transport (RCT). The net result of CETP function is the redistribution of HDL to LDL.

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Plasma HDL-C - Alcohol Intake

Gene SNP ID	Gene Variation	Result	Effect	Comment
CETP rs708272	Intron Variant	GG +/+		The 'GG' genotype is associated with increased plasma CETP activity, reduced HDL level and increased dyslipidemia risk. This genotype is not associated with improved HDL-C levels in response to moderate alcohol intake (defined as 1-2 drinks per day). Reduce alcohol consumption if HDL-C levels are sub-optimal.

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Triglyceride Levels

A triglyceride (TG, triacylglycerol, TAG, or triacylglyceride) is an ester derived from glycerol and frees fatty acids. Triglycerides are a blood lipid that enables the bidirectional transference of adipose fat and blood glucose from the liver. Diets high in refined carbohydrates, with carbohydrates accounting for more than 60% of the total energy intake, can increase triglyceride levels. There is strong correlation for those with a BMI higher than 28 and insulin resistance. If triglyceride levels are elevated; reduce calorie intake from alcohol, saturated fats, refined carbohydrates, and sugar. Increase fibre intake from legumes, wholegrains, and leafy green vegetables. Intake omega-3 fatty acids from fatty fish 2-3 times a week is recommended. Review the CETP gene rs5882 in relation to plant sterol response lowering triglyceride levels.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Lipoprotein Lipase				
LPL rs268	SNV	AA −/−		The 'AA' genotype is associated with typical risk for elevated triglycerides.
Apolipoprotein C3				
APOCIII rs5128	Intron Variant	CC −/−		The 'CC' genotype is not associated with impaired metabolic health. If triglycerides are raised decrease saturated fat intake, increase MUFA intake and avoid processed carbohydrates. Focus on a Mediterranean diet and avoid a typical Western diet. The 'CC' genotype is more responsive to dietary intervention when compared to the 'GG' genotype.
Apolipoprotein A5				
APOA5 rs12286037	Intron Variant	CT −/+		The 'CT' genotype is associated with an increased risk of hypertriglyceridemia. Genetics, diet, and lifestyle contribute triglyceride levels. If triglyceride levels are elevated then limit the intake of sugary foods, foods high in saturated fat, processed carbohydrates, and alcohol. Monitor triglyceride levels.
APOA5 rs662799	Intron Variant	TT −/−		The 'TT' genotype is associated with a typical risk of having elevated triglyceride levels and no increased cardiovascular risk under the age of 45 years. Genetics, diet, and lifestyle contribute triglyceride levels. If triglyceride levels are elevated then limit the intake of sugary foods, foods high in saturated fat, processed carbohydrates, and alcohol. However, there is a direct correlation with increasing BMI and dietary fat intake in individuals homozygous for rs662799 'TT' genotype, BMI increased as total fat intake increased. Conversely, this increase was not present in carriers of the 'C' minor allele. Of the specific fatty acid groups analysed, monounsaturated fatty acids showed the highest statistical significance for this specific interaction. Please refer to the Fat Absorption/Metabolism and Obesity Risk section of this report for additional details.
Endothelial Nitric Oxide Synthase				
NOS3 rs1799983	Asp298Glu	TT +/+		The risk 'T' allele of rs1799983 in the NOS3 gene is associated with a reduction in NOS3 enzyme activity, increased cardiovascular risk markers such as total cholesterol and LDL, and increased risk of high blood pressure. Pregnant females are at increased risk of pre-eclampsia hypertension. Nitric oxide is an antioxidant involved in a broad range of broad range of biological processes including vasodilation of blood vessels. A low-fat dietary intervention has shown that 1.2-1.6 g or 1240mg of EPA and DHA omega-3 fatty acid/day as benefits for those with this variation. Dietary intake of omega 3 rich foods includes salmon, mackerel, and trout. Plant based sources of omega 3 are chia seeds, walnuts, and flaxseed.

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LDL-C and Dietary Saturated Fat Intake

The gene polymorphisms tested are associated with elevated LDL-C level in response to dietary saturated fat intake. Apolipoprotein B (APOB) is one of the main carriers of LDL cholesterol throughout the body and into the cells. The Apolipoprotein B gene encodes for APOB which is the main apolipoprotein of chylomicrons and low-density lipoproteins. APOB occurs in plasma as two main isoforms, apoB-48, and apoB-100; the former is synthesized exclusively in the gut and the latter in the liver. Apolipoprotein B-100 (APOB100) is a key component of LDL-C with an important role in the binding of LDL to the LDL receptors. Genetic variants in the APOB gene can lead to high LDL levels and an increased risk of heart disease. The Low-Density Lipoprotein Receptor (LDL-R) plays a crucial role in lipid metabolism being responsible for the uptake of lipoproteins into the cells.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Apolipoprotein B				
APOB rs693	T7673T	AG −/+		The 'AG' genotype is associated with an increased risk of increased LDL-C, increased triglycerides, increased APOB and increased risk of heart disease. A diet high in saturated fat is associated with elevated LDL-C. Research has shown that there is an inverse relationship between APOB levels and cardiovascular health. Review dietary saturated fat intake.
Apolipoprotein B100				
APOB100 rs754523	Intron Variant	AA −/−		The 'AA' genotype is not associated with an increased risk of elevated LDL-C.
Low-density Lipoprotein Receptor				
LDL-R rs688	Asn591Asn	TT +/+		The 'TT' genotype is associated with an increased risk of elevated LDL-C. A diet high in saturated fat is associated with elevated LDL-C. The presence of the rs688(T) allele is associated with an increased total and LDL-cholesterol in female members of the Framingham Offspring Study. The 'T' allele was not associated with significant differences in HDL-cholesterol. The largest association in relation to cholesterol differences were observed in pre-menopausal women. The 'T' allele is present in approximately 60% of Caucasians, is associated with significant 10% increases in total and LDL-cholesterol in pre-menopausal women. Review dietary saturated fat intake.

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LDL Oxidation

Gene SNP ID	Gene Variation	Result	Effect	Comment
Paraoxonase 1				
PON1 rs662	Gln192Arg	TT −/−		The 'TT' genotype is associated with efficient enzyme activity when compared to the "TC" and "CC" genotypes. Paraoxonase 1 (PON1) is an antioxidant having an important role in preserving the integrity of cell membranes. The "TT" genotype is associated with preventing LDL oxidation and a lower cardiovascular risk when compared to those carrying a 'C' allele.

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LP(a)

Lp(a) is a plasma lipoprotein synthesized by the liver that is composed of a low-density lipoprotein (LDL) molecule, a high molecular weight glycoprotein apolipoprotein(a), and a single molecule of apolipoprotein(B). The increased concentration of Lp(a) has been associated with incidence and severity of cardiovascular disease (CVD), coronary artery disease (CAD), peripheral artery disease, and stroke.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Lipoprotein(a)				
Lp(a) rs10455872	Intron Variant	AA −/−		The 'AA' genotype is not associated with elevated LP(a) levels.
Lp(a) rs3798220	SNV	TT −/−		The 'TT' genotype is associated with a typical risk of having higher plasma lipoprotein(a) or LPA.

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HDL-C and Physical Activity Response

There is genetic variability in the response of HDL-C levels to exercise. The mean HDL-C response to exercise is calculated by adding together the three delta scores obtained from the genotypes when the exercise >8.8 METS. A low HDL-C response is 7.9, an average HDL-C response is 11 and the highest HDL-C response to exercise is 12.5. Add the delta scores together for each genotype to obtain an indication of HDL-C improvement in relation to exercise.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Lipoprotein Lipase				
LPL rs10096633	Intron Variant	CT −/+		Reduced HDL-C response to exercise delta score 2.2; exercise of more than 8 METS/per week improves HDL-C levels. This genotype is optimal for HDL-C response to exercise. Review the results for LPL, LIPC and CETP.
Hepatic Lipase				
LIPC rs1800588	Intron Variant	CC +/+		Reduced HDL-C response to exercise delta score 3.1; this genotype is associated with reduced improvement in HDL-C levels when exercise is less than 8 METS/per week. If one or more genotype results indicates reduced improvement, then the individuals HDL response to exercise may be less than expected. Review the results for LPL, LIPC and CETP.
Cholesterol Ester Transfer Protein				
CETP rs1532624	Intron Variant	CC +/+		Reduced HDL-C response to exercise delta score 2.6; this genotype is associated with reduced improvement in HDL-C levels when exercise is less than 8 METS/per week. Review the results for LPL, LIPC and CETP.

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APOA1 and Physical Activity Repsonse

There is genetic variability in relation to APOA1 levels to exercise. The mean APOA1 response to exercise is calculated by adding together the three delta scores obtained from the genotypes when the exercise >8.8 METS. A low APOA1 response is 8.7, an average APOA1 response is 12.2 and the highest APOA1 response to exercise is 16.9. Add the delta scores together for each genotype to obtain an indication of APOA1 improvement in relation to exercise.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Lipoprotein Lipase				
LPL rs10096633	Intron Variant	CT −/+		Improved APOA1 response to exercise delta score 2.6; exercise of more than 8 METS/per week improves APOA1 levels. This genotype is associated with a moderate improvement of the APOA1 gene in response to exercise. Review the results for LPL, LIPC and CETP.
Hepatic Lipase				
LIPC rs1800588	Intron Variant	CC +/+		Reduced APOA1 response to exercise delta score 3.8; this genotype is associated with reduced improvement in APOA1 levels when exercise is less than 8 METS/per week. Review the results for LPL, LIPC and CETP.
Cholesterol Ester Transfer Protein				
CETP rs1532624	Intron Variant	CC +/+		Reduced APOA1 response to exercise delta score 3.3; this genotype is associated with reduced improvement in APOA1 levels when exercise is less than 8 METS/per week. Review the results for LPL, LIPC and CETP.

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Table 2: Mean HDL-C (mg/dl) Levels per Copy of the Minor Allele at Significant SNPs in the Entire Cohort and Across Median Levels of Physical Activity.

Gene rs Number	MET hours/week (Metabolic equivalent)	Number in Each Group	Mean (SD) HDL-C per allele, copy mg/dl		
			0 **CC**	**1** **CT**	**2** **TT**
LPL rs10096633	≤8.8	11,445	51.5	53.9	54.1
	>8.8	11,493	55.2	56.1	57.7
Delta (HDL-C & MET Physical activity)	N/A	N/A	3.7	2.2	3.6
LIPC rs1800588	≤8.8	11,445	51.3	53.0	54.4
	>8.8	11,491	54.4	56.8	59.3
Delta (HDL-C & Physical activity)	N/A	N/A	3.1	3.8	4.9
Gene rs Number	MET hours/week (Metabolic equivalent)	Number in Each Group	Mean (SD) HDL-C per allele, copy mg/dl		
			0 **CC**	**1** **CA**	**2** **AA**
CETP rs1532624	≤8.8	11,065	50.0	52.2	55.5
	>8.8	11,130	52.6	55.8	59.4
Delta (HDL-C & Physical activity)	N/A	N/A	2.6	3.6	3.9

Adapted from: Ahmad T et al. Physical Activity Modifies the Effect of LPL, LIPC and CETP polymorphisms on HDL-C levels and the Risk of Myocardial Infarction in Caucasian Women. Circulation and Cardiovascular Genetics 4(1), 74-80 (2011). The delta score in red refers to the mean increase in mg/dl for each genotype. For example LPL rs 10096633 CC genotype indicates a 3.7 mg/dl increase in HDL-C when exercise is >8.8 METS.

Table 3: Mean APOA1 (mg/dl) Levels per Copy of the Minor Allele at Significant SNPs in the Entire Cohort and Across Median Levels of Physical Activity.

Gene rs Number	MET hours/week (Metabolic equivalent)	Number in Each Group	Mean (SD) HDL-C per allele, copy mg/dl		
			0 CC	1 CT	2 TT
LPL rs10096633	≤8.8	11,390	148.1	151.1	152.4
	>8.8	11,443	153.0	153.7	154.0
Delta (HDL-C & MET Physical activity)	N/A	N/A	4.9	2.6	1.6
LIPC rs1800588	≤8.8	11,390	147.4	150.6	154.8
	>8.8	11,441	151.2	155.7	161.1
Delta (HDL-C & Physical activity)	N/A	N/A	3.8	5.1	6.8
Gene rs Number	MET hours/week (Metabolic equivalent)	Number in Each Group	Mean (SD) HDL-C per allele, copy mg/dl		
			0 CC	1 CA	2 AA
CETP rs1532624	≤8.8	11,065	145.9	149.3	152.9
	>8.8	11,130	149.2	153.8	158.1
Delta (HDL-C & Physical activity)	N/A	N/A	3.3	4.5	5.2

Adapted from: Ahmad T et al. Physical Activity Modifies the Effect of LPL, LIPC and CETP polymorphisms on HDL-C levels and the Risk of Myocardial Infarction in Caucasian Women. Circulation and Cardiovascular Genetics 4(1), 74-80 (2011). The delta score in red refers to the mean increase in mg/dl for each genotype. For example LPL rs 10096633 CC genotype indicates a 4.9 mg/dl increase in ApoA1 level when exercise is >8.8 METS.

Metabolic Syndrome and Diabetes Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

The long-chain acyl CoA synthetase 1 (ACSL1) and acetyl-CoA carboxylase (ACC2) plays a key role in fatty acid synthesis and oxidation. Disturbance of these pathways is associated with impaired insulin responsiveness and metabolic syndrome (MetS). Moreover, the ACSL1 and ACC2 gene polymorphisms are modulated by dietary fat intake. The Leptin receptor is associated with insulin resistance when PUFA intake is low. Genetic variations detected in the Transcription factor 7-like 2 (TCF7L2) and the Wolfram Syndrome 1 (WFS1) have been reported to play a role in insulin function. The Fat mass and obesity associated (FTO) gene and the Peroxisome Proliferator-Activated Receptor-Gamma (PPARG) gene are associated with an increased likelihood of developing type 2 diabetes due to a higher BMI (FTO), reduced control of blood glucose levels (PPARG) or reduced pancreatic beta cell function (SLC30A8). For individuals with yellow and red dots it may be necessary to assess fasting plasma glucose and glycated haemoglobin A1C.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Insulin Resistance: Leptin Receptor				
LEPR rs1805094	Lys656Asn	GC −/+		Review PUFA intake since the 'GC' genotype is associated with increased risk of insulin resistance when PUFA intake is low.
Metabolic syndrome (METs): Long-Chain-Fatty-Acid-CoA Ligase 1				
ACSL1 rs9997745	Intron Variant	GG +/+		The 'GG' genotype is associated with an increased metabolic syndrome (MetS) risk, elevated fasting glucose, elevated insulin concentrations and increased insulin resistance. A low fat diet <35% of energy or higher PUFA diet was reported to abolished MetS in affected individuals.
Metabolic syndrome (METs): Acetyl-CoA Carboxylase 2				
ACC2 rs4766587	intron Variant	GG −/−		The 'GG' genotype is not associated with an increased risk of metabolic syndrome.
Fat and whole grain food intake: Transcription Factor 7-like 2				
TCF7L2 rs7903146	Intron Variant	TT +/+		The 'TT' genotype is associated with an increased risk of developing insulin resistance and diabetes. Individuals with the 'TT' genotype experience less weight loss when compared to the 'CC' genotype. Long-term diet and exercise intervention is very important for 'T' allele carriers to prevent weight regain, insulin resistance and diabetes. 'T' allele carriers displayed lower reduction in BMI and total body fat when fat intake is high. Consuming whole grain foods can reduce the risk of developing T2D in individuals carrying the 'CT' and 'TT' genotype.

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Metabolic Syndrome and Diabetes Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Fat and whole grain food intake: Transcription Factor 7-like 2				
TCF7L2 rs12255372	Intron	GT −/+		The 'GT' genotype is associated with an increased risk of developing type 2 diabetes due to reduced beta cell function.
Diabetes Insulin secreation: Wolfram Syndrome 1				
WFS1 rs10010131	Intron Variant	GA −/+		The 'GA' genotype is associated with an increased incidence of type 2 diabetes. Review the portion size of carbohydrates in meals. Assess the intake of Low Glycaemic index carbohydrates in the diet.
Pancreatic beta cell function: Solute Carrier Family 30 Member 8				
SLC30A8 rs13266634	Arg325Trp	CT −/+		The 'CT' genotype is associated with increased risk of developing type 2 diabetes. Impaired insulin function may result in elevated blood glucose levels. Avoid refined carbohydrates. Include whole grain foods and complex carbohydrates in the diet.
Insulin Receptor Substrate 1				
IRS1 rs2943641	SNV	TC −/+		The 'TC' genotype is associated with a slightly higher risk of developing type 2 diabetes.
Homeobox				
HHEX rs7923837	SNV	AA −/−		The 'AA' genotype is associated with a typical risk of developing type 2 diabetes.
HHEX rs1111875	SNV	TT −/−		The 'TT' genotype is associated with a typical risk of developing type 2 diabetes.
Potassium inwardly rectifying channel subfamily J member 11				
KCNJ11 rs5219	SNV	CT −/+		The 'CT' genotype is associated with a 1.3x increased risk of developing type 2 diabetes.
Receptor for melatonin, a hormone regulating biorhythms				
MTNR1B rs10830963	SNV	CC −/−		The 'CC' genotype is associated with typical fasting glucose levels.

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Fat Absorption, Fat Metabolism and Obesity Risk Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Polymorphisms in FADS enzymes influence blood levels of omega-6 and omega-3 fatty acids. Individuals with the common minor allele variants in the FADS1-FADS2 gene cluster show blood lipid patterns indicative of slower desaturation. Those individuals with the FADS1/2 polymorphisms (as indicated by a yellow or red dot), may benefit by shifting their dietary intake from omega-6 and to omega-3 intake from fish oil. Flaxseed is a plant-based source of omega 3 however the alpha-linolenic acid must be converted by the FADS enzymes. If reduced enzyme activity is indicated, then there may be reduced benefit from intaking plant-based omega 3's.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Fat metabolism and Obesity Risk: Fat Mass and Obesity Associated				
FTO rs9939609	Intron Variant	TT −/−		The 'TT' genotype is not associated with an increased BMI or obesity. The Fat mass and obesity-associated protein (FTO) gene is expressed in many regions of the brain including the hypothalamus where it controls your food preferences, eating habits, appetite and feelings of fullness or satiety. Stay balanced.
Obesity Risk / Dietary Fats: Peroxisome Proliferator Activated Receptor Gamma				
PPARG rs1801282	Pro12Ala	CC +/+		The 'CC' genotype is found in 80% of the Caucasian population being associated with an increased BMI in response to a high fat diet and an increased risk of type 2 diabetes. PPARG is involved in the regulation of glucose and lipid metabolism, and it has been identified as the nuclear receptor for the thiazolidinedione class of insulin sensitising drugs. Your total fat intake should be balanced between saturated, monounsaturated, and polyunsaturated fat with focus on omega-3 fatty acids. Moderate aerobic exercise for 30 minutes five days a week.
Saturated Fat: Apolipoprotein A-II				
APOA2 rs5082	Intron Variant	TT −/−		The 'TT' genotype is not associated with obesity and increased fat consumption. Apolipoprotein A2 (APOA2), has a complex and largely undefined role in lipoprotein metabolism, insulin resistance, obesity and atherosclerosis susceptibility.

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Fat Absorption, Fat Metabolism and Obesity Risk Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Triglyceride (TGL) and high fat diet: Apolipoprotein A5				
APOA5 rs662799	Intron Variant	TT +/+		Individuals homozygous for the rs662799 'TT' genotype have been reported to increase BMI as total fat intake increased. Therefore, a diet high in total fat increases the risk of obesity. The 'TT' genotype is responsive to dietary intervention therefore decrease total fat intake to improve weight management outcomes. The 'TT' genotype is associated with a typical risk of having elevated triglyceride levels and no increased cardiovascular risk under the age of 45 years. Refer to the Triglyceride section of the report.
Triglyceride and omega-3 intake: Endothelial Nitric Oxide Synthase				
NOS3 rs1799983	Asp298Glu	TT +/+		The risk 'T' allele of rs1799983 in the NOS3 gene is associated with a reduction in NOS3 enzyme activity, increased cardiovascular risk markers such as total cholesterol and LDL, and increased risk of high blood pressure. Pregnant females are at increased risk of pre-eclampsia hypertension. Nitric oxide is an antioxidant involved in a broad range of broad range of biological processes including vasodilation of blood vessels. A low-fat dietary intervention has shown that 1.2-1.6 g or 1240mg of EPA and DHA omega-3 fatty acid/day as benefits for those with this variation. Dietary intake of omega 3 rich foods includes salmon, mackerel, and trout. Plant based sources of omega 3 are chia seeds, walnuts, and flaxseed.
Cholesterol/Triglyceride and Plasma Fat Balance: Hepatic Lipase				
LIPC rs1800588	Intron Variant	CC −/−		The 'CC' genotype is associated with typical hepatic lipase activity, typical cholesterol and triglyceride levels based on this genotype. The genetic result is associated with improved tolerance to the monounsaturated fats found in animal products. Animal-based monounsaturated fatty acids are beef, cheddar cheese, butter, bologna and other processed meats, and pork.
Processing of omega-3 and omega-6 fatty acids: Fatty Acid Desaturase 1				
FADS1 rs174546	Intron Variant	CC −/−		The 'CC' genotype is associated with typical blood levels of Arachidonic acid [ARA; 20:4(n-6)], or Eicosapentaenoic Acid [EPA; 20:5(n-3)] based on this FADS1 genotype.
Linolenic acid: Fatty Acid Desaturase 1				
FADS1 rs174547	Intron Variant	TT −/−		The 'TT genotype has no impact on dietary requirements of linoleic acid an n-6 polyunsaturated fatty acid (PUFA).
Processing of omega-3 and omega-6 fatty acids: Fatty Acid Desaturase 2				
FADS2 rs1535	SNV	AA −/−		The 'AA' genotype is associated with typical FADS2 enzyme activity.

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Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Carbohydrate Sensitivity Section

This AMY1 Copy Number Variation (CNV) is a marker which indicates sensitivity to carbohydrates. CNV's in the AMY1 gene are common and exist across different ethnic groups and geographic regions. Studies, suggest that low AMY1 CNVs are associated with increased cardiovascular disease risk and inflammation, but not glucose metabolism, in overweight or obese adults. A recent report has demonstrated that higher copy numbers of AMY-1 correlate with higher levels of Ruminococcaceae in the gut. The family of bacteria called Ruminococcaceae proliferates in the intestines when more salivary enzyme or amylase is present. Ruminococcaceae are known to break down resistant starch and it is thought that degradation of these hard to digest starches provides nutritional benefits.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Carbohydrate Sensitivity: Alpha-amylase 1				
AMY1 CNV	CNV	9		A higher copy number of the AMY1 gene is associated with increased levels of salivary amylase and a greater efficiency at digesting carbohydrates when compared to a lower AMY-1 gene copy number. It has recently been reported that higher copy numbers of AMY-1 correlate with higher levels of Ruminococcaceae in the gut. A higher copy number is defined as 5 or above.

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Nutrition and Low Grade Chronic Inflammation Section

The inflammatory response is necessary in relation to protection from infection however, chronic inflammation is involved in many disease states including diabetes, osteoporosis, obesity, aging and cardiovascular disease. Susceptibility to an increased inflammatory response is genetically determined. Common inflammatory cytokines known to be involved in chronic low-grade inflammation have been analysed.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Interleukin-6				
IL-6 rs1800795	Intron Variant	GC −/+		The 'GC' genotype is associated with increased gene expression. IL-6 has a complex disease association being associated with low grade chronic inflammation, autoimmune conditions, obesity, insulin resistance, dyslipidaemia and raised systolic blood pressure. If inflammation is present review omega-3 fatty acids intake, review the sodium sensitivity section in relation to hypertension risk, decrease saturated fat intake and maintain a healthy body weight.
Tumor Necrosis Factor-alpha				
TNFA rs1800629	Intron Variant	AG −/+		The 'A' allele is the proinflammatory allele. The 'A' allele is associated with increased TNF-alpha production and higher circulating levels of TNF-alpha in the blood when compared to carriers of the wild-type 'GG' genotype. The 'A' allele is associated with an increased risk for obesity, adiposity, dyslipidaemia, and insulin resistance especially when dietary fat intake is high. Reduce pro-inflammatory saturated fat and increase the intake of omega-3 fatty acids. Weight management and an anti-inflammatory diet are important in relation to this genotype. Like other inflammatory cytokines, TNF alpha levels increase during and after intensive exercise. Ensure extended recovery times. Polyunsaturated fatty acids (n-3 PUFA), glutamine, arginine and nucleotides can be included in a nutrition program when considering an inflammatory phenotype.
C-reactive protein				
CRP rs3093059	SNV	AA −/−		The 'AA' genotype is associated with a typical C reactive protein (CRP) plasma levels.

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Anti-inflammatory Cytokines Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Interleukin-10 (IL-10) is an anti-inflammatory cytokine which acts to inhibit inflammation causing cytokines. Chronic inflammation occurs when our anti-inflammatory cytokines do not control our inflammation causing cytokines. Excess body fat contributes to the production of pro-inflammatory cytokines. If there is a marked decrease of IL-10 production, then the control of pro-inflammatory cytokines maybe reduced. Connectivity: Review pro-inflammatory cytokines.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Interleukin-10				
IL-10 rs1800871	Intron Variant	CT −/+		The 'CT' genotype is associated with decreased IL-10 levels and a reduced ability to control pro-inflammatory cytokine production from TNFA, IL-8, IL-16, IL-1 alpha and IL-1 beta.
IL-10 rs1800872	Intron Variant	CA −/+		The 'CA' genotype is associated with decreased IL-10 levels and a reduced ability to control pro-inflammatory cytokine production from TNFA, IL-8, IL-16, IL-1 alpha and IL-1 beta.
IL-10 rs1800896	Intron Variant	AA +/+		The 'AA' genotype is associated with decreased IL-10 levels and a reduced ability to control pro-inflammatory cytokine production from TNFA, IL-8, IL-16, IL-1 alpha and IL-1 beta.

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Pro-inflammatory Cytokines Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Proinflammatory cytokines trigger a cascade associated with IL-6, IL8 and TNFA, CRP, IL-1beta. This genetic predisposition increases the risk of inflammation associated with obesity, infection, trauma, stress, cardiovascular disease, autoimmune diseases, and periodontal disease. The yellow and red dots indicate increased risk.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Interleukin 1 alpha IL1-α rs17561	Ala114Ser	GG −/−		The 'GG' genotype is not associated with increased IL1-alpha production.
Interleukin 1 alpha 2 IL1-a2 rs1800587	Intron Variant	CC −/−		The 'CC' genotype is not associated with increased IL1-alpha 2 production.
Interleukin 1 beta IL1-β rs16944	Intron Variant	CT −/+		The 'CT' genotype is associated with pro-inflammatory cascade with IL-6, IL-8 and TNF alpha.

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Periodontal Disease Risk/Inflammatory Disorders Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Proinflammatory cytokines trigger a cascade associated with IL-6, IL8 and TNFA, CRP, IL-1beta. This genetic predisposition increases the risk of inflammation associated with periodontal disease, obesity, infection, trauma, and stress. Specifically, the risk of periodontal disease is in part a consequence of the long-term effect of an elevated level of inflammation that can, in the presence of high levels of pathogenic bacteria, result in the destruction of gum tissue, the bone loss which may lead to tooth loss. The yellow and red dots indicate increased risk.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Tumor Necrosis Factor-alpha				
TNFA rs1800629	Intron Variant	AG −/+		The 'A' allele is the proinflammatory allele. The 'A' allele is associated with increased TNF-alpha production and higher circulating levels of TNF-alpha in the blood when compared to carriers of the wild-type 'GG' genotype. The 'AG' genotype increases the risk of periodontitis, migraines, many inflammatory related diseases including gum disease, increased acne risk, nasal polyps and sinus infections, Achilles and knee tendon inflammation and recurrent miscarriages.
Interleukin 1 alpha				
IL1-α rs17561	Ala114Ser	GG −/−		The 'GG' genotype is not associated with increased IL1-alpha production.
Interleukin-1 beta				
IL1-β rs1143634	Phe105Phe	CC −/−		The 'CC' genotype is not associated with increased IL-1B expression.
Interleukin 1 alpha2				
IL1-a2 rs1800587	Intron Variant	CC −/−		The 'CC' genotype is associated with typical IL1-alpha 2 production.
Interleukin-6				
IL-6 rs1800795	Intron Variant	GC −/+		The 'CG' genotype is associated with increased gene expression. The IL-6 'G' allele is associated with moderate to severe periodontitis and the presence of pathogenic periodontal bacteria.
Interleukin-2				
IL2 rs2069763	SNV	CC −/−		The 'C' allele is associated with a typical risk of developing periodontitis.
Interleukin-10				
1L-10 rs1800896	Intron Variant	AA +/+		The 'AA' genotype is associated with gum disease and reduced levels of IL-10 and therefore a reduced anti-inflammatory response.

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Food Responses Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Sodium Sensitivity

Discuss preventative measures in relation to hypertension and cardiovascular disease. Review the intake of processed foods, snacks, canned foods, cheeses, and meats since they have a high salt content.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Angiotensinogen				
AGT rs699	ATG>ACG	CT −/+		The 'CT' genotype is associated with an increased risk of sodium sensitivity associated with increased sodium intake. AGT is the gene that encodes for angiotensinogen, which is a precursor to both angiotensin I and angiotensin II.
Angiotensin-converting enzyme 2				
ACE rs4343	G2328A	GA −/+		The 'GA' genotype is associated with an increased risk of sodium sensitivity when associated with increased sodium intake. Review dietary sodium intake from cheeses, canned foods and processed meats.

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Caffeine Metabolism

Reduce caffeine consumption if it exceeds more than two cups of coffee per day since this may increase the risk of hypertension and heart attack. Reviewing the consumption of caffeinated products may be useful in assessing the individual's overall caffeine intake.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Cytochrome P450 1A2				
CYP1A2 rs762551	Intron Variant	AA −/−		The 'AA' genotype is associated with a reduced risk of heart attack with moderate coffee consumption when compared to those who consume no coffee at all.

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Alcohol Flush

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

The ALDH2 gene produces an enzyme needed for the elimination of acetaldehyde via its conversion to acetic acid. Individuals possessing either one or two copies of the 'A' allele experience alcohol-related sensitivity responses include facial flushing, severe hangovers and hence they are usually not regular drinkers.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Aldehyde dehydrogenase 2 protein				
ALDH2 rs671	SNV	GG -/-		The 'GG' genotype is associated with typical acetaldehyde metabolism.

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Gutbiome - Lactose, Coeliac, FodMap, Infections, Immune, Stress

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Your unique bacterial profile is determined by several factors in including your dietary intake. Approximately 50% of your gut microbiome is determined by your dietary intake. Other contributions to your gut microbiome profile are antibiotics, vaginal or C-section birth, other bacterial exposures, and your innate genome. This section reviews the impact that your genome has on the residents in your intestines.

Lactose Intolerance

The production of the lactase enzyme is regulated by the MCM6 gene. If gastrointestinal symptoms are persisting, then further studies may be warranted such as a hydrogen breath test. This test detects hydrogen because of lactose not being digested. Individuals may be intolerant due to secondary lactose intolerance. Review any medications that the individual may have been prescribed that may affect the bowel flora. This genetic analysis is for primary lactose intolerance not secondary lactose intolerance or congenital lactase deficiency.

Gene SNP ID	Gene Variation	Result	Effect	Comment
MCM6 minichromosome maintenance deficient 6				
MCM6 rs4988235	Intron Variant	TC −/+		The 'TC' genotype is associated with a slightly increased risk of lactose intolerance. If necessary, limit dairy intake and consider almond, rice, oat, soy, or lactose free milk alternatives. Individuals with this genotype may experience gastrointestinal symptoms such as flatulence, bloating, diarrhea and abdominal pain because of low lactase levels.

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Coeliac Disease HLA 2.5/HLA DQ8/HLA DQ2.2

People with coeliac disease are known to have one of either HLA DQ 2.5 (commonest), HLA DQ8 (less common) or HLA DQ2.2 (least common). Speak to your healthcare professional if you experience steatorrhea, diarrhea, cramps, fatigue, flatulence, or joint pain when consuming gluten-containing foods, or if you have a family member with coeliac disease. Having the genetics does not mean that you have coeliac disease. This test is not diagnostic of coeliac disease since only one in 30 people (approximately) with these genes will develop coeliac disease. This analysis is only helpful in relation to the negative predictive value; if the result is negative it means you are very unlikely to ever develop coeliac disease.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Coeliac Disease Risk				
DQ8 rs7454108	Single Nucleotide Variation	AA −/−		
DQ2.5 rs2187668	Single Nucleotide Variation	CC −/−		
HLA DQ2.2 rs2395182	Single Nucleotide Variation	TT +/+		HLA DQ2.2 is associated with a slightly increased risk of coeliac disease.
HLA DQ2.2 rs7775228	Single Nucleotide Variation	AG −/+		
HLA DQ2.2 rs4713586	Single Nucleotide Variation	AA +/+		

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FUT2 Secretor Status

The FUT2 gene encodes for the enzyme, fucosyltransferase 2, which controls whether the oligosaccharides or sugars that make up your blood type will be expressed in your bodily fluids; saliva, sweat, tears, vaginal secretions, semen, or digestive juices (other than your blood). Secretor status is completely independent of ABO blood type, an individual can be blood Type O secretor or blood type O non-secretor and so on for blood types A and B or AB. Approximately 18-20% of the population are non-secretors.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Galactoside 2-alpha-L-Fucosyltransferase 2				
FUT2 rs601338	G428A	AA +/+		The 'AA' genotype is associated with FUT2 non-secretor status, lower levels of Bifidobacterium and resistance to Norovirus infection. H. pylori colonization is also less in non-secretors. Children who are non-secretors are less likely to have diarrheal diseases. Non-secretors have a higher risk of type 1 diabetes, alcohol-induced pancreatitis, Crohn's disease, and adverse outcomes in premature infants. Fucosyltransferase 2 (FUT2) codes for an enzyme involved in the synthesis of oligosaccharides and mediates the expression of gastrointestinal mucosal ABO (blood group) antigens. Oligosaccharides secreted in the intestinal mucosa feed intestinal flora, thereby directly influencing microbial concentrations and diversity. FUT2 produces the prebiotic 2'-fucosyllactose (2'-FL) which an important nutrient for probiotic Bifidobacterium species. Bifidobacterium produce short chain fatty acids and the amino acids lysine, tryptophan, and tyrosine. Low levels of 2'-FL can lead to decreased B12 levels resulting in anaemia, elevated homocysteine levels and an impaired immune system. Gut dysbiosis, Irritable Bowel Syndrome, depression, anxiety is found in association with this genotype.

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FUT2 / Gut Biome Bacterial Profile

FUT2 produces the prebiotic 2'-fucosyllactose (2'-FL) which an important nutrient for probiotic Bifidobacterium species. Bifidobacterium produce short chain fatty acids and the amino acids lysine, tryptophan, and tyrosine.

Gene SNP ID	Gene Variation	Result	Effect	Comment
FUT2 rs601338	G428A	AA +/+		The 'AA' genotype is the FUT2 gene non-secretor of blood type associated with low or no Bifidobacteria (probiotic bacteria), lower Ruminococcus, Clostridium and Akkermansia.

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FUT2 and Norovirus and Rotavirus Infection

Approximately 20% of the worlds population harbours a genetic variant that not only prevents them from secreting their blood type but also protects non-secretors from getting norovirus and rotavirus.

Gene SNP ID	Gene Variation	Result	Effect	Comment
FUT2 rs601338	G428A	AA −/−		The 'AA' FUT2 nonsecretor genotype is associated with resistance to norovirus and rotavirus infection.

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Gut Biome and B12

A strong association exists between rs492602 in the FUT2 gene and plasma vitamin B12 levels in a genome-wide scan was reported (n = 1,658) and an independent replication sample (n = 1,059) from the Nurses' Health Study. Women homozygous for the rs492602(G) allele had higher B12 levels. This allele is in strong linkage disequilibrium with the FUT2 nonsecretor variant rs601338 which suggests a plausible mechanism for altered B12 absorption and plasma levels. Serum B12 levels may not accurately reflect the B12 transported into cells, a methylmalonic acid or MMA test may provide a more accurate assessment in non-secretors.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Galactoside 2-alpha-L-Fucosyltransferase 2				
FUT2 rs492602	SNV	GG −/−		The 'GG' genotype is associated with higher concentrations of plasma B12 levels in women. Serum B12 levels may not accurately reflect the B12 transported into cells, a methylmalonic acid or MMA test may provide a more accurate assessment in non-secretors.

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Gut Microbiome Bifidobacterium

The APOA5 gene not only affects plasma lipoprotein levels, including triglyceride levels, but levels of the probiotic Bifidobacterium gut bacteria. Please review the individuals FUT2 secretor status as this also has an impact on levels of Bifidobacterium species in the gut biome.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Apolipoprotein-A5				
APOA5 rs651821	SNV	TT −/−		The 'TT' genotype is associated with typical levels of Bifidobacterium in the gut microbiome. Please review the individuals FUT2 secretor status as this also has an impact on levels of Bifidobacterium species in the gut biome.

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Gut Microbiome H. pylori

It has been shown that TLR polymorphisms can influence H. pylori susceptibility. H. pylori-related diseases developed during modulation of pro-inflammatory or anti-inflammatory cytokines. Specifically, Toll-like receptor 4 (TLR4) innate immunity role and is protective against gram-negative bacterial infections. TLR4 variants have also been investigated in relation to gastric cancer risk, transplant rejection and vaccine responses. The risk of infection was reported using a recessive model whereby the 'TT' and 'CT' genotypes were compared to the 'CC' genotype. The recessive model of TLR4 rs10759932 showed a decreased risk of H. pylori susceptibility.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Toll-like receptor 4				
TLR4 rs10759932	SNV	TT +/+		The 'TT' genotype is associated with a typical risk of H. pylori infection when compared to the 'CC' genotype which has a decreased risk of H. pylori infection.

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Fodmap Diet Response

Sucrase-isomaltase has two functions; firstly, breaking down sucrose into glucose and fructose, and secondly breaking down some starches into glucose. Foods containing sucrose are sweet fruits and any foods with added sugar. So that's the sucrose component but what about the isomaltase component? Digestion begins in the mouth with salivary amylase however the pancreas secretes amylase into the small intestine to break apart the starches into their molecular components. This final breaking down of the starch molecules is performed by the sucrase-isomaltase enzyme (SI). SI is the major enzyme responsible for 60-80% of the starches and the enzyme maltase-glucoamylase breaks down the remaining 20-40%. Individuals harbouring genetic variants in the SI gene have an increased risk of developing IBS. Dietary intake of sugar and starch combined with a low production of the SI enzyme are associated with constipation and diarrhoea, gas, and bloating. A low fodmap diet does reduce by default some foods with starches and there may be a reduction in IBS symptoms simply because of the reduced dietary intake or starchy foods. However, it has been reported that individuals harbouring, an SI genetic variant are least likely to benefit on a low FODMAP diet. Food high in dietary starches are highly processed food such as in baked breads and cakes, cereals, pasta, gluten free foods, white rice. Fruits low in sugar and starch are berries, lemons, limes, grapefruit, and oranges.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Sucrase-isomaltase				
SI rs9290264	SNV	AC −/+		The 'AC' genotype is associated with reduced enzyme activity when compared to the 'CC' genotype. Reduced enzyme activity is associated with an increased risk of irritable bowel syndrome diarrhoea predominant (IBS-D) and irritable bowel syndrome mixed constipation and diarrhoea (IBS-M). IBS-M is an unstable form of IBS typified by fluctuations between constipation and diarrhoea.

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IBS Immune Response

Tumour necrosis factor (ligand) superfamily, member 15 codes for TNF-like ligand 1A (TL1A), which contributes to the modulation of inflammatory responses. The TLIA protein is expressed in immune cells promoting an inflammatory response in the gut mucosa. The 'G' allele (AG or GG) is associated with an increased risk of irritable bowel syndrome (IBS) and the IBS subtype constipation (IBS-C). Carrying the risk allele is suggested to result in higher TL1A expression and therefore increased T cell activation and immune response. The association with TNFS15 indicates that inflammatory response may be an important mechanism in IBS and IBS with constipation (IBS-C).

Gene SNP ID	Gene Variation	Result	Effect	Comment
Tumor necrosis factor (ligand) superfamily, member 15				
TNFSF15 rs4263839	SNV	AG −/+		The 'AG' genotype is associated with IBS or IBS-C in association with an increased immune response in the gut mucosa.

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Linolenic acid Metabolism Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

The SNP T61803311C or rs174547 is associated with altered lipid profiles with the ‘C’ allele being associated with reduced LDL cholesterol levels in individuals on a diet low in unsaturated fats. This correlated with an earlier study which showed that the largest effects related to this SNP were observed in those with a low unsaturated fat intake.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Linolenic acid: Fatty Acid Desaturase 1				
FADS1 rs174547	Intron Variant	TT −/−		The 'TT genotype has no impact on dietary requirements of linoleic acid an n-6 polyunsaturated fatty acid (PUFA).

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Iron Status Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Anaemia results from a lack of red blood cells or dysfunctional red blood cells in the body. This leads to reduced oxygen flow to the body's organs. Symptoms may include fatigue, skin pallor, shortness of breath, light-headedness, dizziness, or a fast heartbeat. Treatment depends on the underlying diagnosis. The TMPRSS6 gene produces the matriptase-2 protein which affects hepcidin levels which regulates iron balance. TMPRSS6 polymorphisms are associated with lower serum iron (SI) and hemoglobin levels, consistent with their associations to increased iron deficiency and anaemia (IDA) risk. The TFR2 gene produces the TFR2 protein which helps iron enter the cells. The TF gene produces the protein transferrin which is responsible for transferring iron in the body. In combination, these three genes indicate a genetic risk for having decreased iron status. However, it has been reported that polymorphisms in the TMPRSS6 gene are genetic risk factors for not only iron deficiency but moreover iron deficiency anaemia (IDA). In relation to exercise especially at the elite level it is well-known that low iron stores impact hemoglobin production which in turn decreases the oxygen carrying capacity of the blood, leading to a lack of oxygen to working muscles and resulting in impaired muscle contraction and aerobic endurance . As such, genetic markers that impact iron stores in response to intake can indirectly affect performance through the oxygen carrying capacity of hemoglobin. IDA is the most common type of anaemia among athletes, who have higher iron requirements due to increased erythropoietic drive through higher intensities and volumes of training. Female athletes are at particular risk of iron deficiency due to menstruation and generally, a lower total energy or food intake compared to males. Studies using the TMPRSS6, TFR2 and TF homozygous variants in a grouping or haplotype have reported increased risk.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Serum Iron Concentration: Transmembrane Protease Serine 6				
TMPRSS6 rs4820268	Asp512=	GA −/+		The 'GA' genotype in the TMPRSS6 gene is associated with lower serum iron (SI) and hemoglobin levels which is consistent with the associations for increased risk iron deficiency and iron deficiency anaemia (IDA). Review dietary sources of iron such as beef, chicken, fish, and organ meats. Iron levels outside of the typical range may result in undesirable health outcomes. Consider monitoring iron levels and ensuring that they are in the typical range. The recommended daily intake for iron is dependent upon several factors such as gender, age, stage of life and the dietary intake of iron rich foods. Vitamin C is important for the improved absorption of iron. The relationship between dietary vitamin C and circulating levels of ascorbic acid depend on an individual's GSTT1 genotype. Please review this in the Vitamin C section of this report. Individuals who do not meet the Recommended Dietary Allowance (RDA) for vitamin C are significantly more likely to be vitamin C deficient, as assessed by serum ascorbic acid levels than those who meet the RDA, but this effect is much greater in individuals with the GSTT1 NULL/NULL genotype than those with the PRESENT or INSERTED allele.
TMPRSS6 rs855791	SNV	GA −/+		The 'GA' genotype is associated specifically with lower ferritin or iron levels in men.
Serum Iron Concentration: Transferrin Receptor Protein 2				
TFR2 rs7385804	Intron Variant	CA −/+		The 'CA' genotype is associated with low iron status due to a reduced ability of iron to enter the cells. This can impact haematocrit, mean corpuscular volume, and red blood cell count where individuals with the 'CA' genotype have an increased risk of low serum iron levels. Consider monitoring iron levels and ensuring that they are in the typical range. The recommended daily intake for iron is dependent upon several factors such as gender, age, stage of life and the dietary intake of iron rich foods. Dietary requirements for iron are increased in those eating a plant-based diet such as vegans and vegetarians. Vitamin C is important for the improved absorption of iron. The relationship between dietary vitamin C and circulating levels of ascorbic acid depend on an individual's GSTT1 genotype. Please review this in the Vitamin C section of this report. Individuals who do not meet the Recommended Dietary Allowance (RDA) for vitamin C are significantly more likely to be vitamin C deficient, as assessed by serum ascorbic acid levels than those who meet the RDA, but this effect is much greater in individuals with the GSTT1 NULL/NULL genotype than those with the PRESENT or INSERTED allele.

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Iron Status Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Serum Iron Concentration: Tissue Factor				
TF rs3811647	Intron Variant	GA −/+		The 'GA' genotype is associated with an increased risk of low iron status. The TF gene produces the protein transferrin which is responsible to transferring iron in the body. Transferrin is an indicator of how much iron is available to tissues since transferrin controls the levels of free iron contained within our red blood cells. Consider monitoring iron levels and ensuring that they are in the typical range. The recommended daily intake for iron is dependent upon several factors such as gender, age, stage of life and the dietary intake of iron rich foods. Dietary requirements for iron are increased in those eating a plant-based diet such as vegans and vegetarians. Vitamin C is important for the improved absorption of iron. The relationship between dietary vitamin C and circulating levels of ascorbic acid depend on an individual's GSTT1 genotype. Please review this in the Vitamin C section of this report. Individuals who do not meet the Recommended Dietary Allowance (RDA) for vitamin C are significantly more likely to be vitamin C deficient, as assessed by serum ascorbic acid levels than those who meet the RDA, but this effect is much greater in individuals with the GSTT1 NULL/NULL genotype than those with the PRESENT or INSERTED allele.
TF rs1799852	SNV	CC −/−		The 'CC' genotype is associated with typical serum transferrin levels.

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Vitamins Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Vitamin A Metabolism

Beta carotene monooxygenase 1 (BCMO1) codes for an enzyme that converts beta carotene into retinal. Humans are unable to synthesise vitamin A. Vitamin A is a fat-soluble vitamin that is important for skin health, immunity, vision, cellular repair, and embryonic development. Beta-carotene is a precursor to active vitamin A which is an antioxidant found in red- and orange-coloured fruits and vegetables. The yellow and red dots generally indicate increased risk. The predicted impact of increased risk is determined by the combination of these genotypes. If the individual has both SNP's as heterozygous or homozygous or in combination, then the predicted impact of vitamin A deficiency is increased.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin A: Beta,beta-carotene 15,15'-dioxygenase				
BCMO1 rs11645428	Single Nucleotide Variant	GG +/+		The 'GG' genotype is associated with 51% reduced enzyme activity and reduced conversion beta-carotene into retinol. Retinol is essential for vision. Review dietary vitamin A intake.
BCMO1 rs7501331	C1136T	CT −/+		The 'CT' genotype is associated with reduced BCMO1 enzyme activity and conversion of Beta-carotene into vitamin A. Review dietary vitamin A intake.

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Vitamin B2 Metabolism

This section relates specifically to the MTHFR C677T or rs1801133 'TT' genotype and vitamin B2 dietary intake. A red dot indicates that there is a risk of higher levels of homocysteine in individuals with low levels of riboflavin or vitamin B2 intake. Active riboflavin supplementation is shown to reduce the homocysteine levels. Vitamin B2 is found in dairy milk, yogurt, cheese, eggs, lean beef and pork, organ meats (beef liver, chicken breast and salmon.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin B2: Methylenetetrahydrofolate Reductase				
MTHFR rs1801133	C677T	TT +/+		The MTHFR 'TT' genotype is associated with increased plasma levels of homocysteine when dietary intake of vitamin B2 is low. Review vitamin B2 intake.

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Vitamin B6 Metabolism

Vitamin B6 is necessary for the proper functioning of the neurological system, red blood cells and in sugar metabolism. This water-soluble vitamin, called pyridoxine, can lead to anaemia if present in low levels. Other symptoms of low vitamin B6 levels in the body are fatigue, inflammation of the tongue and mouth, cracks at the corner of the mouth and hyper homocysteinemia.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin B6 (Plasma): Neuroblastoma Breakpoint Member 3				
NBPF3 rs4654748	Intron Variant	TT −/−		The 'TT' genotype is associated with typical Vitamin B6 blood concentration.

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Vitamin B12 Transport and Absorption

TCN2 binds to vitamin B12 in the blood stream, transporting it to target cells and tissues. Approximately 30% of plasma vitamin B12 is bound to TCN2, and it is responsible for the majority of B12 transport into tissues. The yellow and red dots indicate lower holo- transcobalamin level and elevated homocysteine when vitamin B12 status is low. Review MTHFR, MTRR, MTR and FUT2 genotypes.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin B12: Transcobalamin-2				
TCN2 rs1801198	C766G	CC −/−		The 'CC' genotype is associated with typical vitamin B12 transport having an increased affinity for binding with vitamin B12.
Vitamin B12: Galactoside 2-alpha-L-Fucosyltransferase 2				
FUT2 rs602662	G772A	AA −/−		The 'AA' genotype is associated with typical vitamin B12 levels and reduced risk of vitamin B12 deficiency. However, this must be assessed since this FUT2 genotype is an FUT2 nonsecretor status. Low levels of 2'-FL can lead to decreased B12 levels resulting in anaemia, elevated homocysteine levels and an impaired immune system. Gut dysbiosis, Irritable Bowel Syndrome, depression, anxiety is found in association with this genotype. The FUT2 rs602662 'AA' genotype and the FUT2 rs601338 'AA' genotype are both associated with non-secretor status. Refer to rs601338 'AA' genotype under the FUT2 status section of this report. Methylmalonic acid (MMA) pathology testing is the gold standard for assessing B12 levels.
FUT2 rs601338	G428A	AA −/−		The 'AA' genotype is associated with typical vitamin B12 levels and reduced risk of vitamin B12 deficiency. However, this must be assessed since this FUT2 genotype is an FUT2 nonsecretor status since this SNP is in linkage with rs602662. Low levels of 2'-FL can lead to decreased B12 levels resulting in anaemia, elevated homocysteine levels and an impaired immune system. Gut dysbiosis, Irritable Bowel Syndrome, depression, anxiety is found in association with this genotype. The FUT2 rs602662 'AA' genotype and the FUT2 rs601338 'AA' genotype are both associated with non-secretor status. Refer to rs601338 'AA' genotype under the FUT2 status section of this report.

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Vitamin C Metabolism

Low blood plasma levels of vitamin C are associated with an increased risk of cardiovascular disease, cancer, and type 2 diabetes. SLC23A1 gene is statistically significantly associated with circulating blood vitamin C concentration. The yellow and red dots indicate increased risk of having low plasma vitamin C levels when individuals do meet the recommended daily intake. Dietary sources of vitamin C are strawberries, pineapple, oranges, broccoli and red peppers.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin C: Solute Carrier family 23 member 1				
SLC23A1 rs33972313	G790A	GG −/−		The 'GG' genotype is associated with typical vitamin C transport and a reduced risk of low vitamin C levels when compared to the 'GA' and 'AA' genotypes.
Vitamin C: Glutathione S-Transferase Theta-1				
GSTT1 CNV	Insertion	Detected PRESENT		The GSTT1 gene has been detected which indicates that the gene is present. This copy number variation is not associated with an increased risk of vitamin C insufficiency. Recent studies have shown that the glutathione transferase genes are involved in the efficient processes of vitamin C.
Vitamin C: Glutathione S-Transferase Mu 1				
GSTM1 CNV	Deletion	Not Detected NULL		The 'AA' genotype is associated with non-functioning (null) genotype. Depending on ethnicity the deletion is relatively common with 50 - 78% of individuals harbouring a NULL genotype. A NULL copy number variation is associated with a reduced ability to process vitamin C from dietary intake and lower blood levels of vitamin C. Recent studies have shown that the glutathione transferase genes are involved in the efficient processes of vitamin C.

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Vitamin E Metabolism

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin E: INTERGENIC nearest gene APOA5				
INTERGENIC rs12272004	Single Nucleotide Variant	AC −/+		The 'AC' genotype is associated with lower levels of alpha-tocopherol in the blood. This result does not mean that the individual's levels are out of balance. Review their dietary intake of vitamin E. Maintain a healthy diet and incorporate foods containing naturally occurring sources of vitamin E such as eggs, nuts, and leafy vegetables. The yellow dot indicates that there is an increased risk of having lower levels of Vitamin E.

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Vitamin D Metabolism

Vitamin D insufficiency has been linked to an increased risk of the following diseases; osteoporosis, fractures, autoimmune diseases such as MS, Crohn's disease, lupus and rheumatoid arthritis, diabetes, depression, mood problems, reduced immunity, and some cancers. The major circulating form of vitamin D is 25(OH)D which can be measured to detect vitamin D insufficiency and to guide dosage. Bone density scans are recommended for females over the age of 40 and males over the age of 50. Vitamin D levels are affected by skin colour, underweight, overweight, obesity, calcium and vitamin D intake, exposure to sunlight, age, pregnancy, and season. Foods rich in vitamin D are liver, egg yolk, oily fish – such as salmon, sardines, herring and mackerel, and fortified foods – such as some fat spreads and breakfast cereals.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin D: 7-dehydrocholesterol reductase, 7-DHC reductase				
DHCR7 rs12785878	Intron Variant	TT −/−		The 'TT' genotype is not associated with vitamin D insufficiency. DHCR7 is a precursor of vitamin D which is found in abundance in the skin. When sunlight penetrates the skin pre-vitamin D is formed. This is the first step in the process. DHCR7 activity is reduced in the presence of benzalkonium chloride (BAC) containing household and personal use products.
Vitamin D: Vitamin D-binding protein				
GC rs2282679	Intron Variant	CC +/+		The 'CC' genotype is associated with reduced vitamin D binding and transport to target tissues such as the skin, kidneys, and liver. The GC gene codes for the vitamin D binding protein (DPB) and it is the major circulating metabolite, transporting vitamin D and its metabolites to target tissues.
Vitamin D: Vitamin D 25-hydroxylase				
CYP2R1 rs10741657	UTR Variant	GG +/+		The 'GG' genotype is associated with slightly reduced vitamin D levels with a reduced response to vitamin D supplementation. The homozygous 'GG' genotype is less responsive than the heterozygotes 'GA' genotype to vitamin D supplementation. This enzymes main roles are in immune support, the vitamin D endocrine system, calcium homeostasis and vitamin D activation.
Vitamin D: Vitamin D3 receptor (FOK1)/Calcium uptake				
VDR-FOK1 rs2228570	FOK1	TC −/+		The Fok1 'TC' genotype is associated with moderately reduced transcriptional activity resulting in reduced activation of vitamin D dependent genes. The 'T' allele has reduced calcium absorption, increased bone turnover and bone loss, a lower bone mineral density in the lumbar spine when compared to the 'C' allele. Reduce caffeine to less than 300mg (2-3 cups of coffee) per day and ensure adequate calcium and vitamin D intake.
Vitamin D: 1,25-dihydroxyvitamin D(3) 24-hydroxylase, mitochondrial, 24-OHase, Vitamin D(3) 24-hydroxylase				
CYP24A1 rs6013897	Single Nucleotide Variant	TA −/+		The 'TA' genotype is associated with an attenuated response to active vitamin D supplementation and slightly reduced vitamin D levels when compared to the 'TT' genotype. The CYP24A1 genetic variation is not a primary factor relating to vitamin D insufficiency. Review the 'TA' genotype in concert with the other genes contributing to vitamin D insufficiency.

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Hormones: Phase I Detoxification and Metabolism Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Phase I Metabolism of Estrogens

The yellow and red dots indicate altered enzyme activity in relation to 2OH, 4OH and 16OH oestrogen metabolites. Review the associated hormone, glutathione, and antioxidant gene MnSOD genes. Review MTHFR, COMT, CYP17A1, CYP19A1 and UGT2B15 since these hormone related genes also metabolise PAHs. Review the glutathione related enzymes GSTT1, GSTM1, GSTP1 since they catalyse the conjugation of glutathione to quinone by-products and reactive oxygen species (ROS) produced as by-products of estrogen metabolism. Review MnSOD since it catalyses the reduction of ROS, which are known to contribute to oxidative stress, lipid peroxidation and DNA damage.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Metabolism of oestrone (E1) and oestradiol (E2)/toxins: Cytochrome P450 1A1				
CYP1A1 rs1048943	A2455G	AA −/−		The 'AA' genotype is associated with typical enzyme activity. CYP1A1 is involved with metabolism of oestrone (E1) and oestradiol (E2) into the 2-hydroxy metabolites, 2OH-E1 and 2OH-E2. CYP1A1 also metabolises environmental pollutants which includes polycyclic aromatic hydrocarbons (PAHs). PAH are present in cigarette smoke, car exhaust fumes and chargrilled meats.
Metabolism of caffeine, hormones including melatonin, estrogens and toxins: Intergenic CYP1A1 and CYP1A2				
CYP1A1/1A2 rs2472297	Intergenic	CT −/+		The 'CT' genotype is associated with slightly increased consumption of caffeine. It is postulated that it may also increase the breakdown and clearance of caffeine. Generally, CYP1A2 catalyses reactions involved in drug metabolism and synthesis of cholesterol, steroids, substances, and other lipids. Inhibitors of the CYP1A2 enzyme include fluoroquinolones (antibiotics), bactericidal, herbs, herbal teas, and certain spices such as turmeric and cumin. Inducers of the enzyme include nicotine and vegetables such as broccoli or cauliflower, environmental toxins such as polycyclic hydrocarbons (PAH's), polychlorinated biphenyls (PCB's), tobacco smoke and chargrilled meat. The polymorphism is not found in those of Asian or African ancestry however it is present in those with European ancestry. This polymorphism exhibits linkage with rs2470893 which has known effects in relation to caffeine breakdown and clearance.
Processing of toxic hydrocarbons and accumulated estrogens: Cytochrome P450 1A1				
CYP1A1_M1 rs4646903	SNV	TC −/+		The 'TC' genotype is associated with increased CYP1A1 enzyme activity. Increased activity favours the formation or 2OH metabolites however it also favours pro-carcinogen activation. Smokers are at increased risk of DNA adduct formation in breast tissue. Review MTHFR, COMT, CYP17A1 and CYP19A1 since these hormone related genes also metabolise PAHs.

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Metabolism of estradiol to 4-OH-estradiol: Cytochrome P450 1B1

The CYP1B1 gene is involved in the phase 1 detoxification of estrogen into 4-OHE1 (E2). Note that the CYP1B1 SNP rs1056836 is referred to in the reverse orientation in this report. This does not affect the information provided. This variant is prone to confusion because the variant is very common and the orientation is often switched in studies. Each of these genetic variants in combination with lifestyle factors such as alcool intake and smoking can have the greatest impact in relation to estrogen related cancers.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Metabolism of estradiol to 4-OH-estradiol: Cytochrome P450 1B1				
CYP1B1 rs1056836	L432V	CC −/−		The 'CC' genotype (or 'GG' genotype in the forward direction Leu432) is associated with slower or decreased estradiol metabolism when compared to the GG genotype (or CC genotype in the reverse direction). Smokers in particular are more prone to hot flushes.
Estrogen metabolites and PAH metabolites: Cytochrome P450 1B1				
CYP1B1 rs1800440	A10106G	AA −/−		The 'AA' genotype is associated with typical enzyme activity, and possibly lower levels of reactive metabolites, when compared to the 'G' allele.

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Phase I Steroidogenesis/ Androgen and Estrogen Metabolism

Gene SNP ID	Gene Variation	Result	Effect	Comment
Circulating estradiol levels: Steroid 17-alpha-hydroxylase/17,20 lyase				
CYP17A1 rs743572	T-34C	GG +/+		The 'GG' genotype is associated with increased CYP17A1 expression and activity. The 'GG' genotype is associated with increased circulating estradiol levels, especially in premenopausal women. Review the hormone related genes COMT, CYP1B1 and CYP19A1.
Conversion of androgens to estrogens: Aromatase				
CYP19A1 rs10046	C19T	CT −/+		The 'CT' genotype is associated with moderate CYP19A1 expression and enzyme activity. This genotype is associated with moderate levels of estrogens and estrogen to androgen ratio's, especially in post-menopausal women. The 'T' allele may increase the risk for hormone-sensitive disorders. Reduce alcohol consumption.
Steoid 5 alpha-reductase				
SRD5A2 rs523349	G264C	CC +/+		The 'CC' genotype is associated with increased enzyme activity and increased conversion of testosterone to dihydrotestosterone (DHT) , with the increased potential risk associated with high DHT levels. If obese then weight loss is recommeded since obesity is shown to influence the activity of SRD5A2 since it results in an increase in catalytic activity and production of biologically active androgens that influence hormonal homeostasis.
UDP glucuronosyltransferase family 2 member B15				
UGT2B15 rs1902023	T253G	CA −/+		The 'A' allele is associated with increased enzyme activity and metabolism of androgenic steroids (androstane-3alpha,17b-diol and dihydrotestosterone (DHT), resulting in a 33% reduction in DHT levels. The 'A' allele is associated with a significant reduction in BPA glucuronidation, an endocrine disrupting plasticizer commonly found in dentistry, food packaging and lacquers. This is shown to potentially increase susceptibility for BPA toxicity.

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Phase I Detoxification/Xenobiotic Metabolism

The CYP2A6 (rs1801272) polymorphism renders the CYP2A6 enzyme unable to metabolise coumarin, interestingly it is also associated with a reduced capacity to metabolise nicotine. In addition, the CYP2A6 enzyme activity affects smoking habits and nicotine dependence. Hence the altered responses may determine strategies are employed to quit smoking. Generally, CYP2A6 is a Phase I detoxification metabolic gene involved in the oxidation and removal of pollutants, herbicides, xenobiotics, and tobacco. Overall, the relevance of Phase I detoxification enzymes in processes connected to the metabolism of toxic substances has been confirmed through scientific investigation.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Drug and herbal medicine metabolism: Cytochrome P450 2C19				
CYP2C19 rs12248560	*17	CC −/−		*1/*1 or the 'CC' genotype is defined as having typical enzyme activity. The cytochrome P450 2C19 (CYP2C19) gene codes for an enzyme is involved in the metabolism of approximately 10% of commonly prescribed drugs. Substrates of CYP2C19 include anti-epileptics, warfarin, proton pump inhibitors and clopidogrel (an antiplatelet drug). CYP2C19 not only converts clopidogrel to its active form, a requirement for the drug to function optimally in the body but it also interacts herbal medicines such as gingko and St. John's Wort in a dose dependent manner.
Drug and herbal medicine metabolism: Cytochrome P450 3A4				
CYP3A4*1B rs2740574	-392A>G	AA −/−		The 'AA' genotype is not associated with a definitive up or down regulation. CYP3A4 works with other CYP genes regulating detox pathways. Despite numerous clinical association studies, the function of CYP3A4*1B remains controversial The CYP3A4*1B is in linkage disequilibrium (LD) with the fully active CYP3A5*1 reference allele in African Americans raising the possibility that CYP3A5 activity could have accounted for any clinical phenotype associated with CYP3A4 *1B. rs2740574 A>G genotype is located 2kb upstream, drug response. CYP3A4 is expressed in the prostate, breast, stomach, colon, small intestine, and liver. CYP3A4 metabolizes many environmental chemicals, therapeutic drugs, and steroid hormones, including testosterone, progesterone, cortisol, and vitamin D. CYP3A4 enzyme activity is significantly modifiable. St. John's Wort, glucocorticoids, smoking, alcohol, and oral contraceptives all induce CYP3A4 expression, whilst grapefruit juice strongly inhibits it. Inhibition is shown to increase circulating levels of estrogen and the bioavailability of many drugs. CYP3A4 is a major catalyst in the hydroxylation of steroid hormones, including estrone, to form the metabolite 16-hydroxyestrone (16-OHE1). Review diet and medication that are metabolised by CYP3A4 to avoid diet and drug interactions. Homozygous TT or AA genotypes have been reported to be associated with decreased gene expression and reduced metabolism of oestrogen and is it also associated with early menarche.
Nicotine metabolism/ Coumarin metabolism: Cytochrome P450 2A6				
CYP2A6 rs1801272	A>T	AA −/−		The 'AA' genotype is associated with faster metabolism of nicotine and coumarin when compared to the 'AT' and 'TT' CYP2A6 genotypes. However, smokers with the 'AA' genotype are likely to not only smoke more cigarettes a day but experience more severe withdrawal symptoms. Nicotine replacement therapy is less effective in those with the 'AA' genotype when compared to individuals with the 'AT' or 'TT' genotypes. Coumarin is a simple plant alkaloid, and it has been used as a sweetener, fixative, stabiliser and previously also as a food additive. It is also present in certain tobacco products, alcoholic beverages, and various kinds of soaps, detergents, and cosmetic preparations. It is found in many plants, where it may serve as a chemical defence against predators. Coumarin-containing preparations have been used as natural medicines in various countries. CYP2A6 is only enzyme able to metabolise coumarin and it is also responsible for the inactivation of nicotine to cotinine.
Cytochrome P450 2D6				
CYP2D6 rs1065852	C>T	CT −/+		The CYP2D6 haplotype (*1/*10) is associated with reduced enzyme activity (CPIC guidelines 2019). CYP2D6 is responsible for the metabolism of many drugs and environmental chemicals that it oxidizes. It is involved in the metabolism of drugs such as antiarrhythmics, adrenoceptor antagonists, and tricyclic antidepressants. The CYP2D6 enzymes metabolize about 25% of pharmaceutical drugs such as SSRIs, opioids, tamoxifen, Nyquil, and beta blockers. This CYP2D6 analysis does not detect duplications, deletions or other variations that may impact on enzyme activity.
CYP2D6 rs3892097	G>A	GG −/−		

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Hormones: Phase II Estrogen and Estrogen Metabolite Elimination Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Catechol-O-methyltransferase				
COMT rs4680	Val158Met	GA −/+		The 'GA' genotype is associated with moderate enzyme activity and methylation of catechol estrogens with possible risk of estrogen-DNA adduct formation and associated tissue damage. Reduced methylation of hydroxylated estrogens may also occur. Reduced methylation of hydroxylated estrogens may result in the accumulation of fat soluble 4-hydroxy-estrones which can be further oxidised to catechol quinones which can be DNA damaging. Assess the individual's weight and discuss weight reduction, if necessary, reduce alcohol consumption if high, review and assess the MTHFR enzyme activity to ensure typical enzyme activity in relation to donating methyl groups to COMT, reduce stress as this may be a factor associated with reduced enzyme activity. Discuss the measurement of urinary estrogen metabolites that comprehensively measure 2, 4 and 16-hydroxylated estrogens.
Glutathione S-Transferase Theta-1				
GSTT1 CNV		Detected PRESENT		The GSTT1 copy number variation is present which is associated with increased enzyme activity and increased clearance of estrogen quinone by-products and ROS, with reduced risk of associated tissue and DNA damage seen with reactive oxygen species accumulation.
Glutathione S-Transferase Mu 1				
GSTM1 CNV		Not Detected NULL		A NULL copy number variation is associated with absent enzyme activity and low clearance of estrogen quinone by-products and ROS, with increased risk of associated tissue and DNA damage seen with ROS accumulation. Depending on ethnicity the deletion is relatively common with 50 - 78% of individuals harbouring a NULL genotype.
Glutathione S-Transferase P				
GSTP1 rs1695	Ile105Val	AG −/+		The 'AG' genotype is associated with decreased conjugation of 18% and decreased clearance of estrogen quinone by-products, with increased risk of associated tissue and DNA damage seen with ROS accumulation. GST enzyme activities are induced in part by sulforaphane found in cruciferous and allium vegetables. These should be increased significantly in the diet to increase activity of other GST enzymes.
Primary anti-oxidant defence system: Superoxide Dismutase [Mn], Mitochondrial				
MnSOD rs4880	Val16Ala	CC −/−		The 'CC' genotype is associated with optimal enzyme activity and a low risk of oxidative damage associated with reactive oxygen species (ROS) production. The 'CC' genotype is however sensitive to inadequate antioxidant intake. There is evidence that Individuals with the 'C' allele consuming a lower intake of fruits and vegetables are at increased risk of developing diseases, including cancers. Review dietary intake of vegetables and fruit, and ensure adequate antioxidant intake from food. Regular, low to moderate intensity exercise increases expression of this gene. Ensure adequate intake of manganese which is a cofactor for SOD2.

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Hormones: LHC, Progesterone and Thyroid Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

This hormone profile is associated with metabolism of luteinising hormone (LH), the steroid hormone progesterone, and thyroid hormones. Polymorphisms in the luteinising hormone/choriogonadotropin receptor (LHCGR) gene are associated with increased insulin responses and an increased risk of polycystic ovary syndrome (PCOS). PCOS is associated with difficulty conceiving, hormonal fluctuations, and irregular menstrual cycles. The LHCGR is the receptor for luteinizing hormone (LH), which triggers ovulation but also human chorionic gonadotropin (hCG), which maintains pregnancy. Increased Progesterone Receptor (PGR) function is associated with increased progesterone receptor function. The FOXE polymorphism is associated with primary hypothyroidism, thyroid disorders and decreased thyroid stimulating hormone. The deiodinase 1 (DIO1) gene produces a protein that converts Thyroxine (T4) to Triiodothyronine (T3) which is involved in the degradation of both T3 and T4 in the liver, kidney, thyroid, and pituitary gland. Iodine and selenium are important for this conversion step. Deiodinase 2 (DIO2) is also involved in converting T4 to T3, which occurs predominantly in skeletal muscles, pituitary, thyroid, brown fat, the heart, and the central nervous system. Selenium and zinc support optimal thyroid function. Enhance your sleep hygiene and support melatonin production by blocking blue light. Optimal iodine levels found in seafood, iodised salt and kelp are important for thyroid health. Environmental factors such as Bisphenols and phthalates can affect thyroid hormones; triclosan found in fluoride toothpaste, mouth washes, facial cleaners, aftershave, deodorants, cosmetics, detergents, and dish washing liquids alter T3 and T4 levels. Animal studies have reported that artificial sweeteners composed of sucralose increase rT3. Avoid perfluoroalkyl a chemical found in cleaners, insecticides, food packaging since it has been shown to affect TSH levels.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Luteinising Hormone Choriogonadotropin Receptor				
LHCGR rs13405728	A226096G	AA −/−		The 'AA' genotype is associated with typical enzyme activity.
Progesterone Receptor				
PGR rs10895068	Intronic	CC −/−		The 'CC' genotype is associated with typical progesterone receptor function.
Thyroid Transcription Factor 2				
FOXE1 rs965513	Intronic	GA −/+		The 'G' allele is associated with increased risk of primary hypothyroidism whilst the 'A' allele is associated with thyroid disorders and decreased thyroid stimulating hormone (TSH). Genetic variants in the TSH-related genes are responsible for approximately 50 – 90% of thyroid hormone variability. The thyroid is sensitive to radiation exposure during childhood whether during treatment of childhood cancers or from environmental exposure, family history and occupational exposures. Review vitamin D, iodine, iron, folate, B12, vitamin C, zinc and selenium intake. Consider measuring vitamin D and thyroid function TSH, T3 and T4.
Deiodinase, Iodothyronine, Type 1				
DIO1 rs2235544	Intronic	CA −/+		The CA' genotype is associated with intermediate enzyme activity. This is the most common genotype frequency across ethnicities.
DI01 rs11206244	UTR_variant	CT −/+		The 'CT' genotype is associated with lower Triiodothyronine (T3). Lower T3 levels are reported to be associated with hypothyroidism, medications, recent thyroid surgery, radiation therapy, pregnancy, or iodine deficiency.
Deiodinase, Iodothyronine, Type 2				
DIO2 rs225014	SNV	TC −/+		The 'TC' genotype is associated with decreased enzyme activity and the reduced conversion of Thyroxine (T4) to Triiodothyronine (T3). Heavy metals, various endocrine disruptors such as bisphenol-A, pesticides and flame retardants have been shown to impair T4 to T3 conversion. Fatigue, depression, constipation, weight gain, dry skin, and hair loss are associated with the reduced conversion of T4 to T3. Dietary intake that are high in sugars and processed carbohydrates can prevent sufficient conversion of T4 to T3. The trace element zinc is required for the conversion of T4 to T3 and good sources include meat, especially red meat, fish, seafood, some nuts and seeds and lentils. Exercise can also improve T4 and T3 levels; beyond helping to stimulate thyroid production, exercise also helps to counteract many of the side effects of hypothyroidism such as gaining weight, loss of muscle, depression, and low energy levels.

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Hormones: Blood Pressure Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Genetic polymorphisms that affect blood pressure and blood clotting may help to guide contraceptive choices, hormonal replacement therapy and pregnancy management.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Angiotensinogen				
AGT rs699	T803C	CT −/+		The 'C' allele is associated with altered plasma AGT levels, with a 10-30% increase among 'C' allele carrier. Individuals with a 'C' allele may be more sensitive to the effects of dietary sodium on vascular function. Limit sodium intake to 2,300mg er day and maintain an adequate intake of potassium rich foods including fruits and vegetables. Review the intake of processed foods, snacks, canned foods, cheeses, and meats since they have a high salt content.
Angiotensin I Converting Enzyme				
ACE rs4343	G2328A	GA −/+		The 'GA' genotype is associated with an increased risk of sodium sensitivity associated with increased sodium intake. The 'G' allele of G2328A is associated with increased levels of ACE which leads to increased processing of angiotensin I to its active form angiotensin II. During exercise this means that blood pressure will increase more rapidly in those carrying the 'G' allele. The increased risk of high blood pressure is associated with an increased cardiovascular risk, and weight gain. Foods that are low in sodium and high in potassium are recommended -since potassium lessens the effect of sodium. The DASH diet is popular among people with high blood pressure. This diet emphasizes fruits and vegetables, both of which are low in sodium and high in potassium. It also includes nuts, whole grains, poultry, and fish. Dairy products also are a good addition to the diet for those individuals that are not lactose intolerant. Milk, yogurt, cheese, and other dairy products are major sources of calcium, vitamin D, and protein. Other low sodium foods include basil, apples, cinnamon, brown rice, kidney beans, and pecans.
Nitric Oxide Synthase 3				
NOS3 rs1799983	G894T	TT +/+		The risk 'T' allele of rs1799983 in the NOS3 gene is associated with a reduction in NOS3 enzyme activity, increased cardiovascular risk markers such as total cholesterol and LDL, and increased risk of high blood pressure. Pregnant females are at increased risk of pre-eclampsia hypertension. Nitric oxide is an antioxidant involved in a broad range of broad range of biological processes including vasodilation of blood vessels. A low-fat dietary intervention has shown that 1.2-1.6 g or 1240mg of EPA and DHA omega-3 fatty acid/day as benefits for those with this variation. Dietary intake of omega 3 rich foods includes salmon, mackerel, and trout. Plant based sources of omega 3 are chia seeds, walnuts, and flaxseed. BH4 or THB is a co-factor necessary for NOS3 function. The 'T' allele of rs1799983 is associated with elevated blood pressure, therefore a reduction in sodium intake to recommended levels should be considered.
Endothelin 1				
EDN1 rs5370	SNV	GG −/−		The 'GG' genotype is associated with typical blood pressure.

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Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Hormones: Coagulation Factors Section

The F5 gene provides instructions for making a protein called coagulation factor V. Coagulation factors are a group of related proteins that make up the coagulation system, a series of chemical reactions that form blood clots. Coagulation factor II (F2) is proteolytically cleaved to form thrombin in the first step of the coagulation cascade which ultimately results in the stemming of blood loss. F2 also plays a role in maintaining vascular integrity during development and postnatal life. Finally, peptides derived from the C-terminus of this protein have antimicrobial activity against E. coli and P. aeruginosa. Mutations in F2 leads to various forms of thrombosis and dysprothrombinemia. The F2 and F5 coagulation-related gene results should be considered together.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Coagulation factor V				
F5 rs6025	R506Q	GG −/−		The 'GG' genotype is associated with typical F5 protein levels, and a reduced risk for blood clotting when compared to the 'GA' and 'AA' genotypes.
Coagulation factor II, thrombin				
F2 rs1799963	G20210A	GG −/−		The 'GG' genotype is associated with typical F2 or prothrombin levels.

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Hormones: Fatty Acid and Choline Metabolism Section

Optimal levels and ratios of PUFAs are important for health in particular reducing inflammation, neural development, new born growth and immune function.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Fatty Acid Desaturase 1				
FADS1 rs174546	G53A	CC −/−		The 'CC' genotype is associated with typical levels of arachidonic acid (Omega-6 polyunsaturated fatty acid) and higher levels of polyunsaturated fatty acid precursors.
FADS1 rs174547	T61803311C	TT −/−		The 'TT genotype has no impact on dietary requirements of linoleic acid an n-6 polyunsaturated fatty acid (PUFA).
Phosphatidylethanolamine N-methyltransferase				
PEMT rs7946	523G>A	AA +/+		The 'AA' genotype is associated with approximately 40% reduced enzyme activity which decreases the ability to produce choline thereby increasing the need for dietary choline. Choline deficiency is associated with several health conditions including neurological disorders, impaired foetal development, and non-alcoholic fatty liver disease. Adequate choline intake is important during all life stages including during pregnancy, whilst breastfeeding and post menopause. Dietary sources of choline are eggs, cauliflower, almonds, and peanut butter. Low dietary choline contributes to high homocysteine levels.

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Phase II: Folate and Homocysteine Metabolism Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

The folate one-carbon metabolism pathway (FOCM) is not only involved in the regulation of homocysteine, methionine and B-vitamin levels but also the methylation of proteins, histones, DNA and RNA.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Vitamin B9: Methylenetetrahydrofolate Reductase				
MTHFR rs1801133	C677T	TT +/+		The 'TT' genotype is associated with 70% reduced MTHFR enzyme activity and an increased risk of reduced folate metabolism and elevated homocysteine level. There is an increased risk of low serum folate and elevated homocysteine levels occurring if the dietary intake of folate and other B group vitamins is not optimal. Low vitamin B2 is associated with reduced MTHFR activity with the 677TT genotype. Please note that enzyme activity requires adequate intake of B vitamins and consideration of other factors such as alcohol intake and medications.
MTHFR rs1801131	A1298C	AA −/−		
Vitamin B9 and choline: Methylenetetrahydrofolate Dehydrogenase				
MTHFD1 rs1076991	C105T	CT −/+		The 'CT' genotype is associated with reduced enzyme activity. The MTHFD1 enzyme supports DNA synthesis and repair, homocysteine metabolism and methylation. Reduced MTHFD1 activity, in association with low folate and/or low dietary choline intake may increase the risk for genome damage, neural tube defects and folate associated health disorders.
MTHFD1 rs2236225	G1958A	GG −/−		The 'GG' genotype is associated with typical enzyme activity. The MTHFD1 enzyme supports DNA synthesis and repair, homocysteine metabolism and methylation.
Folate-mediated one-carbon metabolism/Vitamin B6: Serine Hydroxy Methyltransferase 1				
SHMT1 rs1979277	C1420T	CC −/−		The 'CC' genotype is associated with typical enzyme function. The SHMT1 enzymes regulates key reactions in folate-mediated one-carbon metabolism. Reduced dietary intake of vitamin B6 reduces SHMT1 enzyme activity.

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Phase II: Homocysteine-Methionine Metabolism/Co-factors Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Homocysteine converted to methionine/Vitamin B12: 5-Methyltetrahydrofolate-Homocysteine Methyltransferase				
MTR rs1805087	A2756G	AA +/+		The 'AA' genotype is associated with reduced enzyme activity which results in altered methylation when compared to the 'AG' and 'GG' genotypes. Methionine synthase (MS) is a key enzyme in the methionine cycle, which forms part of the wider methylation pathway. MS generates methionine from homocysteine using 5-MTHF as a donor for a methyl group. A reduction of 5-MTHF occurs in those with a less functional MTHFR enzyme, slowing the reaction which leads to an accumulation of homocysteine. The MS enzyme uses methylated vitamin B12 as a cofactor, zinc and folate for optimal MTR activity and methionine production. Connectivity: Review the MTHFR rs1801133 genotype. If the MTHFR genotype is 'TT' then this variant has been reported to exert a greater effect in pregnant females.
Vitamin B12: 5-Methyltetrahydrofolate-Homocysteine Methyltransferase Reductase				
MTRR rs1801394	A66G	AG −/+		The 'AG' genotype is associated with reduced enzyme activity and increased homocysteine levels when compared to the 'AA' genotype.
Vitamin B12 (cobalamin) transport protein: Transcobalamin 2				
TCN2 rs1801198	C766G	CC −/−		The 'CC' genotype is associated with efficient delivery of vitamin B12 into cells. TCN2 actively facilitates vitamin B12 transport into cells, binding vitamin B12 in the blood plasma and transporting B12 to target cells and tissues. The 'CC' genotype may have more efficient binding of B12.
Plasma homocysteine: Solute Carrier Family 19 Member 1				
SLC19A1 rs4819130	Intron Variant	TT −/−		The 'TT' genotype is not associated with an increase in homocysteine based on this SLC19A1 genotype.

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Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Phase II: Homocysteine-Methionine Metabolism Section

GNMT catalyses the conversion of S-adenosyl-methionine (SAM) to S-adenosyl-homocysteine (SAH), using SAM as the methyl donor, which affects global cellular epigenetic status as the sole source of methyl groups for the cell including those used in DNA, histone, protein and RNA modifications. Cell culture, in vitro L-methionine treatment genotyping of DNA changes including the GNMT SNP rs11752813 and post methionine load testing were performed. The study found that functional differences in the GNMT gene impact an individual's ability to metabolise dietary methionine. The results indicated that the 'GG' genotype at rs11752813 creates a closed chromatin state that inhibits GNMT transcription and methionine metabolism. Reduced methionine metabolism may lead to accumulation of homocysteine (and its dimer, homocystine).

Gene SNP ID	Gene Variation	Result	Effect	Comment
Methionine Metabolism: Glycine N-Methyltransferase				
GNMT rs11752813	SNV	GC −/+		The 'CT' genotype is heterozygous and as such it has the ability to metabolise methionine. The GNMT gene provides instructions for producing the enzyme glycine N-methyltransferase. This enzyme is involved in a multistep process that breaks down the protein building block (amino acid) methionine. The 'CT' genotype may contribute to reduced methionine metabolism. The reduced metabolism of methione may may lead to accumulation of homocysteine (and its dimer, homocystine). In-vitro analysis has reported that the 'G' allele is associated with lower levels of hepatic GNMT protein. However, it maynot be the functional SNP since it may be in linkage disequilibrium with the functional SNP.

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Phase II: Homocysteine Metabolism/Transulphuration Pathway Section

The Cystathionine-Beta-Synthase gene SNP rs234706 has a frequency of 40% for the 'CC' genotype, 40% for the 'CT' genotype and 20% for the 'TT' genotype. All genotypes are relatively common with specific associated effects relating to both homocysteine metabolism and sulphur metabolism.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Plasma homocysteine and sulphur metabolism: Cystathionine-Beta-Synthase				
CBS rs234706	C699T	TT +/+		The 'TT' genotype is associated with active enzyme activity, with low plasma homocysteine and rapid sulphur metabolism. CBS, MTR and BHMT are three enzymes involved in homocysteine metabolism. The "CT" and "TT" CBS genotypes are involved in the up regulation of the CBS gene which results in rapid sulphur metabolism that may correspond with low homocysteine. Both high and low homocysteine are to be avoided. When the CBS gene is highly active, the body cycles through homocysteine rapidly. Since ammonia as a by-product of metabolising sulphur, it has been theorised that increased CBS activity results in higher levels of ammonia, which can cause several health issues. Review the dietary intake of B vitamins and glutathione.

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Phase II: Anti-oxidant Enzymes Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Primary anti-oxidant defence system: MnSOD is located in the cells mitochondria. MnSOD is part of a cascade breaking down the free radical superoxide into hydrogen peroxide, the catalase enzyme (CAT) then neutralises peroxidase to water and oxygen by catalase (CAT) and/or glutathione peroxidase (GPX). MnSOD removes excess reactive oxygen species maintaining the balance between anti-oxidant activity and oxidative stress.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Primary anti-oxidant defence system: Superoxide dismutase [Mn], mitochondrial				
MnSOD rs4880	Val16Ala	CC −/−		The 'CC' genotype is sensitive to inadequate antioxidant intake. This genotype has a complex interaction. Enzyme activity for this genotype has been reported as typical through to being reduced in other studies. The effect of this genotype is disease specific. There is evidence that Individuals with the 'C' allele consuming a lower intake of fruits and vegetables are at increased risk of developing diseases, including some cancers. Review dietary intake of vegetables and fruit and ensure adequate antioxidant intake from food. Regular, low to moderate intensity exercise increases expression of this gene. Ensure adequate intake of manganese which is a cofactor for SOD2.
Primary anti-oxidant defence system: Catalase				
CAT rs1001179	-262G>A	GG −/−		The 'GG' genotype is associated with typical enzyme activity. The protection offered by the 'G' allele is further pronounced in individuals who have a high dietary intake of antioxidant and polyphenol rich foods. Typical enzyme function requires adequate dietary intake and lifestyle changes irrespective of genetics.
Primary anti-oxidant defence system: Glutathione Peroxidase 1				
GPX1 rs1050450	Pro200Leu	CC −/−		The 'CC' genotype is associated with typical enzyme activity. Assess selenium intake to ensure optimal enzyme activity. Brazil nuts, sunflower seeds, fish, shellfish, meat, eggs, mushrooms, grains and onions are good dietary sources of selenium.
Cardiovascular:Paraoxonase 1				
PON1 rs662	Q192R	TT −/−		The 'TT' genotype is associated with efficient enzyme activity when compared to the "TC" and "CC" genotypes. Paraoxonase 1 (PON1) is an antioxidant having an important role in preserving the integrity of cell membranes. The "TT" genotype is associated with preventing LDL oxidation and a lower cardiovascular risk when compared to those carrying a 'C' allele.

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Phase II: Anti-oxidant Enzymes Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Plasma Vitamin C: Solute Carrier Family 23 Member 1				
SLC23A1 rs33972313	G790A	GG −/−		The 'GG' genotype is associated with typical vitamin C transport and reduced risk of low vitamin C levels compared to the 'GA' and 'AA' genotypes. Vitamin C is an essential nutrient which must be obtained from dietary sources. Low blood plasma levels are associated with an increased risk of cardiovascular disease, cancer, and type 2 diabetes.
Heme Oxygenase 1				
HMOX-1 rs2071746	SNV	AA −/−		The 'AA' genotype is associated with typical enzyme activity. This anti-inflammatory antioxidant is located in the brain, intestine, lungs and endothelial cells.
NAD(P)H Quinone Dehydrogenase 1				
NQO1 rs1800566	C609T	CT −/+		In vitro analysis has demonstrated that the rs1800566 'CT' genotype is associated with unstable enzyme activity and reduced antioxidant capacity. Reduced NQO1 enzymatic activity prevents the one electron reduction of quinones which results in the production of radical species. Synthetic antioxidants and extracts of cruciferous vegetables are potent inducers of NQO1. When ubiquinone, the oxidised from of CoQ10, is ingested, the body transforms it to ubiquinol. The NQO1 enzyme encodes for the enzyme that catalyses this conversion. The bioavailability of CoQ10 may be affected in those with the 'T' allele since the conversion of CoQ10 to ubiquinol may be compromised. COQ10 is important for cardiovascular health, cognitive function, nerve health, mitochondrial function, and cellular energy. Reduce exposure to benzene.

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Phase II Detoxification: Glutathione Enzymes Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Glutathione (GSH), referred to as gamma-glutamyl-cysteinylglycine, is a tripeptide formed from glutamic acid, cysteine, and glycine. Glutathione is not only the first defence line against oxidative stress, but it is also involved in a number of metabolic processes such as regulation of enzyme activity, gene expression, signal transduction, and improved cytoplasmic and transmembrane transport. The main role of glutathione is detoxification of xenobiotics and some endogenous compounds. Glutathione is present in cells and body fluids such as blood. Inside the cells, glutathione is present in cytosol and organelles, endoplasmic reticulum, nucleus, and mitochondria.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Glutamate-Cysteine Ligase Catalytic Subunit				
GCLC rs17883901	C53410037T	GG −/−		The 'GG' genotype is associated with typical enzyme activity. Glutamate-cysteine ligase regulatory subunit (GCLC) together with Glutamate-cysteine ligase catalytic subunit (GCLM) forms an enzyme which is the first step in the cellular glutathione (GSH) biosynthetic pathway and is encoded for by the GCLC gene.
Glutamate-Cysteine Ligase Modifier Subunit				
GCLM rs41303970	C590T	GG −/−		The 'GG' genotype is associated with typical enzyme activity. Glutamate-cysteine ligase catalytic subunit (GCLM) together with glutamate-cysteine ligase regulatory subunit (GCLC) forms an enzyme which is the first step in the cellular glutathione (GSH) biosynthetic pathway and is encoded for by the GCLM gene.
Glutathione S-Transferase Pi 1				
GSTP1 rs1695	Ile105Val	AG −/+		The 'AG' genotype is associated with 18% decreased conjugation. GSTP1 is the most abundant glutathione-s-transferase subtype in the lungs and is known to metabolise many carcinogenic compounds. GST enzyme activities are induced in part by sulforaphane found in cruciferous and allium vegetables. These should be increased significantly in the diet to increase activity of other GST enzymes.
GSTP1 rs1138272	C341T	CC −/−		The 'CC' genotype is associated with typical enzyme activity. GSTP1 metabolises many carcinogenic compounds. Reduce exposure to water soluble environmental toxins, including many solvents, herbicides, fungicides, lipid peroxidases and heavy metals such as mercury, cadmium, and lead.
Glutathione S-Transferase Theta 1				
GSTT1 CNV		Detected PRESENT		The GSTT1 copy number variation is present therefore glutathione conjugation capacity IS NOT compromised. PRESENT means that at least one copy of the enzyme has been detected. Glutathione S-transferase T1 is a member of a super family of proteins that catalyse the conjugation of reduced glutathione to a variety of electrophilic and hydrophobic compounds.
Glutathione S-Transferase Mu 1				
GSTM1 CNV		Not Detected NULL		The GSTM1 copy number variation is not detected or null. Depending on ethnicity the deletion is relatively common with 50 - 78% of individuals harbouring a NULL genotype. NULL copy number variations or a deletion result in an absence of the enzyme, leading to reduced capacity for hepatic detoxification and increases the likelihood that mutagenic DNA adducts will be produced. The GSTM1 null genotype is associated with increased risk of various cancers, chemical sensitivity, coronary artery disease in smokers, atopic asthma, and deficits in lung function. Reduce exposure to water soluble environmental toxins, including many solvents, herbicides, fungicides, lipid peroxidases and heavy metals such as mercury, cadmium, and lead. If the exposure to environmental toxins is increased, then discuss risk reduction strategies.

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Phase II Detoxification: N-acetyltransferase 2 (NAT2) Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Acetylation is the major degradation pathway for aromatic amines found in caffeine, meat and fish cooked at high temperatures, tobacco smoke and exhaust fumes.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Deactivation of arylamine and hydrazine drugs and carcinogens. N-Acetyltransferase 2				
NAT2 rs1495741	Tag SNP G>A	GA −/+		The 'GA' genotype is associated with a NAT2 intermediate metaboliser status. NAT2 functions to both activate and deactivate arylamine and hydrazine drugs and carcinogens. Support acetylation with vitamin B5, vitamin C. Short chain fatty acids produced by probiotic bacteria in the gut microbiome also support acetylation.

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Phase II: Amine Metabolism Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

The Monoamine oxidase A (MAO-A) gene located on the X chromosome encodes for the enzyme, Monoamine oxidase. The MAO-A enzyme found in brain metabolises the neurotransmitter's noradrenaline, adrenaline, serotonin, dopamine, and xenobiotic amines. The 'AA' genotype is associated with high MAO-A activity or expression (4-5R) which is associated with reduced levels of these neurotransmitters in the brain. Conversely the 'GG' genotype is associated with low MAO-A activity or expression (2-3R) which is associated with increased levels of the neurotransmitter's adrenalin, dopamine, and serotonin in the brain. High and low levels of MAO-A are both associated with negative effects. The 'R' or repeat indicates the expression or transcription. Simply, an increase in the number of repeats is associated with increased transcription and subsequent increased expression of MAO-A protein. Since MAO-A is located on the X chromosome (X-Linked) females are either heterozygous or homozygous for MAOA since females have two X chromosomes, whereas males only carry one copy (one X and one Y sex chromosome) and are therefore hemizygous. Our algorithm reports males as homozygous. This MAO-A result and its expression depend upon the interaction with other genes, environment, childhood development and psychology.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Serotonin/Dopamine/Norepinephrine metabolism: Monoamine Oxidase A				
MAO-A rs909525	Intron Variant	GA −/+		The 'GA' genotype is associated with intermediate MAOA activity. The 'A' allele is considered the "non-warrior" allele and it is generally not associated with aggression It is suggested that this is the most favourable genotype for healthy brain function.

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Dopamine metabolism

COMT 'GA' and 'AA' genotypes should ensure adequate availability of methylating agents, which may include Vitamin B12, folate and S-adenosyl methionine. Avoid alcohol and a high caffeine intake. Focus on upregulating activity of other phase two detoxification enzymes, by increasing intake of foods such as cruciferous and allium vegetables.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Dopamine metabolism: Catechol-O-methyltransferase				
COMT rs4680	Val158Met	GA −/+		The 'GA' genotype is associated with slow enzyme activity an increased synaptic dopamine when compared to the 'G' genotype, advantage in memory and attention tasks. COMT inactivates catecholamines, catechol estrogens and catechol drugs such as L-DOPA. Reduced methylation of hydroxylated estrogens may also occur. Reduced methylation of hydroxylated estrogens may result in the accumulation of fat soluble 4 hydroxy estrone which can be further oxidised to catechol quinones which can be DNA damaging.

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Histamine Metabolism

Histamine can exert many responses within the body. It can directly stimulate the heart, causing contraction or relaxation of extravascular smooth muscle, stimulates both motor and sensorial neurons, and control gastric secretion. It also mediates primary and immediate symptoms in allergic responses. Potentially elevated levels of histamines in the digestive tract due to variants in APB1 gene that encodes for the DAO enzyme. You may be more sensitive to gut dysbiosis and fluctuating estrogen levels (females). Histamine is lowered by Vitamin C, choline, folate, magnesium, chamomile, basil, stinging nettle, echinacea, fennel, ginger and wild oregano.

Gene SNP ID	Gene Variation	Result	Effect	Comment
D-Amino Acid Oxidase				
AOC1/DAO rs2052129	G4586T	CA −/+		The 'CA' genotype is associated with reduced enzyme function when compared to the 'CC' genotype. Diamine oxidase encoded by the DAO gene also known as histaminase is involved in the metabolism, oxidation, and inactivation of histamine.
AOC1/DAO rs1049793	His664Asp	CC −/−		The 'CC' genotype is associated with typical enzyme function. Diamine oxidase encoded by the DAO gene also known as histaminase is involved in the metabolism, oxidation, and inactivation of histamine.
AOC1/DAO rs1049742	Ser332Phe	CC −/−		The 'CC' genotype is associated with typical enzyme function. Diamine oxidase encoded by the DAO gene also known as histaminase is involved in the metabolism, oxidation, and inactivation of histamine.
AOC1/DAO rs10156191	Thr16Met	CT −/+		The 'CT' genotype is associated with reduced levels of the Diamine Oxidase (DAO) protein, increased sensitivity to aspirin and ibuprofen and increased risk of migraines, especially women.

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Histamine Degradation

HNMT produces the enzyme that uses a methyl group to degrade histamine in the body. Potentially elevated levels of histamine in the central nervous system, skin and bronchial tissue are due to a heterozygous or homozygous HMNT gene polymorphisms. You may be more sensitive to gut dysbiosis and fluctuating estrogen levels (females). Histamine is lowered by Vitamin C, choline, folate, magnesium, chamomile, basil, stinging nettle, echinacea, fennel, ginger and wild oregano.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Histamine N-Methyltransferase				
HNMT rs11558538	Thr105Ile	CC −/−		The 'CC' genotype is associated with typical enzyme activity.
HNMT rs1050891	T939C	AA +/+		The 'AA' genotype is associated with probable increased histamine levels. Reported to contribute to ADHD behaviour when individuals have been exposed to certain food additives: sunset yellow, carnosine, tartrazine, ponceau 4R, quinoline yellow, Allura red AC and sodium benzoate.

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Neurotransmitters, Mood, Cognition and Circadian Rhythm Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Polymorphisms in genes that affect neurotransmitter function via changes to enzyme function have been reported to assist with cognitive support strategies. The MAO-A genotypes are an example of genetics, environment, early life trauma and other genetic contributions. This is a complex association which requires further research to fully elucidate all of the genetic contributions to low mood or aggression.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Serotonin metabolism: Monoamine Oxidase A				
MAO-A rs909525	Intron Variant	GA −/+		The 'GA' genotype is associated with lower levels of neurotransmitters, but it is often assigned as having a neutral status. Monoamine Oxidase A (MAO-A) is an enzyme which is encoded by the MAOA gene which is located on the X chromosome. Therefore, women can be heterozygous or homozygous for MAOA (as women have two X chromosomes), whereas men only carry one copy (as they have one X and one Y sex chromosome) and are therefore hemizygous. Increased MAO-A activity is associated with reduced levels of neurotransmitters in the brain, such as serotonin, noradrenaline, adrenaline, and dopamine. Monoamine oxidase therefore plays an important role in regulating mood. The 'A' allele is considered the "non-warrior" allele and individuals may have anxiety and/or low mood. Males are more susceptible to this effect however environmental contributions should be considered.
MAO-A rs6323	R297R	GT −/+		The 'GT' genotype is associated with increased monoamine oxidase A activity, and it is associated with increased outward anger in females and aggression in males. Monoamine Oxidase A (MAO-A) is an enzyme which is encoded by the MAOA gene which is located on the X chromosome. Therefore, women can be heterozygous or homozygous for MAOA (as women have two X chromosomes), whereas men only carry one copy (as they have one X and one Y sex chromosome) and are therefore hemizygous. Individuals who produce more MAO-A protein being carriers of the 'G' allele or 'GG' genotypes are not as affected by the placebo effect. If there is a conflict in enzyme activity between the two SNPs; rs909525 and rs6323 then the number of repeats inherited are likely to be less than 5 repeats.
Dopamine metabolism: Catechol-O-methyltransferase				
COMT rs4680	Val158Met	GA −/+		The 'GA' genotype is associated with slow enzyme activity and increased synaptic dopamine when compared to the 'GG' genotype, advantage in memory and attention tasks. COMT inactivates catecholamines, catechol estrogens and catechol drugs such as L-DOPA. Reduced methylation of hydroxylated estrogens may also occur. Reduced methylation of hydroxylated estrogens may result in the accumulation of fat soluble 4-hydroxy estrone which can be further oxidised to catechol quinones which can be DNA damaging.
DRD2 receptor activity: Dopamine receptor D2/Ankyrin repeat and Kinase Domain Containing 1				
DRD2/ANNK1 rs1800497	Taq1a	CC −/−		The 'CC' genotype is associated with typical DRD2 receptor activity.

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Neurotransmitters, Mood, Cognition and Circadian Rhythm Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Tyrosine Hydroxylase				
TH rs10770141	C824T	CT −/+		The 'CT' genotype is associated with stress induced high blood pressure, hypertension, and increased expression of catecholamines which includes dopamine, noradrenalin and adrenalin. The 'T' allele is associated with higher catecholamine excretion and greater changes in blood pressure to cold stress, such as cold weather and cold water. Since this polymorphism is also associated with “white coat” hypertension it is suggested that the individuals delays the taking of blood pressure for 5 minutes to assist with obtaining a more accurate blood pressure reading. It has also been reported that low serum cortisol levels and elevated catecholamine typify anxiety caused physical and emotional stress.
Brain Derived Neurotrophic Factor				
BDNF rs6265	Val66Met	AG −/+		The 'AG' genotype is associated with an increased risk of depression and reduced BDNF levels when compared to the 'GG' genotype. Chronic stress can severely impair memory; try relaxation techniques such as meditation and mindfulness. Exercise may improve mood and general feelings via improved expression of BDNF.
Glutamate Decarboxylase 1				
GAD1 rs3749034	147G>A	GA −/+		The 'A' allele is associated with lower GAD1 transcription and possible reduced enzyme activity, resulting in lower levels of GABA when compared to the 'GG' genotype. The 'A' allele is associated with poorer working memory and may increase the risk for mood disorders and is also associated with an increased risk of panic disorder in females when compared the 'G' allele. Reduce glutamate rich foods, reduce sugar intake if high, reduce stress and monitor via functional pathology with organic acids and neurotransmitter profiles.

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Memory Performance

Human memory and recall involve many genetic contributions or polygenic genetic contributions. The KIBRA gene has been widely studied and the evidence shows that genetic variation is associated with hippocampal activation during memory retrieval.

Gene SNP ID	Gene Variation	Result	Effect	Comment
WW domain-containing protein 1S_Brain Health				
KIBRA rs17070145	Intron Variant	CC +/+		The 'CC' genotype is associated with reduced verbal memory, attention, concentration, and delayed recall performance than individuals with the 'T' allele. Review daily exercise, establish a regular sleep pattern, play brain games and meditation as these activities have been reported to improve brain health.

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Stress Reponses - HPA axis

The brain controls the release of cortisol from the adrenal glands. A cascade of events occurs when the hypothalamus signals using corticotrophin releasing hormone (CRH) to the pituitary gland. In response to this signal the pituitary gland releases adrenocorticotropic hormone (ACTH), which then dials up the production of cortisol from the adrenal cortex. This signalling system originating in the Hypothalamus triggers the Pituitary which then sends a signal to the Adrenals is aptly named the HPA axis. There are two binding to two cortisol binding sites the mineralocorticoid (MR) and glucocorticoid (GR) receptors. The MR are triggered by low circulating levels of cortisol controlling typical bodily functions. Conversely, the GR are triggered by higher levels of cortisol under stress situations activating the flight or fight response. Dysfunction of the HPA axis can result in altered patterns of cortisol production across a 24-hour period. This altered pattern is expressed as either inappropriate stress responses or chronically elevated cortisol. Genetic vulnerability, childhood trauma and repeated stress either emotional or physical can result in prolonged elevated cortisol levels. Infertility, weight gain, heart disease, elevated blood glucose, diabetes, insulin resistance, autoimmune diseases, depression, and anxiety.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Mineralocorticoid Receptor				
MR rs5522	Intron Variant	AA −/−		The 'AA' genotype is associated with typical enzyme activity.
MR rs56149945	Intron Variant	TT −/−		The 'TT' genotype is associated with typical enzyme activity.

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Circadian Rhythm

Disruptions to the sleep wake cycle are associated with weight gain, depression, anxiety, social activity and seasonal affective disorder.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Clock Circadian Regulator				
CLOCK rs1801260	Intron Variant	TC −/+		The 'TC' genotype is associated with higher plasma ghrelin concentrations, resistance to weight loss, shorter sleep duration associated with evening preference, and a delayed breakfast time. High plasma ghrelin concentrations correlate with poor food choices when breakfast is delayed until mid to late morning.
CLOCK rs2412646	Intron Variant	GA −/+		The 'GA' genotype is associated with typical levels of social activity. Social activity is important for brain health and healthy ageing.
Neuronal PAS Domain Protein 2				
NPAS2 rs6725296	Intron Variant	GA −/+		The 'GA' genotype is associated with seasonal variation in weight and appetite. Review lifestyle choices such as diet, stress and physical activity levels.
NPAS2 rs2305160	Intron Variant	AG −/+		The 'AG' genotype is associated with typical seasonal variation of sleep length, social activity, mood, weight, appetite, and energy level.

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Weight Management Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Peroxisome Proliferator Activated Receptor Delta				
PPARD rs2016520	Intron Variant	GA −/+		The 'GA' genotype is associated with decreased cardiovascular disease risk, decreased cholesterol, a lower fasting plasma glucose and decreased fasting insulin.
Apolipoprotein A-II				
APOA2 rs5082	Intron Variant	TT −/−		The 'TT' genotype is not associated with obesity with increased fat consumption. Apolipoprotein A2 (APOA2), has a complex and largely undefined role in lipoprotein metabolism, insulin resistance, obesity and atherosclerosis susceptibility.
Triglyceride (TGL) and high fat diet: Apolipoprotein A5				
APOA5 rs662799	Intron Variant	TT +/+		Individuals homozygous for the rs662799 'TT' genotype have been reported to increase BMI as total fat intake increased. Therefore, a diet high in total fat increases the risk of obesity. The 'TT' genotype is responsive to dietary intervention therefore decrease total fat intake to improve weight management outcomes. The 'TT' genotype is associated with a typical risk of having elevated triglyceride levels and no increased cardiovascular risk under the age of 45 years. Refer to the Triglyceride section of the report.
Taste 2 Receptor Member 38				
TAS2R38 rs713598	Ala49Pro	GG −/−		The 'GG' genotype is associated with being a non-taster of bitter flavours and an increased preference for bitter vegetables such as cabbage, soy, broccoli, coffee, and green tea. Conversely you have a reduced preference for foods high in salt and fat.

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Weight Management Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Fat metabolism and Obesity Risk: Fat Mass and Obesity Associated				
FTO rs9939609	Intron Variant	TT −/−		The 'TT' genotype is not associated with an increased BMI or obesity. The Fat mass and obesity-associated protein (FTO) gene is expressed in many regions of the brain including the hypothalamus where it controls your food preferences, eating habits, appetite and feelings of fullness or satiety. The randomised Pounds Lost study recommended a diet consisting of 15% protein, 20-40% fat and 45-65% carbohydrates for weight loss associated with this genotype. Calorie restriction of up to 750 kcal /day and increased fibre intake improved weight loss. However, other genetic contributions relating to weight management or other related health areas should be reviewed. Similarly, any pre-existing health conditions should be taken into consideration and discussed with a health practitioner.
Melanocortin 4 Receptor				
MC4R rs17782313	Intron Variant	TT −/−		The 'TT' genotype is not associated with an increased risk of having a higher BMI due to increased snacking. The Melanocortin-4 Receptor (MC4R) gene is expressed in the hypothalamus an important centre for energy balance and the regulation of food intake.
DRD2 receptor activity: Dopamine receptor D2/Ankyrin repeat and Kinase Domain Containing 1				
DRD2/ANNK1 rs1800497	Taq1a	CC −/−		The 'CC' genotype is associated with typical DRD2 receptor activity.
Solute Carrier Family 2 Member 2: Sugar consumption				
SLC2A2 rs5400	Intron Variant	CC −/−		The 'CC' genotype is associated with typical sugar consumption when compared to the 'CT' and 'TT' genotypes.
Adiponectin				
ADIPOQ rs17366568	Intron Variant	GG −/−		The 'GG' genotype is associated with typical adiponectin levels which are associated with a lower risk of obesity and an increased ability to metabolise fat.

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Weight Management Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Adrenoceptor Beta 2				
ADRB2 rs1042713	Arg16Gly	AG −/+		The 'AG' genotype is associated with slightly reduced weight loss. A Mediterranean diet lower in saturated fat will assist with weight loss.
Adrenoceptor Beta 3				
ADRB3 rs4994	Trp64Arg	TT −/−		The 'TT' genotype is associated with typical regulation of lipolysis and thermogenesis. Weight loss will be relatively easy.
Adiponectin				
ADIPOQ rs17300539	Intron Variant	GG +/+		The 'GG' genotype is more likely to regain weight after eating a low calorie diet.
Fat mass and obesity-associated protein				
FTO rs1558902	Intron Variant	AT −/+		The 'AT' genotype is associated with obesity when sedentary. However, improved weight loss was reported when a diet higher protein is consumed. A low protein diet of less than 18% total energy is associated with a significantly higher BMI.
FTO rs1121980	Intron Variant	CT −/+		The 'T' allele is associated with an increased BMI. However, exercise has been reported to have a positive impact on weight loss in those with a 'T' allele.

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Sports and Exercise Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Insulin Induced Gene 2				
INSIG2 rs7566605	Intron Variant	GG −/−		The 'GG' genotype is not associated with an increase in fat volume associated with strength training.
Collagen Type I Alpha 1 Chain				
COL1A1 rs1800012	Intron Variant	GG −/−		The 'GG' genotype is associated with the typical production of type 1 collagen, typical bone strength and bone mineral density.
Vitamin D: Vitamin D3 receptor (FOK1)/Calcium uptake				
VDR-FOK rs2228570	FOK1	TC −/+		The Fok1 'CT' genotype is associated with moderately reduced transcriptional activity resulting reduced activation of vitamin D dependent genes. The 'T' allele is associated with reduced calcium absorption, increased bone turnover and bone loss, a lower bone mineral density in the lumbar spine compared to the 'C' allele. Reduce caffeine to less than 300mg (2-3 cups of coffee) per day and ensure adequate calcium and vitamin D intake. Incorporate weight-bearing exercises into the training routine. Increase vigilance for BMD screening. Test for vitamin D status.
Vascular Endothelial Growth Factor A				
VEGFR2 rs1870377	Gln472His	AA −/−		
ACE rs4341	Intron Variant	CG −/+		
ACE rs4343	Thr776=	AG −/+		This haplotype is associated with mixed sprint and endurance ability.
ACTN3 rs1815739	Arg620Ter	CC −/−		
HIF1 rs11549465	Pro583Ser	CT −/+		

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Sports and Exercise Section

Support Definitions
- No Action
- Practitioner Review/Action
- Practitioner Action

Gene SNP ID	Gene Variation	Result	Effect	Comment
Hypoxia Inducible Factor 1 Subunit Alpha				
HIF1 rs11549465	Pro583Ser	CT −/+		The 'CT' genotype is associated with a lower change in VO2 max.
Monocarboxylate transporter 1: Lactate removal				
MCT-1 rs1049434	Asp490Glu	AT −/+		The 'AT' genotype is associated with moderate lactate removal when compared to the 'AA' genotype. MCT1 mediates the movement of lactate and pyruvate across the cell membrane. Slower removal of lactate from cells which results in longer recovery time after intense physical exercise.
Nitric Oxide Synthase 3:Vasodilation				
eNOS3 rs2070744	Intron Variant	CC +/+		The 'CC' genotype is associated with reduced endothelial nitric oxide synthesis which is not associated with an endurance and power athletic performance.
Angiotensin I Converting Enzyme: Serum Ace Activity				
ACE rs4341	Intron Variant	CG −/+		The 'CG' genotype is associated with higher serum and ACE activity giving the individual endurance and sprint ability.
ACE rs4343	Thr776=	AG −/+		The 'AG' genotype is associated with higher serum and ACE activity giving the individual endurance and sprint ability.
Interleukin 6: Muscle repair, Inflammation and CRP levels				
IL-6 rs1800795	Intron Variant	GC −/+		The 'C' allele has been linked to increased IL-6 and CRP production in muscle cells in response to exercise, which may induce more pronounced fatigue due to excess inflammation and prolong recovery times. Higher IL-6 levels are also associated with a lower VO2max, which may affect aerobic capacity. The 'C' allele has been linked to increases in the muscle damage marker, creatine kinase, following exercise.
Collagen Type V Alpha 1 Chain: Achilles Tendon Risk				
COL5A1 rs12722	Intron Variant	CT −/+		The 'CT' genotype is associated with moderate risk of ACL injury and a reduced range of motion when compared to the 'CC' genotype. Conditioning and flexibility training are important parts of injury prevention for at risk individuals.

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Concussion Recovery

Recovery from concussion has recently been demonstrated to be influenced by genotype. A single nucleotide polymorphism (SNP) in the SLC17A7 gene has been reported to be associated with delayed recovery time from head injuries. Immediately after a concussion, the brain is in crisis. Many brain cells become activated at the same time, which can affect stress, steroids, appetite, peptides, neurotransmitters, and other hormones, creating an imbalance and a host of unpleasant symptoms. This cascade of events has multiple chemical consequences; astroglial activation and β-amyloid (Aβ) dysmetabolism in the brain, glutamate an excitatory neurotransmitter is disrupted resulting in diminished transport from the synaptic cleft, GABA is an inhibitory neurotransmitter has diminished activity and the delicate balance between glutamate and GABA is skewed to an excitatory state resulting in neurological injury and dysfunction of neurons. Restoration of the neurotransmitter balance is part of a concussion recovery management. Human and animal studies have demonstrated that zinc may be beneficial to those as risk of head injury by improving resilience, fish oil has been reported to reduce neural inflammation in an animal study. Similarly, an animal study reported that vitamin D3 may protect against glutamate toxicity via upregulation of the vitamin D receptor (VDR), antioxidants within the first 24 hours may prove beneficial since oxidative stress is heightened during this window. Vitamin C has been reported to be depleted following a concussion. Exercise following a concussion has also been reported to improve recovery.

Gene SNP ID	Gene Variation	Result	Effect	Comment
Solute Carrier Family 17				
SLC17A7 rs74174284	SNV	CG −/+		The 'CG' and 'GG' genotypes, (one two copies of the 'G' allele) are 6.33 times more likely to experience prolonged recovery exceeding 20 days. Review the APOE genotype since there is a possible relationship between the ε4 allele and post-concussion impairment, as well as between the ε4 allele and neurocognitive performance variability, suggesting that the ε4 genotype may be a risk factor for less efficient cognitive processing in concussed athletes.

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