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DATOS GENÉTICOS

En esta prueba genética se identificaron 59 variantes genéticas, de un total de 102 variantes evaluadas, con impacto en la definición de un plan nutricional y/o en la promoción de la salud metabólica. Las variantes con impacto en cada una de las características se pueden consultar en las respectivas secciones de detalle. En la siguiente tabla se indican las variantes genéticas que se tuvieron en cuenta en la realización de este informe. Los resultados se describen de acuerdo con la nomenclatura HGVS (`http:www.hgvs.org`) consultada a fecha de 1 de julio de 2020.

Gen	Referencia de la variante genética		Cambio nucleotídico [1]	Cambio aminoacídico	Resultado
	HGMD	Ensembl			
ADD1	CM021240	rs4961	c.1378G>T	p.Gly460Trp	G
ADIPOQ	CR052432	rs17300539	c.-1138A>G	–	G
ADORA2A	–	rs5751876	g.24837301T>C	–	C
ADRB2	CM950016	rs1042713	c.46A>G	p.Arg16Gly	G
AHR	–	rs4410790	g.17284577T>C	–	C
ALPL	–	rs4654748	c.134-9113T>C	–	T
AMDHD1	–	rs10745742	g.95964751C>T	–	CT
APOA1	CR900263	rs670	c.-113A>G	–	GA
APOA2	CR024268	rs5082	c.-323T>C	–	T
APOA5	CM023881	rs3135506	c.56G>C	p.Trp19Ser	C
APOA5	CM032546	rs2075291	c.553G>T	p.Gly185Cys	G
APOA5	CR033141	rs662799	c.-620C>T	–	T
APOB	–	rs512535	c.-965A>G	–	G
APOE	CM860003	rs7412	c.526C>T	p.Arg176Cys	C
APOE	CM900020	rs429358	c.388T>C	p.Cys130Arg	T
BCO1	CM091857	rs12934922	c.801A>T	p.Arg267Ser	AT
BCO1	CM091858	rs7501331	c.1136C>T	p.Ala379Val	CT
BDNF	–	rs10767664	c.-22+16205A>T	–	TA
CLCNKA	–	rs848307	n.530+427C>T	–	C
CLOCK	CR121503	rs3749474	c.*897G>A	–	GA
CLOCK	CR984677	rs1801260	c.*213T>C	–	T
COMT	CM960420	rs4680	c.472G>A	p.Val158Met	G
CRY1	–	rs2287161	c.-562G>C	–	CG
CRY2	–	rs11605924	c.32+4259A>C	–	CA
CYP1A1	–	rs2470893	c.-1694G>A	–	AG
CYP1A1	–	rs2472297	g.74735539C>T	–	CT
CYP1A2	CR993820	rs762551	c.-9-154C>A	–	A
CYP24A1	–	rs17216707	g.54115823T>C	–	T
CYP2R1	–	rs10741657	g.14914878A>G	–	GA
DHCR7	–	rs12785878	c.146+1233G>T	–	GT
DRD2	CM041241	rs1800497	c.2137G>A	p.Glu713Lys	A
FABP2	CM950433	rs1799883	c.163G>A	p.Ala55Thr	AG
FADS1	CR1510437	rs174546	c.*53A>G	–	GA
FTO	–	rs1121980	c.46-34805G>A	–	G
FTO	CR119357	rs1558902	c.46-40478T>A	–	T
FTO	CS076623	rs9939609	c.46-23525T>A	–	T
FTO	CS088104	rs8050136	c.46-27777C>A	–	C
FUT2	CM042988	rs602662	c.772A>G	p.Ser258Gly	GA
GC	–	rs2282679	c.*26-796A>C	–	A
GCKR	CR118767	rs780094	c.1423-418T>C	–	CT
GHSR	CR084002	rs490683	g.172175074C>G	–	G
GIPR	–	rs2287019	c.886+14T>C	–	C
GRB14	–	rs10195252	g.165513091C>T	–	C
GRK4	CM025429	rs2960306	c.194G>T	p.Arg65Leu	GT
GRK4	CM025430	rs1024323	c.425C>T	p.Ala142Val	CT
HLADQA1	–	rs2187668	g.32638107C>T	–	C
HLADQB1	–	rs4713586	g.32691805A>G	–	A
HLADQB1	–	rs7454108	g.32713706T>C	–	CT
HLADQB1	–	rs7775228	g.32690302T>C	–	T
HLADRA	–	rs2395182	c.*406+494G>T	–	T
IL6	CR983402	rs1800795	c.-237G>C	–	G
IM11	–	rs12272004	g.116733008C>A	–	CA
IM19	–	rs4420638	g.44919689A>G	–	GA

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	HGMD	Ensembl			
IRS1	CR096329	rs2943641	g.227093745TC>T	–	CT
LDLR	–	rs6511720	c.67+2015G>T	–	G
LEPR	–	rs11208659	c.-20-51949T>C	–	T
LIPC	CR971949	rs1800588	c.-557C>T	–	TC
LPL	CM900164	rs328	c.1421G>C	p.Term474Ser	C
LPL	CS890131	rs285	c.1019-1582C>T	–	CT
LPL	CS931395	rs320	c.1322+483G>T	–	T
LYPLAL1	–	rs2605100	g.219470882A>G	–	G
MC4R	–	rs11152221	g.60350016C>T	–	T
MC4R	–	rs12970134	g.60217517G>A	–	AG
MC4R	–	rs17700633	g.60262199G>A	–	A
MC4R	–	rs17782313	g.60183864T>C	–	TC
MC4R	CM030481	rs2229616	c.307A>G	p.Ile103Val	G
MC4R	CM030483	rs52820871	c.751C>A	p.Leu251Ile	A
MCM6	CR024269	rs4988235	c.1917+326C>T	–	CT
MCM6	CR024379	rs182549	c.1362+117G>A	–	AG
MCM6	CR070424	rs145946881	c.1917+226G>C	–	G
MCM6	CR070425	rs41380347	c.1917+321T>G	–	T
MCM6	CR070426	rs41525747	c.1917+329C>G	–	C
MSRA	–	rs545854	g.9860080C>G	–	C
MTHFR	CM950819	rs1801133	c.665C>T	p.Ala222Val	TC
MTHFR	CM981315	rs1801131	c.1286A>C	p.Glu429Ala	A
MTNR1B	CR110512	rs10830963	c.223+5596C>G	–	C
NR1D1	–	rs12941497	c.31+723C>T	–	TC
NR1D1	–	rs2314339	c.370+106A>G	–	G
OPRM1	CM003770	rs1799971	c.118A>G	p.Asn40Asp	A
PCSK1	CM083013	rs6232	c.661A>G	p.Asn221Asp	A
PCSK1	CM1311914	rs6235	c.2069C>G	p.Thr690Ser	C
PEMT	CR063410	rs12325817	g.17486519C>G	–	CG
PER2	–	rs2304672	c.-12C>G	–	CG
PER2	–	rs4663302	g.238295120C>T	–	C
PLIN	CS045669	rs894160	c.772-799G>A	–	G
PNPLA3	CM086892	rs738409	c.444C>G	p.Ile148Met	CG
PPARD	CR035869	rs2016520	c.-87C>T	–	T
PPARG	CM981614	rs1801282	c.34C>G	p.Pro12Ala	C
PPM1K	–	rs1440581	n.133-6526T>C	–	TC
PROX1	–	rs340874	c.-68+2590T>C	–	CT
SEC16B	–	rs539515	g.177919890A>C	–	A
SEC23A	–	rs8018720	g.39086981G>C	–	CG
SIRT1	–	rs1467568	c.1916-864A>G	–	AG
SLC23A1	CM0911294	rs33972313	c.790G>A	p.Val264Met	G
SLC2A2	CM941277	rs5400	c.329C>T	p.Thr110Ile	CT
SLC30A8	CM072050	rs13266634	c.826C>T	p.Arg276Trp	C
SOD2	CM962694	rs4880	c.47T>C	p.Val16Ala	T
SORT1	–	rs629301	c.*1635G>T	–	GT
TCF7L2	CS065626	rs7903146	c.382-41435C>T	–	CT
TFAP2B	–	rs987237	c.602-724A>G	–	A
TM6SF2	CM143615	rs58542926	c.499G>A	p.Glu167Lys	G
TMEM18	–	rs2867125	g.622827T>C	–	CT

[1] El ID numérico asociado a cada una de las alteraciones, está indexado a una secuencia de referencia obtenida de la base de datos Ensembl (`http://www.ensembl.org/index.html`).

Haplotipos *APOE* identificados:

- *APOE*-$\varepsilon_3/\varepsilon_3$ (T,T) (C,C)

Haplotipos *HLA identificados:

- DQ8/DQX