

MOMA Exome Endocrinology v2 (2016-03-01), 212 genes

Approved symbol	Approved name	HGNC ID	Location
ABCC8	ATP binding cassette subfamily C member 8	HGNC:59	11p15.1
AIP	aryl hydrocarbon receptor interacting protein	HGNC:358	11q13.3
AIRE	autoimmune regulator	HGNC:360	21q22.3
ALB	albumin	HGNC:399	4q13.3
ALPL	alkaline phosphatase, liver/bone/kidney	HGNC:438	1p36.12
AMER1	APC membrane recruitment protein 1	HGNC:26837	Xq11.1
ANKH	ANKH inorganic pyrophosphate transport regulator	HGNC:15492	5p15.2
ANOS1	anosmin 1	HGNC:6211	Xp22.32
AP2S1	adaptor related protein complex 2 sigma 1 subunit	HGNC:565	19q13.32
APOA2	apolipoprotein A-II	HGNC:601	1q23.3
AQP2	aquaporin 2	HGNC:634	12q13.12
AR	androgen receptor	HGNC:644	Xq12
ATP6V0A4	ATPase H+ transporting V0 subunit a4	HGNC:866	7q34
ATP6V1B1	ATPase H+ transporting V1 subunit B1	HGNC:853	2p13
ATP7B	ATPase copper transporting beta	HGNC:870	13q14.3
AVP	arginine vasopressin	HGNC:894	20p13
AVPR2	arginine vasopressin receptor 2	HGNC:897	Xq28
AXL	AXL receptor tyrosine kinase	HGNC:905	19q13.1
BLK	BLK proto-oncogene, Src family tyrosine kinase	HGNC:1057	8p23-p22
BMP1	bone morphogenetic protein 1	HGNC:1067	8p21
BRAF	B-Raf proto-oncogene, serine/threonine kinase	HGNC:1097	7q34
BSND	barttin CLCNK type accessory beta subunit	HGNC:16512	1p32.3
CASR	calcium sensing receptor	HGNC:1514	3q21.1
CBS	cystathionine-beta-synthase	HGNC:1550	21q22.3
CDC73	cell division cycle 73	HGNC:16783	1q25
CDKN1A	cyclin-dependent kinase inhibitor 1A	HGNC:1784	6p21.1
CDKN1B	cyclin-dependent kinase inhibitor 1B	HGNC:1785	12p13.1-p12
CDKN1C	cyclin-dependent kinase inhibitor 1C	HGNC:1786	11p15.5
CEL	carboxyl ester lipase	HGNC:1848	9q34.3
CHD7	chromodomain helicase DNA binding protein 7	HGNC:20626	8q12.2
CISD2	CDGSH iron sulfur domain 2	HGNC:24212	4q24
CLCN5	chloride voltage-gated channel 5	HGNC:2023	Xp11.23
CLCN7	chloride voltage-gated channel 7	HGNC:2025	16p13.3
CLCNKB	chloride voltage-gated channel Kb	HGNC:2027	1p36.13
CLDN16	claudin 16	HGNC:2037	3q28
COL1A1	collagen type I alpha 1	HGNC:2197	17q21.33
COL1A2	collagen type I alpha 2	HGNC:2198	7q21.3
COL4A5	collagen type IV alpha 5	HGNC:2207	Xq22

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CREB3L1	cAMP responsive element binding protein 3-like 1	HGNC:18856	11q11
CRTAP	cartilage associated protein	HGNC:2379	3p22
CTSK	cathepsin K	HGNC:2536	1q21
CYP11B1	cytochrome P450 family 11 subfamily B member 1	HGNC:2591	8q21-q22
CYP11B2	cytochrome P450 family 11 subfamily B member 2	HGNC:2592	8q21-q22
CYP17A1	cytochrome P450 family 17 subfamily A member 1	HGNC:2593	10q24.3
CYP19A1	cytochrome P450 family 19 subfamily A member 1	HGNC:2594	15q21
CYP21A2	cytochrome P450 family 21 subfamily A member 2	HGNC:2600	6p21.3
DGCR10	DiGeorge syndrome critical region gene 10 (non-protein coding)	HGNC:17864	22q11
DGCR11	DiGeorge syndrome critical region gene 11 (non-protein coding)	HGNC:17226	22q11.21
DGCR14	DiGeorge syndrome critical region gene 14	HGNC:16817	22q11.21
DGCR2	DiGeorge syndrome critical region gene 2	HGNC:2845	22q11.21
DGCR5	DiGeorge syndrome critical region gene 5 (non-protein coding)	HGNC:16757	22q11
DGCR6	DiGeorge syndrome critical region gene 6	HGNC:2846	22q11.21
DGCR6L	DiGeorge syndrome critical region gene 6-like	HGNC:18551	22q11.21
DGCR8	DGCR8 microprocessor complex subunit	HGNC:2847	22q11.2
DGCR9	DiGeorge syndrome critical region gene 9 (non-protein coding)	HGNC:17227	22q11.21
DMP1	dentin matrix acidic phosphoprotein 1	HGNC:2932	4q21
DMRT1	doublesex and mab-3 related transcription factor 1	HGNC:2934	9p24.3
DMXL2	Dmx like 2	HGNC:2938	15q21.2
EGLN1	egl-9 family hypoxia-inducible factor 1	HGNC:1232	1q42.1
EIF2AK3	eukaryotic translation initiation factor 2 alpha kinase 3	HGNC:3255	2p12
ENPP1	ectonucleotide pyrophosphatase/phosphodiesterase 1	HGNC:3356	6q22-q23
EPAS1	endothelial PAS domain protein 1	HGNC:3374	2p21-p16
FBN1	fibrillin 1	HGNC:3603	15q21.1
FEZF1	FEZ family zinc finger 1	HGNC:22788	7q31.32
FGF17	fibroblast growth factor 17	HGNC:3673	8p21.3
FGF23	fibroblast growth factor 23	HGNC:3680	12p13
FGF8	fibroblast growth factor 8	HGNC:3686	10q25-q26
FGFR1	fibroblast growth factor receptor 1	HGNC:3688	8p11.23-p11.22
FKBP10	FK506 binding protein 10	HGNC:18169	17q21.2
FOXP3	forkhead box P3	HGNC:6106	Xp11.23
FTO	fat mass and obesity associated	HGNC:24678	16q12.2
GATA3	GATA binding protein 3	HGNC:4172	10p15
GATA4	GATA binding protein 4	HGNC:4173	8p23.1-p22
GATA6	GATA binding protein 6	HGNC:4174	18q11-q12
GCK	glucokinase	HGNC:4195	7p15.3-p15.1
GCM2	glial cells missing homolog 2	HGNC:4198	6p24.2

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GHRHR	growth hormone releasing hormone receptor	HGNC:4266	7p14
GLIS3	GLIS family zinc finger 3	HGNC:28510	9p24.2
GNA11	guanine nucleotide binding protein (G protein), alpha 11 (Gq class)	HGNC:4379	19p13.3
GNAS	GNAS complex locus	HGNC:4392	20q13.2-q13.3
GNRH1	gonadotropin releasing hormone 1	HGNC:4419	8p21-p11.2
HESX1	HESX homeobox 1	HGNC:4877	3p14.3
HNF1A	HNF1 homeobox A	HGNC:11621	12q24.31
HNF1B	HNF1 homeobox B	HGNC:11630	17q12
HNF4A	hepatocyte nuclear factor 4 alpha	HGNC:5024	20q13.12
HS6ST1	heparan sulfate 6-O-sulfotransferase 1	HGNC:5201	2q21
HSD11B1	hydroxysteroid (11-beta) dehydrogenase 1	HGNC:5208	1q32-q41
HSD3B2	hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2	HGNC:5218	1p12
IER3IP1	immediate early response 3 interacting protein 1	HGNC:18550	18q12
IFITM5	interferon induced transmembrane protein 5	HGNC:16644	11p15.5
IGF2	insulin like growth factor 2	HGNC:5466	11p15.5
IGFALS	insulin like growth factor binding protein acid labile subunit	HGNC:5468	16p13.3
IL17RD	interleukin 17 receptor D	HGNC:17616	3p21.1
INS	insulin	HGNC:6081	11p15.5
KCNJ1	potassium voltage-gated channel subfamily J member 1	HGNC:6255	11q24
KCNJ11	potassium voltage-gated channel subfamily J member 11	HGNC:6257	11p15.1
KISS1	KISS-1 metastasis-suppressor	HGNC:6341	1q32
KISS1R	KISS1 receptor	HGNC:4510	19p13.3
KLF11	Kruppel-like factor 11	HGNC:11811	2p25
KRAS	Kirsten rat sarcoma viral oncogene homolog	HGNC:6407	12p12.1
LEP	leptin	HGNC:6553	7q31
LEPR	leptin receptor	HGNC:6554	1p31
LHX3	LIM homeobox 3	HGNC:6595	9q34.3
LHX4	LIM homeobox 4	HGNC:21734	1q25.3
LRP5	LDL receptor related protein 5	HGNC:6697	11q13.4
MAS1	MAS1 proto-oncogene, G protein-coupled receptor	HGNC:6899	6q24-q27
MC4R	melanocortin 4 receptor	HGNC:6932	18q22
MEN1	menin 1	HGNC:7010	11q13
MMP2	matrix metalloproteinase 2	HGNC:7166	16q12.2
MNX1	motor neuron and pancreas homeobox 1	HGNC:4979	7q36
NEUROD1	neuronal differentiation 1	HGNC:7762	2q32
NEUROG3	neurogenin 3	HGNC:13806	10q21.3
NF1	neurofibromin 1	HGNC:7765	17q11.2
NF2	neurofibromin 2 (merlin)	HGNC:7773	22q12.2

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NIPA1	non imprinted in Prader-Willi/Angelman syndrome 1	HGNC:17043	15q11.2
NIPA2	non imprinted in Prader-Willi/Angelman syndrome 2	HGNC:17044	15q11.2
NKX2-2	NK2 homeobox 2	HGNC:7835	20p11.22
NPFFR2	neuropeptide FF receptor 2	HGNC:4525	4q21
NROB1	nuclear receptor subfamily 0 group B member 1	HGNC:7960	Xp21.3
NR5A1	nuclear receptor subfamily 5 group A member 1	HGNC:7983	9q33
NRAS	neuroblastoma RAS viral (v-ras) oncogene homolog	HGNC:7989	1p13.2
NSMF	NMDA receptor synaptonuclear signaling and neuronal migration factor	HGNC:29843	9q34.3
OCRL	oculocerebrorenal syndrome of Lowe	HGNC:8108	Xq25
OTUD4	OTU deubiquitinase 4	HGNC:24949	4q31.21
P3H1	prolyl 3-hydroxylase 1	HGNC:19316	1p34.1
PAX4	paired box 4	HGNC:8618	7q32.1
PAX6	paired box 6	HGNC:8620	11p13
PCSK1	proprotein convertase subtilisin/kexin type 1	HGNC:8743	5q15-q21
PDX1	pancreatic and duodenal homeobox 1	HGNC:6107	13q12.1
PHEX	phosphate regulating endopeptidase homolog, X-linked	HGNC:8918	Xp22.2-p22.1
PHF8	PHD finger protein 8	HGNC:20672	Xp11.22
PIK3CA	phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha	HGNC:8975	3q26.3
PLAGL1	PLAG1 like zinc finger 1	HGNC:9046	6q24-q25
PLOD2	procollagen-lysine, 2-oxoglutarate 5-dioxygenase 2	HGNC:9082	3q24
PLS3	plastin 3	HGNC:9091	Xq23
PNPLA6	patatin like phospholipase domain containing 6	HGNC:16268	19p13.2
POLD1	polymerase (DNA directed), delta 1, catalytic subunit	HGNC:9175	19q13.3
POLR3B	polymerase (RNA) III (DNA directed) polypeptide B	HGNC:30348	12q23.3
POMC	proopiomelanocortin	HGNC:9201	2p23
POR	P450 (cytochrome) oxidoreductase	HGNC:9208	7q11.2
POU1F1	POU class 1 homeobox 1	HGNC:9210	3p11.2
PPARG	peroxisome proliferator activated receptor gamma	HGNC:9236	3p25
PIIB	peptidylprolyl isomerase B	HGNC:9255	15q21-q22
PRKAR1A	protein kinase cAMP-dependent type I regulatory subunit alpha	HGNC:9388	17q24.2
PROK2	prokineticin 2	HGNC:18455	3p21.1
PROKR2	prokineticin receptor 2	HGNC:15836	20p12.3
PROP1	PROP paired-like homeobox 1	HGNC:9455	5q35.3
PRRT2	proline rich transmembrane protein 2	HGNC:30500	16p11.2
PTEN	phosphatase and tensin homolog	HGNC:9588	10q23
PTF1A	pancreas specific transcription factor, 1a	HGNC:23734	10p12.31
PTH	parathyroid hormone	HGNC:9606	11p15.3
PTPN11	protein tyrosine phosphatase, non-receptor type 11	HGNC:9644	12q24.1

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PTTG1	pituitary tumor-transforming 1	HGNC:9690	5q35.1
RAF1	Raf-1 proto-oncogene, serine/threonine kinase	HGNC:9829	3p25
RB1	retinoblastoma 1	HGNC:9884	13q14.2
RET	ret proto-oncogene	HGNC:9967	10q11.2
RFX6	regulatory factor X6	HGNC:21478	6q22.31
RNF216	ring finger protein 216	HGNC:21698	7p22.1
SCNN1B	sodium channel epithelial 1 beta subunit	HGNC:10600	16p12.2-p12.1
SCNN1G	sodium channel epithelial 1 gamma subunit	HGNC:10602	16p12
SDHA	succinate dehydrogenase complex flavoprotein subunit A	HGNC:10680	5p15
SDHAF2	succinate dehydrogenase complex assembly factor 2	HGNC:26034	11q12.2
SDHB	succinate dehydrogenase complex iron sulfur subunit B	HGNC:10681	1p36.1-p35
SDHC	succinate dehydrogenase complex subunit C	HGNC:10682	1q23.3
SDHD	succinate dehydrogenase complex subunit D	HGNC:10683	11q23
SEMA3A	semaphorin 3A	HGNC:10723	7p12.1
SEMA7A	semaphorin 7A (John Milton Hagen blood group)	HGNC:10741	15q22.3-q23
SERPINA7	serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 7	HGNC:11583	Xq21-q22
SERPINF1	serpin peptidase inhibitor, clade F (alpha-2 antiplasmin, pigment epithelium derived factor), member 1	HGNC:8824	17p13.3
SERPINH1	serpin peptidase inhibitor, clade H (heat shock protein 47), member 1, (collagen binding protein 1)	HGNC:1546	11q13.5
SF1	splicing factor 1	HGNC:12950	11q13
SH2B1	SH2B adaptor protein 1	HGNC:30417	16p11.2
SHOX	short stature homeobox	HGNC:10853	Xp22.33 and Yn11.32
SLC12A1	solute carrier family 12 member 1	HGNC:10910	15q21.1
SLC12A3	solute carrier family 12 member 3	HGNC:10912	16q13
SLC19A2	solute carrier family 19 member 2	HGNC:10938	1q23.3
SLC26A4	solute carrier family 26 member 4	HGNC:8818	7q31
SLC2A2	solute carrier family 2 member 2	HGNC:11006	3q26.2
SLC34A1	solute carrier family 34 member 1	HGNC:11019	5q35.3
SLC34A3	solute carrier family 34 member 3	HGNC:20305	9q34.3
SLC4A1	solute carrier family 4 member 1 (Diego blood group)	HGNC:11027	17q21.31
SLC7A7	solute carrier family 7 member 7	HGNC:11065	14q11.2
SLC7A9	solute carrier family 7 member 9	HGNC:11067	19q13.11
SLC9A3R1	SLC9A3 regulator 1	HGNC:11075	17q25.1
SOS1	SOS Ras/Rac guanine nucleotide exchange factor 1	HGNC:11187	2p21
SOST	sclerostin	HGNC:13771	17q21.31
SOX10	SRY-box 10	HGNC:11190	22q13.1
SOX2	SRY-box 2	HGNC:11195	3q26.33
SOX3	SRY-box 3	HGNC:11199	Xq27.1
SOX9	SRY-box 9	HGNC:11204	17q24.3

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SQSTM1	sequestosome 1	HGNC:11280	5q35
SRD5A2	steroid 5 alpha-reductase 2	HGNC:11285	2p23.1
SREBF1	sterol regulatory element binding transcription factor 1	HGNC:11289	17p11.2
SRY	sex determining region Y	HGNC:11311	Yp11.3
STAR	steroidogenic acute regulatory protein	HGNC:11359	8p11.2
STAT3	signal transducer and activator of transcription 3 (acute-phase response factor)	HGNC:11364	17q21
TAC3	tachykinin 3	HGNC:11521	12q13.3
TACR3	tachykinin receptor 3	HGNC:11528	4q25
TBCE	tubulin folding cofactor E	HGNC:11582	1q42.3
TBX1	T-box 1	HGNC:11592	22q11.21
THRB	thyroid hormone receptor beta	HGNC:11799	3p24.2
TMEM127	transmembrane protein 127	HGNC:26038	2q11.2
TMEM38B	transmembrane protein 38B	HGNC:25535	9q31.3
TNFRSF11A	tumor necrosis factor receptor superfamily member 11a	HGNC:11908	18q22.1
TNFRSF11B	tumor necrosis factor receptor superfamily member 11b	HGNC:11909	8q24
VDR	vitamin D (1,25- dihydroxyvitamin D3) receptor	HGNC:12679	12q13.11
VHL	von Hippel-Lindau tumor suppressor, E3 ubiquitin protein ligase	HGNC:12687	3p25.3
WDR11	WD repeat domain 11	HGNC:13831	10q26
WFS1	wolframin ER transmembrane glycoprotein	HGNC:12762	4p16.1
WNT1	wingless-type MMTV integration site family member 1	HGNC:12774	12q13.12
WNT4	wingless-type MMTV integration site family member 4	HGNC:12783	1p36.23-p35.1
WT1	Wilms tumor 1	HGNC:12796	11p13