

LMM / eMERGE III Network Reference Sequences

October 2016

LMM / eMERGE III Network Consensus Actionable Gene List

*ACMG56 gene

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Gene	Region	Transcript
ACTA2*	Exon 02-09	NM_001613.2
ACTC1*	Exon 01-06	NM_005159.4
APC*	Exon 01-15	NM_000038.4
APC*	Exon 01A	NM_001127511.2
APOB*	Exon 01-29	NM_000384.2
BMPR1A	Exon 03-13	NM_004329.2
BRCA1*	Exon 02A-23A	NM_007294.3
BRCA1*	Exon 13	NM_007300.3
BRCA2*	Exon 02-27	NM_000059.3
CACNA1A	Exon 01-48	NM_023035.2
CACNA1A	Exon 37D	NM_001127221.1
CACNA1S*	Exon 01-44	NM_000069.2
COL3A1*	Exon 01-51	NM_000090.3
COL5A1	Exon 01-66	NM_000093.4
COL5A1	Exon 64A	NM_001278074.1
DSC2*	Exon 01-16	NM_024422.3
DSC2*	Exon 15A	NM_024422.3
DSC2*	Exon 15B	NM_004949.3

Gene	Region	Transcript
DSG2*	Exon 01-15	NM_001943.3
DSP*	Exon 01-24	NM_004415.2
FBN1*	Exon 01-65	NM_000138.4
GLA*	Exon 01-07	NM_000169.2
GLA*	Intron 04 (c.640-801G>A; c.639+919G>A)	NM_000169.2
HNF1A	Exon 01-10	NM_000545.5
HNF1B	Exon 01-09	NM_000458.2
KCNE1	Exon 03	NM_000219.3
KCNH2*	Exon 01-15	NM_000238.3
KCNH2*	Exon 01B	NM_172057.2
KCNH2*	Exon 09A	NM_172056.2
KCNJ2	Exon 02	NM_000891.2
KCNQ1*	Exon 01-16	NM_000218.2
KCNQ1*	Exon 01A	NM_181798.1
LDLR*	Exon 01-18	NM_000527.4
LMNA*	Exon 01-12	NM_170707.2
LMNA*	Exon 01B	NM_001257374.2
LMNA*	Exon 13B	NM_001257374.2

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Gene	Region	Transcript
MEN1*	Exon 03-11	NM_130804.2
MLH1*	Exon 01-19	NM_000249.3
MSH2*	Exon 01-16	NM_000251.2
MSH6*	Exon 01-10	NM_000179.2
MUTYH*	Exon 01-16	NM_001128425.1
MYBPC3*	Exon 01-34	NM_000256.3
MYH11*	Exon 02-42	NM_001040113.1
MYH11*	Exon 42A	NM_001040114.1
MYH7*	Exon 03-40	NM_000257.2
MYL2*	Exon 01-07	NM_000432.3
MYL3*	Exon 01-06	NM_000258.2
MYLK*	Exon 04-34	NM_053025.3
NF2*	Exon 01-15	NM_016418.5
NF2*	Exon 16B	NM_000268.3
NF2*	Exon 16C	NM_181825.2
OTC	Exon 01-10	NM_000531.5
PALB2	Exon 01-13	NM_024675.3

Gene	Region	Transcript
PCSK9*	Exon 01-12	NM_174936.3
PKP2*	Exon 01-14	NM_004572.3
PMS2*	Exon 01-15	NM_000535.5
POLD1	Exon 02A	NM_001256849.1
POLD1	Exon 03-27	NM_002691.3
POLE	Exon 01-49	NM_006231.2
PRKAG2*	Exon 01-16	NM_016203.3
PTEN*	Exon 01-09	NM_000314.4
RB1*	Exon 01-27	NM_000321.2
RET*	Exon 01-20	NM_020975.4
RET*	Exon 19A	NM_020630.4
RYR1*	Exon 01-106	NM_000540.2
RYR2*	Exon 01-105	NM_001035.2
SCN5A*	Exon 02-28	NM_198056.2
SCN5A*	Exon 06A	NM_001099404.1
SDHAF2*	Exon 01-04	NM_017841.2
SDHB*	Exon 01-08	NM_003000.2

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SDHC*	Exon 01-05	NM_003001.3
SDHC*	Exon 05A	NM_001035511.1
SDHD*	Exon 01-04	NM_003002.2
SMAD3*	Exon 01-09	NM_005902.3
SMAD3*	Exon 01B	NM_001145103.1
SMAD4	Exon 02-12	NM_005359.5
STK11*	Exon 01-09	NM_000455.4
TGFBR1*	Exon 01-09	NM_004612.2
TGFBR2*	Exon 01-07	NM_003242.5
TGFBR2*	Exon 01A	NM_001024847.2
TMEM43*	Exon 01-12	NM_024334.2
TNNI3*	Exon 01-08	NM_000363.4
TNNT2*	Exon 02-16	NM_001001430.1
TNNT2*	Exon 04A	NM_000364.2
TP53*	Exon 02-11	NM_000546.4
TP53*	Exon 10A	NM_001126114.2
TP53*	Exon 10B	NM_001126113.2
TPM1*	Exon 01-09, 06A	NM_000366.5
TPM1*	Exon 06B, 10	NM_001018005.1

Gene	Region	Transcript
TPM1*	Exon 02A, 10A	NM_001018008.1
TPM1*	Exon 01A, 10B	NM_001018020.1
TSC1*	Exon 03-23	NM_000368.4
TSC2*	Exon 02-42	NM_000548.3
VHL*	Exon 01-03	NM_000551.2
WT1*	Exon 01-10	NM_024426.4
WT1*	Exon 01B	NM_001198551.1

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LMM / eMERGE III Network Consensus Actionable "SNV" List

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Gene/Build	Transcript/Chr	Region	AA	rsID
ACADM	NM_001127328.1	c.997A>G	p.Lys333Glu	rs77931234
ALDOB	NM_000035.3	c.360_363delCAAA	p.Asn120LysfsX32	rs387906225
BCKDHB	NM_000056.3	c.548G>C	p.Arg183Pro	rs79761867
BCKDHB	NM_000056.3	c.832G>A	p.Gly278Ser	rs386834233
BLM	NM_000057.2	c.2207_2212delins	p.Tyr736LeufsX5	rs113993962 (delinsTAGATTC)
CPT2	NM_000098.2	c.1239_1240delGA	p.Lys414ThrfsX7	rs397509431 (c.1238_1239delAG)
CYP21A2	NM_000500.7	c.293-13C>G	p.?	rs6467
F5	NM_000130.4	c.1601A	p.Gln534	rs6025
FAH	NM_000137.2	c.782C>T	p.Pro261Leu	rs80338898
G6PC	NM_000151.3	c.247C>T	p.Arg83Cys	rs1801175
HFE	NM_000410.3	c.845G>A	p.Cys282Tyr	rs1800562
MEFV	NM_000243.2	c.2177T>C	p.Val726Ala	rs28940579
MEFV	NM_000243.2	c.2080A>G	p.Met694Val	rs61752717
MSH2*	NM_000251.1	c.942+3A>T	NA	rs193922376

MSH2* We cannot exclude the presence of a variant in this location due to challenging sequence

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Additional eMERGE III Network Reportable Genes Available to Sites (by specific request)

Gene	Region	Transcript
ANGPTL3	Exon 01-07	NM_014495.3
ANGPTL4	Exon 01-07	NM_139314.1
ANK2	Exon 01-46	NM_001148.4
ANK2	Exon 02B, 28B	NM_001127493.1
APOA5	Exon 02-04	NM_052968.4
APOC3	Exon 02-04	NM_000040.1
APOE	Exon 02-04	NM_000041.2
ATM	Exon 02-63	NM_000051.3
ATP1A2	Exon 01-23	NM_000702.3
BMPR2	Exon 01-13	NM_001204.6
CACNA1B	Exon 01-47	NM_000718.3
CACNA1C	Exon 01-50	NM_199460.2
CACNA1C	Exon 08V, 42V	NM_001167625.1
CACNA1C	Exon 32C, 39C	NM_001129829.1
CFH	Exon 01-22	NM_000186.3
CFH	Exon 10A	NM_001014975.2
CFTR	Exon 01-27	NM_000492.3
CFTR	Intron 12 (c.1680-886A>G)	NM_000492.3
CFTR	Intron 22 (c.3808-2477C>T)	NM_000492.3

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Gene	Region	Transcript
CHEK2	Exon 02B-15B	NM_007194.3
CHEK2	Exon 03	NM_001005735.1
CORIN	Exon 01-22	NM_006587.3
CORIN	Exon 14B	NM_001278586.1
FLG	Exon 02	NM_002016.1
<i>Not covered</i>	<i>Exon 03</i>	<i>NM_002016.1</i>
GRM1	Exon 02-09	NM_001278065.1
GRM1	Exon 08B	NM_001278064.1
GRM2	Exon 02-06	NM_000839.3
GRM5	Exon 01-09	NM_001143831.2
GRM7	Exon 01-10	NM_181874.2
GRM7	Exon 10A	NM_000844.3
GRM8	Exon 02-10	NM_001127323.1
GRM8	Exon 10A	NM_000845.2
IL33	Exon 02-08	NM_033439.3
IL4	Exon 01-04	NM_000589.3
MC4R	Exon 01	NM_005912.2
MTHFR	Exon 02-12	NM_005957.4

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Gene	Region	Transcript
NTRK1	Exon 01-17	NM_002529.3
NTRK1	Exon 01A, 02A	NM_001007792.1
PLTP	Exon 02-16	NM_006227.3
PON1	Exon 01-09	NM_000446.5
SCN1A	Exon 03-28	NM_001202435.1
SCN9A	Exon 02-27	NM_002977.3
SERPINA1	Exon 04-07	NM_001127701.1
SLC25A40	Exon 03-12	NM_018843.3
SLC2A10	Exon 01-05	NM_030777.3
TCF4	Exon 01-20	NM_001243226.1
TCF4	Exon 01D	NM_001243230.1
TCF4	Exon 01F	NM_001243231.1
TCF4	Exon 01G	NM_001243232.1
TCF4	Exon 01I	NM_001243234.1
TCF4	Exon 01K	NM_001243236.1
TCF4	Exon 09A	NM_001243228.1
TCIRG1	Exon 02-20	NM_006019.3
TNF	Exon 01-04	NM_000594.3
TSLP	Exon 01-04	NM_033035.4

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Gene	Region	Transcript
TTR	Exon 01-04	NM_000371.3
TYK2	Exon 03-25	NM_003331.4
UMOD	Exon 02-12	NM_001278614.1
VDR	Exon 01A, 02A, 03A	NM_001017536.1
VDR	Exon 05-11	NM_001017535.1

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Additional eMERGE III Network Reportable "SNVs" Available to Sites (by specific request) For a full list of positions genotyped, contact the LMM

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Gene/Build	Transcript/Chr	Region	AA	rsID
ABCC8	NM_000352.3	c.3989-9G>A	NA	rs151344623
ADAMTS2	NM_014244.4	c.673C>T	p.Gln225X	rs137853146
APOB	NM_000384.2	c.10580G>A	p.Arg3527Gln	rs5742904 (p.Arg3500Gln)
APOB	NM_000384.2	c.10579C>T	p.Arg3527Trp	rs144467873 (p.Arg3500Trp)
APOC3	NM_000040.1	c.179+1G>T	NA	rs140621530
APOC3	NM_000040.1	c.55+1G>A	NA	rs138326449
ARSA	NM_001085425.2	c.1136C>T	p.Pro379Leu	rs74315478
ASPA	NM_001128085.1	c.693C>A	p.Tyr231X	rs12948217
ASPA	NM_001128085.1	c.854A>C	p.Glu285Ala	rs28940279
ATP7B	NM_001243182.1	c.2874C>A	p.His958Gln	rs76151636
CARD9	NM_052813.4	c.1434+1G>C	NA	rs141992399
CHD8	NM_020920.3	c.2875C>T	p.Gln959X	rs397514551
CHD8	NM_020920.3	c.3172C>T	p.Arg1058X	rs397514552
CLRN1	NM_174878.2	c.144T>G	p.Asn48Lys	rs111033258
COL4A5	NM_033380.2	c.5048G>A	p.Arg1683Gln	rs104886308
COL5A2	NM_000393.3	c.1924-2_1928del	p.?	rs786205103 (delAGGGAGC)
COL5A2	NM_000393.3	c.2031+1G>T	NA	rs786205104
COL5A2	NM_000393.3	c.3445G>C	p.Gly1149Arg	rs121912930
COQ4	NM_016035.3	c.718C>T	p.Arg240Cys	rs143441644
CPT2	NM_000098.2	c.1342T>C	p.Phe448Leu	rs74315297
CYP1B1	NM_000104.3	c.1103G>A	p.Arg368His	rs79204362
CYP1B1	NM_000104.3	c.1159G>A	p.Glu387Lys	rs55989760
DHCR7	NM_001163817.1	c.964-1G>C	NA	rs138659167
DHCR7	NM_001360.2	c.453G>A	p.Trp151X	rs104894213
DHDDS	NM_024887.3	c.124A>G	p.Lys42Glu	rs147394623
DLD	NM_000108.3	c.685G>T	p.Gly229Cys	rs121964990
F11	NM_000128.3	c.403G>T	p.Glu135X	rs121965063
F11	NM_000128.3	c.901T>C	p.Phe301Leu	rs121965064
FAM161A	NM_001201543.1	c.1355_1356delCA	p.Thr452SerfsX3	rs397704718
FANCC	NM_000136.2	c.456+4A>T	NA	rs104886456
FKTN	NM_001079802.1	c.1167_1168insA	p.Phe390IlefsX14	rs398123555 (c.1167dup)

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Gene/Build	Transcript/Chr	Region	AA	rsID
GBA	NM_001005741.2	c.27+1G>A	NA	rs80356759
GJB2	NM_004004.5	c.101T>C	p.Met34Thr	rs35887622
HEXA	NM_000520.4	c.1274_1277dup	p.Tyr427IlefsX5	rs387906309 (dupTATC)
HEXA	NM_000520.4	c.1421+1G>C	NA	rs147324677
HPS3	NM_032383.3	c.1163+1G>A	NA	rs201227603
JAK2	NM_004972.3	c.1849G>A	p.Val617Ile	rs77375493
KCNE2	NM_172201.1	c.161T>C	p.Met54Thr	rs74315447
KCNE2	NM_172201.1	c.29C>T	p.Thr10Met	rs199473648
LRRK2	NM_198578.3	c.6055G>A	p.Gly2019Ser	rs34637584
MCOLN1	NM_020533.2	c.406-2A>G	NA	rs104886461
MEFV	NM_000243.2	c.1105C>T	p.Pro369Ser	rs11466023
MEFV	NM_000243.2	c.2084A>G	p.Lys695Arg	rs104895094
MEFV	NM_000243.2	c.442G>C	p.Glu148Gln	rs3743930
MPL	NM_005373.2	c.79+2T>A	NA	rs146249964
MTTP	NM_000253.2	c.2593G>T	p.Gly865X	rs146064714
PCDH15	NM_033056.3	c.733C>T	p.Arg245X	rs111033260
PEX2	NM_001172086.1	c.355C>T	p.Arg119X	rs61752123
PHGDH	NM_006623.3	c.1468G>A	p.Val490Met	rs121907987
PMM2	NM_000303.2	c.422G>A	p.Arg141His	rs28936415
RTEL1	NM_032957.4	c.3724+139G>A	NA	rs201540674
SLC1A4	NM_003038.4	c.766G>A	p.Glu256Lys	rs201278558
SMPD1	NM_000543.4	c.1493G>T	p.Arg498Leu	rs120074117
SMPD1	NM_000543.4	c.1829_1831delGCC	p.Arg610del	rs120074118
SMPD1	NM_000543.4	c.911T>C	p.Leu304Pro	rs120074124
SMPD1	NM_000543.4	c.996delC	p.Phe333SerfsX52	rs387906289
SPG7	NM_003119.2	c.1529C>T	p.Ala510Val	rs61755320
TERT	NM_198253.2	c.1892G>A	p.Arg631Gln	rs199422294
TMEM216	NM_001173991.2	c.218G>T	p.Arg73Leu	rs201108965
TOR1A	NM_000113.2	c.907_909delGAG	p.Glu303del	rs724159981 (c.907_909del)