

SUPPLEMENTARY MATERIAL**Whole exome sequencing in a family with thromboangiitis obliterans**

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Table: VTE candidate genes

Gene_symbol	Gene_name
A4GALT	<i>alpha 1,4-galactosyltransferase (P blood group)</i>
ABCA6	<i>ATP binding cassette subfamily A member 6</i>
ABO	<i>ABO, alpha 1-3-N-acetylgalactosaminyltransferase and alpha 1-3-galactosyltransferase</i>
ACE	<i>angiotensin converting enzyme</i>
ADAMTS13	<i>ADAM metalloproteinase with thrombospondin type 1 motif 13</i>
AGBL1	<i>ATP/GTPbinding protein like1</i>
ANXA5	<i>annexin A5</i>
APOH	<i>apolipoprotein H</i>
ARID4A	<i>AT-rich Interaction domain 4A</i>
ASH2L	<i>ASH2 like, histone lysine methyltransferase complex subunit</i>
BAZ1B	<i>bromodomain adjacent to zinc finger domain 1B</i>
BCOR	<i>BCL6 corepressor</i>
BRCC3	<i>BRCA1/BRCA2-containing complex subunit 3</i>
C1orf42	<i>chromosome 12 open reading frame 42</i>
C1orf198	<i>chromosome 1 open reading frame 198</i>
C4BPA	<i>complement component 4 binding protein alpha</i>
C4BPB	<i>complement component 4 binding protein beta</i>
CACFD1	<i>calcium channel flower domain containing 1</i>
CADM1	<i>cell adhesion molecule 1</i>
CALR	<i>calreticulin</i>
CATSPERB	<i>cation channel sperm associated auxiliary subunit B</i>

Gene_symbol	Gene_name
CBS	<i>cystathionine-beta-synthase</i>
CD36	<i>CD36 molecule</i>
CD93	<i>CD93 molecule</i>
CHD3	<i>chromodomain helicase DNA binding protein 3</i>
CNOT3	<i>CCR4-NOT transcription complex subunit 3</i>
CPB2	<i>carboxypeptidase B2</i>
CRP	<i>C-reactive protein</i>
CSRNP1	<i>cysteine and serine rich nuclear protein 1</i>
CYP1A2	<i>cytochrome P450, family 1 subfamily A member 2</i>
CYP27C1	<i>cytochrome P450 family 27 subfamily C member 1</i>
CYP2C9	<i>cytochrome P450, family 2 subfamily C member 9</i>
CYP3A5	<i>cytochrome P450, family 3, subfamily A member 5</i>
CYP4V2	<i>cytochrome P450, family 4, subfamily V member 2</i>
DENND2D	<i>DENN domain containing 2D</i>
EDEM2	<i>ER degradation-enhancing alpha-mannosidase-like 2</i>
EFEMP2	<i>EGF-containing fibulin-like extracellular matrix protein 2</i>
EIF5A	<i>eukaryotic translation initiation factor 5A</i>
EPHA3	<i>EPH receptor A3</i>
F10	<i>coagulation factor X</i>
F11	<i>coagulation factor XI</i>
F12	<i>coagulation factor XII</i>
F13A1	<i>coagulation factor XIII A chain</i>
F13B	<i>coagulation factor XIII B chain</i>
F2	<i>coagulation factor II</i>

Gene_symbol	Gene_name
F2R	<i>coagulation factor II thrombin receptor</i>
F2RL1	<i>F2R like trypsin receptor 1</i>
F2RL2	<i>coagulation factor II thrombin receptor-like 2</i>
F2RL3	<i>coagulation factor II thrombin receptor-like 3</i>
F3	<i>coagulation factor III</i>
F5	<i>coagulation factor V</i>
F7	<i>coagulation factor VII</i>
F8	<i>coagulation factor VIII</i>
F9	<i>coagulation factor IX</i>
FGA	<i>fibrinogen alpha chain</i>
FGB	<i>fibrinogen beta chain</i>
FGG	<i>fibrinogen gamma chain</i>
FOXA2	<i>forkhead box A2</i>
GAS6	<i>growth arrest-specific 6</i>
GCKR o GCK	<i>glucokinase regulator protein</i>
GGCX	<i>gamma-glutamyl carboxylase</i>
GP1BA	<i>glycoprotein Ib platelet subunit alpha</i>
GP1BB	<i>glycoprotein Ib platelet subunit beta</i>
GP5	<i>glycoprotein V platelet</i>
GP6	<i>glycoprotein VI platelet</i>
GP9	<i>glycoprotein IX platelet</i>
GPR25	<i>G protein-coupled receptor 25</i>
GRK5	<i>G protein-coupled receptor kinase 5</i>
HABP2	<i>hyaluronan binding protein 2</i>

Gene_symbol	Gene_name
HIVEP1	<i>human immunodeficiency virus type I enhancer binding protein 1</i>
HLA-C	<i>major histocompatibility complex, class I, C</i>
HRG	<i>histidine-rich glycoprotein</i>
HSPBAP1	<i>HSPB1 associated protein 1</i>
IL13	<i>interleukin 13</i>
IL1A	<i>interleukin 1 alpha</i>
IL4	<i>interleukin 4</i>
IL6	<i>interleukin 6</i>
ITGA1	<i>integrin subunit alpha 1</i>
ITGA2	<i>integrin subunit alpha 2</i>
ITGA2B	<i>integrin subunit alpha 2b</i>
ITGB2	<i>integrin subunit beta 2</i>
ITGB3	<i>integrin subunit beta 3</i>
JAK2	<i>Janus kinase 2</i>
KCNQ3	<i>potassium voltage-gated channel subfamily Q member 3</i>
KIF26B	<i>kinesin family member 26B</i>
KLF2	<i>KLF transcription factor 2</i>
KLK1	<i>kallikrein 1</i>
KLK10	<i>kallikrein-related peptidase 10</i>
KLK11	<i>kallikrein-related peptidase 11</i>
KLK12	<i>kallikrein-related peptidase 12</i>
KLK13	<i>kallikrein-related peptidase 13</i>
KLK14	<i>kallikrein-related peptidase 14</i>
KLK15	<i>kallikrein-related peptidase 15</i>

Gene_symbol	Gene_name
KLK2	<i>kallikrein-related peptidase 2</i>
KLK3	<i>kallikrein-related peptidase 3</i>
KLK4	<i>kallikrein-related peptidase 4</i>
KLK5	<i>kallikrein-related peptidase 5</i>
KLK6	<i>kallikrein-related peptidase 6</i>
KLK7	<i>kallikrein-related peptidase 7</i>
KLK8	<i>kallikrein-related peptidase 8</i>
KLK9	<i>kallikrein-related peptidase 9</i>
KLKB1	<i>kallikrein B1</i>
KNG1	<i>kininogen 1</i>
LMOD1	<i>leiomodisin 1</i>
LRP4	<i>LDL receptor related protein 4</i>
LYST	<i>lysosomal trafficking regulator</i>
MAST2	<i>microtubule associated serine/threonine kinase 2</i>
MFGE8	<i>milk fat globule-EGF factor 8 protein</i>
MPHOSPH9	<i>M-phase phosphoprotein 9</i>
MPL	<i>MPL proto-oncogene,thrombopoietin receptor</i>
MTHFR	<i>methylene tetrahydrofolate reductase</i>
MYRF	<i>myelin regulatory factor</i>
NAT8B	<i>N-acetyltransferase 8B</i>
NCAM2	<i>neuronal cell adhesion molecule 2</i>
NCOA1	<i>nuclear receptor coactivator 1</i>
NLRP2	<i>NLR family pyrin domain containing 2</i>
NOS3	<i>nitric oxide synthase 3</i>

Gene_symbol	Gene_name
NQO1	<i>NAD(P)H quinone dehydrogenase 1</i>
NR1I2	<i>nuclear receptor subfamily 1 group I member 2</i>
NRG3	<i>neuregulin 3</i>
NUGGC	<i>nuclear GTPase, germinal center associated</i>
OSMR-AS1	<i>OSMR antisense RNA 1</i>
PAR1	<i>Prader-Willi/Angelman region-1</i>
PDIA2	<i>protein disulfide isomerase family A member 2</i>
PEAR1	<i>platelet endothelial aggregation receptor 1</i>
PEPD	<i>peptidase D</i>
PF4	<i>platelet factor 4</i>
PLAT	<i>tissue plasminogen activator</i>
PLAU	<i>urinary plasminogen activator</i>
PLAUR	<i>urinary plasminogen activator receptor</i>
PLCG2	<i>phospholipase C gamma 2</i>
PLEK	<i>pleckstrin</i>
PLG	<i>plasminogen</i>
POLE4	<i>DNA polimerase epsilon 4, accessory subunit</i>
PPIF	<i>peptidylprolyl isomerase F</i>
PROC	<i>protein C, inactivator of coagulation factors Va and VIIIa</i>
PROCR	<i>protein C receptor</i>
PROS1	<i>protein S</i>
PROZ	<i>protein Z, vitamin K-dependent plasma glycoprotein</i>
PSG8	<i>pregnancy specific beta 1-glycoprotein 8</i>
PTGIR	<i>prostaglandin I2 receptor</i>

Gene_symbol	Gene_name
PTGIS	<i>prostaglandin I2 synthase</i>
QSER1	<i>glutamine and serine rich 1</i>
RGS7	<i>regulator of G-protein signaling 7</i>
RGSL1	<i>regulator of G protein signaling like 1</i>
SAA1	<i>serum amyloid A1</i>
SAA2	<i>serum amyloid A2</i>
SAA4	<i>serum amyloid A4, constitutive</i>
SBNO1	<i>strawberry notch homolog 1</i>
SCARA5	<i>scavenger receptor class A member 5</i>
SELE	<i>selectin E</i>
SELP	<i>selectin P</i>
SERPINA1	<i>serpin family A member 1</i>
SERPINA10	<i>serpin family A member 10</i>
SERPINA5	<i>serpin family A member 5</i>
SERPINB2	<i>serpin family B member 2</i>
SERPINC1	<i>serpin family C member 1</i>
SERPIND1	<i>serpin family D member 1</i>
SERPINE1	<i>serpin family E member 1</i>
SERPINE2	<i>serpin family E member 2</i>
SERPINF1	<i>serpin family F member 1</i>
SERPINF2	<i>serpin family F member 2</i>
SERPING1	<i>serpin family G member 1</i>
SH2B3/LNK	<i>SH2B adapter protein 3</i>
SHBG	<i>sex hormone-binding globulin</i>

Gene_symbol	Gene_name
SLC44A2	<i>solute carrier family 44 member 2</i>
SLC4A1	<i>solute carrier family 4 member 1 (Diego blood group)</i>
SMG6	<i>SMG6 nonsense mediated mRNA decay factor</i>
SPP1	<i>secreted phosphoprotein 1</i>
STAB2	<i>stabilin 2</i>
STX10	<i>syntaxin 10</i>
STX2	<i>syntaxin 2</i>
STXBP5	<i>syntaxin binding protein 5</i>
SULT1A1	<i>sulfotransferase family 1A member 1</i>
TC2N	<i>tandem C2 domains, nuclear</i>
TFPI	<i>tissue factor pathway inhibitor</i>
TFPI2	<i>tissue factor pathway inhibitor 2</i>
THBD	<i>thrombomodulin</i>
THBS1	<i>thrombospondin 1</i>
TMCO1	<i>transmembrane and coiled coil domains1</i>
TNF	<i>tumor necrosis factor</i>
TNFRSF11B	<i>TNF receptor superfamily member 11b</i>
TSPAN15	<i>tetraspanin 15</i>
U2SURP	<i>U2 snRNP associated SURP domain containig</i>
UGT1A1	<i>UDP glucuronosyltransferase family 1 member A1</i>
UGT1A3	<i>UDP glucuronosyltransferase family 1 member A3</i>
UGT1A9	<i>UDP glucuronosyltransferase family 1 member A9</i>
UGT2B7	<i>UDP glucuronosyltransferase family 2 member B7</i>
USF1	<i>upstream transcription factor 1</i>

Gene_symbol	Gene_name
VKORC1	<i>vitamin K epoxide reductase complex subunit 1</i>
VTN	<i>vitronectin</i>
VWF	<i>von Willebrand factor</i>
ZFPM2	<i>zinc finger protein, FOG family member 2</i>

Table: TAO candidate genes

Gene_symbol	Gene_name	References
IL17RC	interleukin 17 receptor C	<i>Shi ZF et al, 2016</i>
SEMA5B	semaphorin 5B	<i>Shi ZF et al, 2016</i>
CD14	CD14 molecule	<i>Sun XL et al, 2017</i>
HLA-A	major histocompatibility complex, class I, A	<i>Sun XL et al, 2017</i>
HLA-B	major histocompatibility complex, class I, B	<i>Sun XL et al, 2017</i>
HLA-DPB1	major histocompatibility complex, class II, DP beta 1	<i>Sun XL et al, 2017</i>
HLA-DRB1	major histocompatibility complex, class II, DR beta 1	<i>Sun XL et al, 2017</i>
IL33	interleukin 33	<i>Sun XL et al, 2017</i>
MyD88	MYD88 innate immune signal transduction adaptor	<i>Sun XL et al, 2017</i>
NOS3	nitric oxide synthase 3	<i>Sun XL et al, 2017</i>
ST2	suppression of tumorigenicity 2	<i>Sun XL et al, 2017</i>

Table: TAO transcriptome mRNA Chen B

mRNA	regulation	gene	References
ABCD2	down	ATP binding cassette subfamily D member 2	Chen B et al, 2021
ADGRA3	down	adhesion G protein-coupled receptor A3	Chen B et al, 2021
AGPAT1	up	1-acylglycerol-3-phosphate O-acyltransferase 1	Chen B et al, 2021
AKAP5	down	A-kinase anchoring protein 5	Chen B et al, 2021
ALDH5A1	up	aldehyde dehydrogenase 5 family member A1	Chen B et al, 2021
AOC3	up	amine oxidase copper containing 3	Chen B et al, 2021
AR	down	androgen receptor	Chen B et al, 2021
C12orf74	down	PLEKHG7 pleckstrin homology and RhoGEF domain containing G7	Chen B et al, 2021
CADM1	down	cell adhesion molecule 1	Chen B et al, 2021
CAPN5	up	calpain 5	Chen B et al, 2021
CCDC141	down	coiled-coil domain containing 141	Chen B et al, 2021
CTTNBP2	down	cortactin binding protein 2	Chen B et al, 2021
CXCL8	down	C-X-C motif chemokine ligand 8	Chen B et al, 2021
DSC1	down	desmocollin 1	Chen B et al, 2021
DVL3	up	dishevelled segment polarity protein 3	Chen B et al, 2021
EML3	up	EMAP like 3	Chen B et al, 2021
ENPP4	down	ectonucleotide pyrophosphatase/phosphodiesterase 4	Chen B et al, 2021

mRNA	regulation	gene	References
FAM218A	down	family with sequence similarity 218 member A	Chen B et al, 2021
FPR3	down	formyl peptide receptor 3	Chen B et al, 2021
GCNT4	down	glucosaminyl (N-acetyl) transferase 4	Chen B et al, 2021
GNA12	up	G protein subunit alpha 12	Chen B et al, 2021
HPCAL1	up	hippocalcin like 1	Chen B et al, 2021
IGSF10	down	immunoglobulin superfamily member 10	Chen B et al, 2021
KIF21A	down	kinesin family member 21A	Chen B et al, 2021
KLF13	up	KLF transcription factor 13	Chen B et al, 2021
LDB2	down	LIM domain binding 2	Chen B et al, 2021
LRRC8A	up	orthologs from human, Leucine rich repeat containing 8 VRAC subunit A	Chen B et al, 2021
LRRN3	down	leucine rich repeat neuronal 3	Chen B et al, 2021
MFSD2B	up	MFSD2 lysolipid transporter B, sphingolipid	Chen B et al, 2021
MINK1	up	misshapen like kinase 1	Chen B et al, 2021
NAP1L2	down	nucleosome assembly protein 1 like 2	Chen B et al, 2021
NDUFAF4	down	NADH:ubiquinone oxidoreductase complex assembly factor 4	Chen B et al, 2021
NLGN4Y	down	neuroligin 4 Y-linked	Chen B et al, 2021
NUAK1	down	NUAK family kinase 1	Chen B et al, 2021
NUAK2	up	NUAK family kinase 2	Chen B et al, 2021
OSBPL6	up	oxysterol binding protein like 6	Chen B et al, 2021
PLPP7	up		Chen B et al, 2021

mRNA	regulation	gene	References
		<i>phospholipid phosphatase 7 (inactive)</i>	
<i>RANBP10</i>	up	<i>RAN binding protein 10</i>	<i>Chen B et al, 2021</i>
<i>RCAN2</i>	down	<i>regulator of calcineurin 2</i>	<i>Chen B et al, 2021</i>
<i>RPS6KA6</i>	down	<i>ribosomal protein S6 kinase A6</i>	<i>Chen B et al, 2021</i>
<i>RUNDC1</i>	up	<i>RUN domain containing 1</i>	<i>Chen B et al, 2021</i>
<i>SDC2</i>	down	<i>syndecan 2</i>	<i>Chen B et al, 2021</i>
<i>SH2D1A</i>	down	<i>SH2 domain containing 1A</i>	<i>Chen B et al, 2021</i>
<i>SH2D1B</i>	down	<i>SH2 domain containing 1B</i>	<i>Chen B et al, 2021</i>
<i>SNX22</i>	up	<i>sorting nexin 22</i>	<i>Chen B et al, 2021</i>
<i>SOCS6</i>	down	<i>suppressor of cytokine signaling 6</i>	<i>Chen B et al, 2021</i>
<i>STMN1</i>	down	<i>stathmin 1</i>	<i>Chen B et al, 2021</i>
<i>TIGIT</i>	down	<i>T cell immunoreceptor with Ig and ITIM domains</i>	<i>Chen B et al, 2021</i>
<i>TRIM58</i>	up	<i>tripartite motif containing 58</i>	<i>Chen B et al, 2021</i>
<i>TTN</i>	down	<i>titin</i>	<i>Chen B et al, 2021</i>
<i>ZNF471</i>	down	<i>zinc finger protein 471</i>	<i>Chen B et al, 2021</i>
<i>ZNF705E</i>	down	<i>zinc finger protein 705E</i>	<i>Chen B et al, 2021</i>

Table: TAO transcriptome mRNA Oztan G

Gene_symbol	Gene_name	References	Regulation
ANPEP	alanyl (membrane) aminopeptidase	<i>Oztan G et al, 2024</i>	down
CCL4; CCL4L1; CCL4L2	chemokine (C-C motif) ligand 4; chemokine (C-C motif) ligand 4-like 1; chemokine (C-C motif) ligand 4-like 2	<i>Oztan G et al, 2024</i>	down
DGKZ	diacylglycerol kinase, zeta	<i>Oztan G et al, 2024</i>	down
EGR1	early growth response 1	<i>Oztan G et al, 2024</i>	down
EIF4A1; SNORD10	eukaryotic translation initiation factor 4A1; Small nucleolar RNA, C/D box 10	<i>Oztan G et al, 2024</i>	up
EIF4A2; SNORA63	eukaryotic translation initiation factor 4A2; Small nucleolar RNA, H/ACA box 63	<i>Oztan G et al, 2024</i>	up
EIF4G2; SNORD97	eukaryotic translation initiation factor 4 γ, 2; Small nucleolar RNA, C/D box 97	<i>Oztan G et al, 2024</i>	up
FMNL1	formin like 1	<i>Oztan G et al, 2024</i>	up
HBB	hemoglobin, β	<i>Oztan G et al, 2024</i>	down
LAMP2	lysosomal-associated membrane protein 2	<i>Oztan G et al, 2024</i>	down
NDUFA1	NADH dehydrogenase (ubiquinone) 1 α subcomplex, 1, 7.5kDa	<i>Oztan G et al, 2024</i>	down
NEIL3	nei-like DNA glycosylase 3	<i>Oztan G et al, 2024</i>	up
PHC3	polyhomeotic homolog 3 (Drosophila)	<i>Oztan G et al, 2024</i>	up
PLP2	proteolipid protein 2 (colonic epithelium-enriched)	<i>Oztan G et al, 2024</i>	down

Gene_symbol	Gene_name	References	Regulation
PRKACA	protein kinase, cAMP-dependent, catalytic, α	<i>Oztan G et al, 2024</i>	down
RNF149; SNORD89	ring finger protein 149; small nucleolar RNA, C/D box 89	<i>Oztan G et al, 2024</i>	up
RPL27A; SNORA3A	ribosomal protein L27a; small nucleolar RNA, H/ACA box 3A	<i>Oztan G et al, 2024</i>	up
RPL32	ribosomal protein L32	<i>Oztan G et al, 2024</i>	down
RPL9	ribosomal protein L9	<i>Oztan G et al, 2024</i>	down
SNORA54	small nucleolar RNA, H/ACA box 54	<i>Oztan G et al, 2024</i>	up
TCIRG1	T-cell, immune regulator 1, ATPase, H⁺ transporting, lysosomal V0 subunit A3	<i>Oztan G et al, 2024</i>	down
TOP2B	topoisomerase (DNA) II β	<i>Oztan G et al, 2024</i>	down