Supplementary Materials: **A homozygous deep intronic variant causing von Willebrand factor deficiency and lack of endothelial-specific secretory organelles, Weible-Palade bodies**

Table S1. List of homozygous variants located in entire von Willebrand factor (VWF) gene of the index patient, detected by next-generation sequencing, and subsequent bioinformatics analysis by Ensemble Variant Effect Predictor (VEP)

	Location	Allele	Consequence	SYMBOL	Existing variation	SIFT	PolyPhen	CLIN_SIG	SpliceAI_pred DS_AG	SpliceAI_pred DS_AL	SpliceAI_pred DS_DG	SpliceAI_pred DS_DL
1	NC_000012:6121122-6121122	G	intron_variant	VWF	rs6489694	-	-	-	0.00	0.00	0.00	0.00
2	NC_000012:6116076-6116076	G	intron_variant	VWF	rs4764537	-	-	-	0.00	0.00	0.00	0.00
3	NC_000012:6107670-6107670	T	intron_variant	VWF	rs4485164	-	-	-	0.00	0.00	0.00	0.00
4	NC_000012:6095736-6095737	-	intron_variant	VWF	rs5796209	-	-	-	0.00	0.00	0.00	0.00
5	NC_000012:6093442-6093442	A	intron_variant	VWF	rs2058223	-	-	-	0.00	0.00	0.00	0.00
6	NC_000012:6093296-6093296	G	intron_variant	VWF	rs12317079	-	-	-	0.00	0.00	0.00	0.00
7	NC_000012:6092634-6092634	A	intron_variant	VWF	rs142695490	-	-	-	0.00	0.00	0.00	0.00
8	NC_000012:6092632-6092632	A	intron_variant	VWF	rs148892733	-	-	-	0.00	0.00	0.00	0.00
9	NC_000012:6092630-6092630	A	intron_variant	VWF	rs143663161	-	-	-	0.00	0.00	0.00	0.00
10	NC_000012:6092628-6092628	A	intron_variant	VWF	rs144959320	-	-	-	0.00	0.00	0.00	0.00
11	NC_000012:6092626-6092626	A	intron_variant	VWF	rs150759772	-	-	-	0.00	0.00	0.00	0.00
12	NC_000012:6092624-6092624	A	intron_variant	VWF	rs144868844	-	-	-	0.00	0.00	0.00	0.00
13	NC_000012:6092622-6092622	A	intron_variant	VWF	rs145245519	-	-	-	0.00	0.00	0.00	0.00
14	NC_000012:6092620-6092620	A	intron_variant	VWF	rs147129737	-	-	-	0.00	0.00	0.00	0.00
15	NC_000012:6092519-6092519	G	intron_variant	VWF	rs3063460	-	-	-	0.00	0.00	0.00	0.00
16	NC_000012:6092515-6092515	TG	intron_variant	VWF	rs66551998	-	-	-	-	-	-	-
17	NC_000012:6092028-6092028	G	intron_variant	VWF	rs2239139	-	-	-	0.00	0.00	0.00	0.00
18	NC_000012:6091277-6091277	T	intron_variant	VWF	rs2239140	-	-	-	0.00	0.00	0.00	0.00
19	NC_000012:6089912-6089912	C	intron_variant	VWF	rs2239142	-	-	-	0.00	0.00	0.00	0.00
20	NC_000012:6089774-6089774	G	intron_variant	VWF	rs1034933	-	-	-	0.00	0.00	0.00	0.00
21	NC_000012:6089557-6089557	T	intron_variant	VWF	rs2109119	-	-	-	0.00	0.00	0.00	0.00

22	NC_000012:6089081-6089081	T	intron_variant	VWF	rs10849382	-	-	-	0.00	0.00	0.00	0.00
23	NC_000012:6089075-6089075	T	intron_variant	VWF	rs10849381	-	-	-	0.00	0.00	0.00	0.00
24	NC_000012:6089065-6089065	G	intron_variant, non_coding transcript_variant	VWF	rs2238101	-	-	-	0.00	0.00	0.00	0.00
25	NC_000012:6088302-6088302	C	intron_variant	VWF	rs2238102	-	-	-	0.00	0.00	0.00	0.00
26	NC_000012:6085489-6085489	C	intron_variant	VWF	rs2109118	-	-	-	0.00	0.00	0.00	0.00
27	NC_000012:6085077-6085077	G	intron_variant	VWF	rs1860364	-	-	-	0.00	0.00	0.00	0.00
28	NC_000012:6085017-6085017	A	intron_variant	VWF	rs2238103	-	-	-	0.00	0.00	0.01	0.00
29	NC_000012:6084371-6084371	G	intron_variant	VWF	rs763580	-	-	-	0.00	0.00	0.00	0.00
30	NC_000012:6083930-6083930	T	intron_variant	VWF	rs3819537	-	-	-	0.00	0.00	0.00	0.00
31	NC_000012:6083720-6083720	G	intron_variant	VWF	rs3819538	-	-	-	0.00	0.00	0.00	0.00
32	NC_000012:6081339-6081339	T	intron_variant	VWF	rs7133854	-	-	-	0.00	0.00	0.00	0.00
33	NC_000012:6073501-6073501	C	intron_variant	VWF	-	-	-	-	0.00	0.00	**0.95**	0.00
34	NC_000012:6070224-6070224	T	intron_variant	VWF	rs137873960	-	-	-	0.00	0.00	0.00	0.00
35	NC_000012:6066859-6066859	C	intron_variant	VWF	rs2238106	-	-	-	0.00	0.00	0.00	0.00
36	NC_000012:6063182-6063182	A	intron_variant	VWF	rs2239159	-	-	-	0.00	0.00	0.00	0.00
37	NC_000012:6062336-6062336	G	intron_variant	VWF	rs2191161	-	-	-	0.00	0.00	0.00	0.00
38	NC_000012:6060567-6060567	C	intron_variant	VWF	rs4764482	-	-	-	0.01	0.00	0.01	0.00
39	NC_000012:6060277-6060277	A	intron_variant	VWF	rs980133	-	-	-	0.00	0.00	0.00	0.00
40	NC_000012:6060032-6060032	C	intron_variant	VWF	rs980131	-	-	-	0.00	0.00	0.00	0.00
41	NC_000012:6059861-6059861	C	intron_variant	VWF	rs980130	-	-	-	0.00	0.00	0.00	0.00
42	NC_000012:6054787-6054787	A	intron_variant	VWF	rs36100099	-	-	-	0.01	0.00	0.00	0.00
43	NC_000012:6053686-6053686	C	intron_variant	VWF	rs2283333	-	-	-	0.00	0.00	0.00	0.00
44	NC_000012:6053588-6053588	G	intron_variant	VWF	rs2239160	-	-	-	0.00	0.00	0.00	0.00
45	NC_000012:6053495-6053495	A	intron_variant	VWF	rs2239161	-	-	-	0.00	0.00	0.00	0.00
46	NC_000012:6051577-6051578	TG	intron_variant	VWF	rs386759880	-	-	-	-	-	-	-
47	NC_000012:6051526-6051526	A	intron_variant	VWF	rs4764531	-	-	-	0.00	0.00	0.00	0.00
48	NC_000012:6051039-6051039	Ins.	intron_variant	VWF	rs75336349	-	-	-	-	-	-	-
49	NC_000012:6050980-6050980	C	intron_variant	VWF	rs11064010	-	-	-	0.00	0.00	0.00	0.00
50	NC_000012:6050879-6050879	T	intron_variant	VWF	rs10849379	-	-	-	0.01	0.00	0.00	0.00

51	NC_000012:6050371-6050371	G	intron_variant	VWF	rs216304	-	-	-	0.00	0.00	0.00	0.00
52	NC_000012:6049879-6049879	T	intron_variant	VWF	rs216303	-	-	-	0.00	0.00	0.00	0.00
53	NC_000012:6048856-6048856	A	intron_variant	VWF	rs216302	-	-	-	0.00	0.00	0.00	0.00
54	NC_000012:6048822-6048822	G	intron_variant	VWF	rs216301	-	-	-	0.00	0.00	0.00	0.00
55	NC_000012:6047469-6047469	G	intron_variant	VWF	rs216299	-	-	-	0.00	0.00	0.00	0.00
56	NC_000012:6047311-6047311	T	intron_variant	VWF	rs216298	-	-	-	0.00	0.00	0.00	0.00
57	NC_000012:6045504-6045504	G	intron_variant	VWF	rs216296	-	-	-	0.00	0.00	0.00	0.00
58	NC_000012:6045040-6045040	C	intron_variant	VWF	rs216295	-	-	-	0.00	0.00	0.00	0.00
59	NC_000012:6044584-6044584	G	intron_variant	VWF	rs216294	-	-	-	0.00	0.00	0.00	0.00
60	NC_000012:6044493-6044493	T	intron_variant	VWF	rs216293	-	-	benign	0.01	0.00	0.00	0.00
61	NC_000012:6044098-6044098	G	intron_variant	VWF	rs11064008	-	-	-	0.00	0.00	0.00	0.00
62	NC_000012:6043810-6043810	A	intron_variant	VWF	rs216292	-	-	-	0.00	0.00	0.00	0.00
63	NC_000012:6043683-6043683	G	intron_variant	VWF	rs73034873	-	-	-	0.00	0.00	0.00	0.00
64	NC_000012:6042707-6042707	T	intron_variant	VWF	rs216291	-	-	-	0.00	0.00	0.00	0.00
65	NC_000012:6042431-6042431	A	intron_variant	VWF	rs216290	-	-	-	0.00	0.00	0.00	0.00
66	NC_000012:6041658-6041658	A	intron_variant	VWF	rs183356	-	-	-	0.00	0.00	0.00	0.00
67	NC_000012:6040118-6040118	C	intron_variant	VWF	rs216339	-	-	-	0.00	0.00	0.00	0.00
68	NC_000012:6039160-6039160	G	intron_variant	VWF	rs216338	-	-	-	0.00	0.00	0.00	0.00
69	NC_000012:6039143-6039143	C	intron_variant	VWF	rs216337	-	-	-	0.00	0.00	0.00	0.00
70	NC_000012:6039064-6039064	G	intron_variant	VWF	rs216336	-	-	-	0.00	0.00	0.00	0.00
71	NC_000012:6038839-6038839	C	intron_variant	VWF	rs216335	-	-	-	0.00	0.00	0.00	0.00
72	NC_000012:6038445-6038445	T	intron_variant	VWF	rs216334	-	-	-	0.00	0.00	0.00	0.00
73	NC_000012:6037953-6037953	C	intron_variant	VWF	rs216333	-	-	-	0.00	0.00	0.00	0.00
74	NC_000012:6037890-6037890	A	intron_variant	VWF	rs216332	-	-	-	0.00	0.00	0.00	0.00
75	NC_000012:6037848-6037848	T	intron_variant	VWF	rs216331	-	-	-	0.00	0.00	0.00	0.00
76	NC_000012:6037464-6037464	C	intron_variant	VWF	rs216330	-	-	-	0.00	0.00	0.01	0.00
77	NC_000012:6037341-6037341	C	intron_variant	VWF	rs216329	-	-	-	0.00	0.00	0.02	0.00
78	NC_000012:6037317-6037317	T	intron_variant	VWF	rs216328	-	-	-	0.00	0.00	0.00	0.00
79	NC_000012:6036983-6036983	C	intron_variant	VWF	rs216327	-	-	-	0.00	0.00	0.00	0.00

80	NC_000012:6036782-6036782	A	intron_variant	VWF	rs216326	-	-	-	0.00	0.00	0.00	0.00
81	NC_000012:6036363-6036363	A	intron_variant	VWF	rs216325	-	-	benign	0.00	0.00	0.00	0.00
82	NC_000012:6036059-6036059	G	intron_variant	VWF	rs216324	-	-	-	0.00	0.00	0.00	0.00
83	NC_000012:6035943-6035943	T	intron_variant	VWF	rs216323	-	-	-	0.00	0.00	0.00	0.00
84	NC_000012:6035731-6035731	T	intron_variant	VWF	rs216322	-	-	-	0.00	0.00	0.00	0.00
85	NC_000012:6035003-6035003	G	intron_variant	VWF	rs669667	-	-	-	0.00	0.00	0.00	0.00
86	NC_000012:6034998-6034998	C	intron_variant	VWF	rs499648	-	-	-	0.00	0.00	0.00	0.00
87	NC_000012:6034818-6034818	C	missense_variant	VWF	rs216321	Tolerated (1)	Benign (0)	benign	0.00	0.00	0.00	0.00
88	NC_000012:6034775-6034775	T	synonymous_variant	VWF	rs756662315	-	-	-	0.00	0.00	0.00	0.00
89	NC_000012:6034190-6034190	C	intron_variant	VWF	rs216320	-	-	-	0.00	0.00	0.01	0.00
90	NC_000012:6032875-6032875	AC	intron_variant	VWF	rs796891068	-	-	-	-	-	-	-
91	NC_000012:6030095-6030095	C	intron_variant	VWF	rs216318	-	-	-	0.00	0.00	0.00	0.00
92	NC_000012:6027748-6027748	G	intron_variant	VWF	rs216315	-	-	-	0.00	0.00	0.00	0.00
93	NC_000012:6027414-6027414	C	intron_variant	VWF	rs216314	-	-	-	0.00	0.00	0.00	0.00
94	NC_000012:6019277-6019277	C	missense_variant	VWF	rs216311	Tolerated (1)	Benign (0)	Benign, likely_benign	0.00	0.00	0.00	0.00
95	NC_000012:6018777-6018777	G	synonymous_variant	VWF	rs216310	-	-	likely_benign, benign	0.00	0.00	0.00	0.00
96	NC_000012:6017761-6017761	T	intron_variant	VWF	rs216309	-	-	-	0.00	0.00	0.00	0.00
97	NC_000012:6015500-6015500	C	intron_variant	VWF	rs216308	-	-	-	0.00	0.00	0.00	0.00
98	NC_000012:6011912-6011912	T	intron_variant	VWF	rs216305	-	-	-	0.01	0.00	0.00	0.00
99	NC_000012:6009094-6009094	T	intron_variant	VWF	rs216814	-	-	-	0.00	0.00	0.00	0.00
100	NC_000012:6008617-6008617	G	intron_variant	VWF	rs216813	-	-	-	0.00	0.00	0.00	0.00
101	NC_000012:6005478-6005478	C	intron_variant	VWF	rs216810	-	-	-	0.00	0.00	0.00	0.00
102	NC_000012:6004278-6004278	C	intron_variant	VWF	rs216808	-	-	-	0.00	0.00	0.00	0.00
103	NC_000012:6003290-6003290	A	intron_variant	VWF	rs216807	-	-	-	0.00	0.00	0.00	0.00
104	NC_000012:6002359-6002359	G	intron_variant	VWF	rs216806	-	-	-	0.00	0.00	0.00	0.00
105	NC_000012:6002245-6002245	A	intron_variant	VWF	rs216804	-	-	-	0.00	0.00	0.00	0.00
106	NC_000012:6000796-6000796	T	intron_variant	VWF	rs188819154	-	-	-	0.00	0.00	0.00	0.00
107	NC_000012:6000790-6000792	-	intron_variant	VWF	rs149367153	-	-	-	0.00	0.00	0.00	0.00
108	NC_000012:6000736-6000736	C	intron_variant	VWF	rs216803	-	-	-	0.00	0.00	0.00	0.00

109	NC_000012:5999937-5999937	A	intron_variant	VWF	rs216802	-	-	-	0.00	0.00	0.00	0.00
110	NC_000012:5999347-5999347	A	intron_variant	VWF	rs216801	-	-	-	0.00	0.00	0.00	0.00
111	NC_000012:5998780-5998780	T	intron_variant	VWF	rs216800	-	-	-	0.00	0.00	0.00	0.00
112	NC_000012:5998451-5998451	C	intron_variant	VWF	rs7398722	-	-	-	0.00	0.00	0.00	0.00
113	NC_000012:5996852-5996852	C	intron_variant	VWF	rs216904	-	-	-	0.00	0.00	0.00	0.00
114	NC_000012:5996333-5996333	C	intron_variant	VWF	rs216903	-	-	-	0.00	0.00	0.00	0.00
115	NC_000012:5996221-5996221	A	synonymous_variant	VWF	rs216902	-	-	likely_benign, benign	0.00	0.00	0.00	0.00
116	NC_000012:5995702-5995702	A	intron_variant	VWF	rs216901	-	-	-	0.00	0.00	0.00	0.00
117	NC_000012:5994832-5994832	T	intron_variant	VWF	rs12099542	-	-	-	0.00	0.00	0.00	0.00
118	NC_000012:5993598-5993598	A	intron_variant	VWF	rs216898	-	-	-	0.00	0.00	0.00	0.00
119	NC_000012:5993441-5993441	C	intron_variant	VWF	rs216897	-	-	-	0.00	0.00	0.00	0.00
120	NC_000012:5991630-5991630	T	intron_variant	VWF	rs216896	-	-	-	0.00	0.00	0.00	0.00
121	NC_000012:5991429-5991429	T	intron_variant	VWF	rs216895	-	-	-	0.00	0.00	0.00	0.00
122	NC_000012:5991411-5991411	G	intron_variant	VWF	rs216894	-	-	-	0.00	0.00	0.00	0.00
123	NC_000012:5991319-5991319	G	intron_variant	VWF	rs216893	-	-	-	0.00	0.00	0.00	0.00
124	NC_000012:5990909-5990909	A	intron_variant	VWF	rs4348960	-	-	-	0.01	0.00	0.00	0.00
125	NC_000012:5990862-5990862	A	intron_variant	VWF	rs216892	-	-	-	0.00	0.00	0.00	0.00
126	NC_000012:5990683-5990683	G	intron_variant	VWF	rs216891	-	-	-	0.01	0.00	0.00	0.00
127	NC_000012:5990372-5990372	C	intron_variant	VWF	rs216890	-	-	-	0.00	0.00	0.00	0.00
128	NC_000012:5990287-5990287	C	intron_variant	VWF	rs216889	-	-	-	0.00	0.00	0.00	0.00
129	NC_000012:5989691-5989691	G	intron_variant	VWF	rs216888	-	-	-	0.00	0.00	0.00	0.00
130	NC_000012:5989286-5989286	C	intron_variant	VWF	rs216887	-	-	-	0.00	0.00	0.00	0.00
131	NC_000012:5988799-5988799	T	intron_variant	VWF	rs216886	-	-	-	0.00	0.00	0.00	0.00
132	NC_000012:5988789-5988789	C	intron_variant	VWF	rs216885	-	-	-	0.00	0.00	0.00	0.00
133	NC_000012:5988662-5988662	A	intron_variant	VWF	rs216884	-	-	-	0.00	0.00	0.00	0.00
134	NC_000012:5987650-5987650	T	intron_variant	VWF	rs216882	-	-	-	0.00	0.00	0.00	0.00
135	NC_000012:5987546-5987546	G	intron_variant	VWF	rs216881	-	-	-	0.00	0.00	0.00	0.00
136	NC_000012:5987525-5987525	A	intron_variant	VWF	rs216880	-	-	-	0.00	0.00	0.00	0.00
137	NC_000012:5987510-5987510	A	intron_variant	VWF	rs526881	-	-	-	0.00	0.00	0.00	0.00

138	NC_000012:5987507-5987507	G	intron_variant	VWF	rs526861	-	-	-	0.00	0.00	0.00	0.00
139	NC_000012:5987489-5987489	T	intron_variant	VWF	rs216879	-	-	-	0.00	0.00	0.00	0.00
140	NC_000012:5987217-5987217	C	intron_variant	VWF	rs216878	-	-	-	0.00	0.00	0.00	0.00
141	NC_000012:5987128-5987128	C	intron_variant	VWF	rs216877	-	-	-	0.00	0.00	0.00	0.00
142	NC_000012:5987090-5987090	A	intron_variant	VWF	rs216876	-	-	-	0.00	0.00	0.00	0.00
143	NC_000012:5986735-5986735	G	intron_variant	VWF	rs216875	-	-	-	0.00	0.00	0.00	0.00
144	NC_000012:5986641-5986642	TT	intron_variant	VWF	rs71450640	-	-	-	-	-	-	-
145	NC_000012:5986591-5986591	C	intron_variant	VWF	rs216874	-	-	-	0.00	0.00	0.00	0.00
146	NC_000012:5986108-5986108	C	intron_variant	VWF	rs216873	-	-	-	0.00	0.00	0.00	0.00
147	NC_000012:5985679-5985679	A	intron_variant	VWF	rs177702	-	-	benign	0.00	0.00	0.00	0.00
148	NC_000012:5984916-5984916	A	intron_variant	VWF	rs24177	-	-	-	0.00	0.00	0.00	0.00
149	NC_000012:5984374-5984374	C	intron_variant	VWF	rs216872	-	-	-	0.00	0.00	0.00	0.00
150	NC_000012:5983882-5983882	A	intron_variant	VWF	rs216870	-	-	-	0.00	0.00	0.00	0.00
151	NC_000012:5982531-5982531	C	intron_variant	VWF	rs190156617	-	-	-	0.00	0.00	0.02	0.00
152	NC_000012:5981998-5981998	A	splice_region_variant ,intron_variant	VWF	rs216868	-	-	benign	0.00	0.00	0.00	0.00
153	NC_000012:5981834-5981834	G	synonymous_variant	VWF	rs216867	-	-	likely_benign, benign	0.00	0.00	0.00	0.00
154	NC_000012:5981137-5981137	C	intron_variant	VWF	rs216865	-	-	-	0.00	0.00	0.00	0.00
155	NC_000012:5978365-5978365	C	intron_variant	VWF	rs216859	-	-	-	0.00	0.00	0.00	0.00
156	NC_000012:5977875-5977875	A	intron_variant	VWF	rs216858	-	-	-	0.00	0.00	0.00	0.00
157	NC_000012:5977770-5977770	T	intron_variant	VWF	rs216857	-	-	-	0.00	0.00	0.00	0.00
158	NC_000012:5977099-5977099	C	intron_variant	VWF	rs216856	-	-	-	0.00	0.00	0.00	0.00
159	NC_000012:5976347-5976347	G	intron_variant	VWF	rs216855	-	-	-	0.00	0.00	0.00	0.00
160	NC_000012:5975698-5975698	C	intron_variant	VWF	rs216854	-	-	-	0.00	0.00	0.00	0.00
161	NC_000012:5975547-5975547	A	intron_variant	VWF	rs216853	-	-	-	0.00	0.00	0.00	0.00
162	NC_000012:5975157-5975157	G	intron_variant	VWF	rs4764521	-	-	-	0.00	0.00	0.00	0.00
163	NC_000012:5975025-5975025	C	intron_variant	VWF	rs216852	-	-	-	0.00	0.00	0.00	0.00
164	NC_000012:5973633-5973633	G	intron_variant	VWF	rs12829271	-	-	-	0.00	0.00	0.00	0.00
165	NC_000012:5973586-5973586	G	intron_variant	VWF	rs10849366	-	-	-	0.00	0.00	0.00	0.00
166	NC_000012:5973392-5973392	T	intron_variant	VWF	rs2239138	-	-	-	0.00	0.00	0.00	0.00

167	NC_000012:5972947-5972947	G	intron_variant	VWF	rs917859	-	-	-	0.00	0.00	0.00	0.00
168	NC_000012:5972772-5972772	T	intron_variant	VWF	rs917858	-	-	-	0.00	0.00	0.00	0.00
169	NC_000012:5971429-5971429	A	intron_variant	VWF	rs11063965	-	-	-	0.00	0.00	0.00	0.00
170	NC_000012:5971411-5971411	C	intron_variant	VWF	rs2238110	-	-	-	0.00	0.00	0.00	0.00
171	NC_000012:5971368-5971368	C	intron_variant	VWF	rs10849365	-	-	-	0.00	0.00	0.00	0.00
172	NC_000012:5971124-5971124	A	intron_variant	VWF	rs7301070	-	-	-	0.02	0.00	0.00	0.00
173	NC_000012:5970158-5970158	T	intron_variant	VWF	rs60982029	-	-	-	0.00	0.00	0.00	0.00
174	NC_000012:5969892-5969892	T	intron_variant	VWF	rs12300917	-	-	-	0.00	0.00	0.00	0.00
175	NC_000012:5969772-5969772	G	intron_variant	VWF	rs11063964	-	-	-	0.00	0.00	0.00	0.00
176	NC_000012:5969450-5969450	G	intron_variant	VWF	rs2270239	-	-	-	0.00	0.00	0.00	0.00
177	NC_000012:5969258-5969258	T	missense_variant	VWF	rs35335161	Tolerated (0.27)	Benign (0.021)	Benign, likely_benign	0.00	0.00	0.00	0.00
178	NC_000012:5968959-5968959	A	intron_variant	VWF	rs4764478	-	-	-	0.00	0.00	0.00	0.00
179	NC_000012:5968741-5968741	A	intron_variant	VWF	rs769080	-	-	-	0.00	0.00	0.00	0.00
180	NC_000012:5968688-5968688	C	intron_variant	VWF	rs2012829	-	-	-	0.00	0.00	0.00	0.00
181	NC_000012:5968656-5968656	C	intron_variant	VWF	rs12307110	-	-	-	0.00	0.00	0.00	0.00
182	NC_000012:5967775-5967775	G	intron_variant	VWF	rs216851	-	-	-	0.00	0.00	0.02	0.00
183	NC_000012:5965306-5965306	A	intron_variant	VWF	rs73049469	-	-	-	0.00	0.00	0.00	0.00
184	NC_000012:5964354-5964354	G	intron_variant	VWF	rs12369177	-	-	-	0.00	0.00	0.00	0.00
185	NC_000012:5964017-5964017	C	intron_variant	VWF	rs12307884	-	-	-	0.00	0.00	0.00	0.00
186	NC_000012:5964012-5964012	C	intron_variant	VWF	rs12307883	-	-	-	0.00	0.00	0.00	0.00
187	NC_000012:5962770-5962770	C	intron_variant	VWF	rs11063957	-	-	-	0.00	0.00	0.00	0.00
188	NC_000012:5962637-5962645	-	intron_variant	VWF	rs60444970	-	-	-	-	-	-	-
189	NC_000012:5961673-5961673	A	intron_variant	VWF	rs2363309	-	-	-	0.00	0.00	0.00	0.00
190	NC_000012:5958468-5958468	A	intron_variant	VWF	rs11063956	-	-	-	0.00	0.00	0.00	0.00
191	NC_000012:5958348-5958348	C	intron_variant	VWF	rs11063955	-	-	-	0.00	0.00	0.00	0.00
192	NC_000012:5957986-5957986	T	intron_variant	VWF	rs7964554	-	-	-	0.00	0.00	0.00	0.00
193	NC_000012:5957520-5957520	T	intron_variant	VWF	rs35926634	-	-	-	0.00	0.00	0.00	0.00
194	NC_000012:5957191-5957191	G	intron_variant	VWF	rs10849363	-	-	-	0.00	0.00	0.00	0.00
195	NC_000012:5955904-5955904	G	intron_variant	VWF	rs11063953	-	-	-	0.00	0.00	0.00	0.00

196	NC_000012:5955526-5955526	A	intron_variant	VWF	rs12819922	-	-	-	0.00	0.00	0.00	0.00
197	NC_000012:5954793-5954793	T	intron_variant	VWF	rs723189	-	-	-	0.00	0.00	0.00	0.00
198	NC_000012:5953976-5953976	T	intron_variant	VWF	rs12317523	-	-	-	0.00	0.00	0.00	0.00
199	NC_000012:5953817-5953817	A	intron_variant	VWF	rs7958883	-	-	-	0.00	0.00	0.00	0.00
200	NC_000012:5953723-5953723	G	intron_variant	VWF	rs12297442	-	-	-	0.00	0.00	0.00	0.00
201	NC_000012:5953712-5953712	A	intron_variant	VWF	rs3759321	-	-	-	0.02	0.00	0.00	0.00
			non_coding_transcript_variant									
202	NC_000012:5953687-5953687	T	intron_variant	VWF	rs3759320	-	-	-	0.00	0.00	0.00	0.00
			non_coding_transcript_variant									
203	NC_000012:5953213-5953213	G	intron_variant	VWF	rs12831864	-	-	-	0.00	0.00	0.00	0.00
			non_coding_transcript_variant									
204	NC_000012:5952781-5952781	C	intron_variant	VWF	rs7133777	-	-	-	0.00	0.00	0.01	0.00
			non_coding_transcript_variant									
205	NC_000012:5948745-5948745	G	upstream_gene_variant	ANO2	rs1990326	-	-	-	-	-	-	-
206	NC_000012:5947502-5947502	C	upstream_gene_variant	ANO2	rs11063951	-	-	-	-	-	-	-
207	NC_000012:5947326-5947326	C	upstream_gene_variant	ANO2	rs3809242	-	-	-	-	-	-	-
208	NC_000012:5947284-5947284	T	upstream_gene_variant	ANO2	rs3809241	-	-	-	-	-	-	-
209	NC_000012:5946627-5946630	-	upstream_gene_variant	ANO2	rs3842300	-	-	-	-	-	-	-
210	NC_000012:5945966-5945966	G	downstream_gene_variant	VWF	rs6489682	-	-	-	0.00	0.00	0.00	0.00
			Intron-Variant	ANO2								
211	NC_000012:5945874-5945874	T	downstream_gene_variant	VWF	rs6416322	-	-	-	0.00	0.00	0.00	0.00
			Intron Variant	ANO2								
212	NC_000012:5945582-5945582	C	downstream_gene_variant	VWF	rs6489681	-	-	-	0.00	0.00	0.01	0.00
			Intron Variant	ANO2								
213	NC_000012:5945229-5945229	G	downstream_gene_variant	VWF	rs6489680	-	-	-	0.00	0.00	0.00	0.00
			Intron Variant	ANO2								
214	NC_000012:5945034-5945034	A	downstream_gene_variant	VWF	rs117766816	-	-	-	0.00	0.00	0.00	0.00
			intron_variant	ANO2								0
215	NC_000012:5943702-5943702	T	intron_variant	ANO2	rs7314566	-	-	-	0.00	0.00	0.00	0.00

Table S1 presents a complete list of homozygous variants identified in VWF gene (*VWF*; chr12:5,948,877-6,124,770 [GRCh38/hg38]) after NGS and subsequent bioinformatics analysis by VEP, including variant annotation, variant location, the existing variant in public variant databases (SNPdb, SNPs database) and predicting its pathogenic effect. The impact of exonic variants was evaluated by SIFT and PolyPhen prediction tools, as well as splicing prediction tool SpliceAl. The intronic variants were only validated by Splice Al (Illumina artificial intelligence splicing prediction software). **SIFT** predicts whether an amino acid substitution affects protein function based on sequence homology and the physical properties of amino acids. PolyPhen is a tool that predicts the possible impact of an amino acid substitution on the structure and function of a human protein using straightforward physical and comparative considerations. For both SIFT and PolyPhen, VEP can report either a score between 0 and 1, a prediction in words, or both. SpliceAl calculations are including **DS_AG (**Delta score for acceptor gain), **DS_AL (**Delta score for acceptor loss), **DS_DG** (Delta score for donor gain), and **DS_DL** (Delta score for donor loss). The calculated scores range from 0 to 1 and can be interpreted as the probability of the variant being splice-altering. The suggested cutoffs are: 0.2 (high recall), 0.5 (recommended), and 0.8 (high precision)**.**

Table S2. Allele frequency data for homozygous variants located in von Willebrand factor (VWF) gene of the index patient, from genotyping project, the gnomAD

	Location	SNPdb ID	Allele	SYMBOL	AFR_AF	AMR_AF	EAS_AF	EUR_AF	SAS_AF
1	NC_000012:6121122-6121122	rs6489694	G	VWF	1	0.9957	1	0.9841	0.9847
2	NC_000012:6116076-6116076	rs4764537	G	VWF	1	0.9957	1	0.9841	0.9857
3	NC_000012:6107670-6107670	rs4485164	T	VWF	0.7791	0.5519	0.6349	0.7376	0.6892
4	NC_000012:6095736-6095737	rs5796209	-	VWF	0.2141	0.5259	0.7222	0.66	0.7362
5	NC_000012:6093442-6093442	rs2058223	A	VWF	0.4773	0.5793	0.7113	0.6968	0.7761
6	NC_000012:6093296-6093296	rs12317079	G	VWF	0.1929	0.5144	0.7113	0.6282	0.7219
7	NC_000012:6092634-6092634	rs142695490	A	VWF	-	-	-	-	-
8	NC_000012:6092632-6092632	rs148892733	A	VWF	-	-	-	-	-
9	NC_000012:6092630-6092630	rs143663161	A	VWF	-	-	-	-	-
10	NC_000012:6092628-6092628	rs144959320	A	VWF	-	-	-	-	-
11	NC_000012:6092626-6092626	rs150759772	A	VWF	-	-	-	-	-
12	NC_000012:6092624-6092624	rs144868844	A	VWF	-	-	-	-	-
13	NC_000012:6092622-6092622	rs145245519	A	VWF	-	-	-	-	-
14	NC_000012:6092620-6092620	rs147129737	A	VWF	-	-	-	-	-
15	NC_000012:6092519-6092519	rs3063460	G	VWF	-	-	-	-	-
16	NC_000012:6092515-6092515	rs66551998	TG	VWF	-	-	-	-	-
17	NC_000012:6092028-6092028	rs2239139	G	VWF	0.9395	0.6383	0.753	0.6998	0.7822
18	NC_000012:6091277-6091277	rs2239140	T	VWF	0.0356	0.4207	0.3026	0.495	0.3517
19	NC_000012:6089912-6089912	rs2239142	C	VWF	0.0363	0.428	0.2867	0.5	0.3609
20	NC_000012:6089774-6089774	rs1034933	G	VWF	0.9705	0.6326	0.7827	0.6988	0.7883
21	NC_000012:6089557-6089557	rs2109119	T	VWF	0.4236	0.5807	0.753	0.6968	0.7863
22	NC_000012:6089081-6089081	rs10849382	T	VWF	0.4682	0.5879	0.7897	0.6958	0.7883
23	NC_000012:6089075-6089075	rs10849381	T	VWF	0.4682	0.5879	0.7897	0.6958	0.7904
24	NC_000012:6089065-6089065	rs2238101	G	VWF	0.4168	0.5865	0.7609	0.6948	0.7863
25	NC_000012:6088302-6088302	rs2238102	C	VWF	0.9735	0.647	0.7867	0.7018	0.7914
26	NC_000012:6085489-6085489	rs2109118	C	VWF	0.0386	0.451	0.2857	0.498	0.3333
27	NC_000012:6085077-6085077	rs1860364	G	VWF	0.9599	0.6614	0.7946	0.7048	0.7658
28	NC_000012:6085017-6085017	rs2238103	A	VWF	0.7496	0.6066	0.7917	0.6362	0.6963
29	NC_000012:6084371-6084371	rs763580	G	VWF	0.9599	0.6484	0.7956	0.7008	0.7658
30	NC_000012:6083930-6083930	rs3819537	T	VWF	0.9599	0.6484	0.7956	0.6998	0.7658
31	NC_000012:6083720-6083720	rs3819538	G	VWF	0.9599	0.6484	0.7956	0.7008	0.7658
32	NC_000012:6081339-6081339	rs7133854	T	VWF	0.6067	0.5331	0.2897	0.5616	0.4039
34	NC_000012:6070224-6070224	rs137873960	T	VWF	0.0023	0.0029	0	0.0159	0
35	NC_000012:6066859-6066859	rs2238106	C	VWF	0.27	0.4323	0.6339	0.6163	0.6043
36	NC_000012:6063182-6063182	rs2239159	A	VWF	0.3964	0.245	0.4306	0.3837	0.5348
37	NC_000012:6062336-6062336	rs2191161	G	VWF	0.5234	0.5432	0.6647	0.7425	0.6902
38	NC_000012:6060567-6060567	rs4764482	C	VWF	0.1346	0.611	0.6915	0.5278	0.6074
39	NC_000012:6060277-6060277	rs980133	A	VWF	0.6921	0.7205	0.7222	0.6123	0.7249
40	NC_000012:6060032-6060032	rs980131	C	VWF	0.6921	0.7176	0.7242	0.6123	0.7249
41	NC_000012:6059861-6059861	rs980130	C	VWF	0.7738	0.7695	0.8879	0.6829	0.7955
42	NC_000012:6054787-6054787	rs36100099	A	VWF	0.3154	0.621	0.6845	0.493	0.6033
43	NC_000012:6053686-6053686	rs2283333	C	VWF	0.8964	0.8905	0.7679	0.8887	0.8783
44	NC_000012:6053588-6053588	rs2239160	G	VWF	0.8533	0.8948	0.7669	0.8887	0.8783
45	NC_000012:6053495-6053495	rs2239161	A	VWF	0.9599	0.902	0.7649	0.8887	0.8773

46	NC_000012:6051577-6051578	rs386759880	TG	VWF	-	-	-	-	-
47	NC_000012:6051526-6051526	rs4764531	A	VWF	0.3359	0.6542	0.6796	0.5338	0.6861
48	NC_000012:6051039-6051039	rs75336349	Ins.	VWF	0.3933	0.6657	0.6806	0.5517	0.681
49	NC_000012:6050980-6050980	rs11064010	C	VWF	0.6679	0.8602	0.7708	0.8767	0.863
50	NC_000012:6050879-6050879	rs10849379	T	VWF	0.382	0.6686	0.6825	0.5537	0.6851
51	NC_000012:6050371-6050371	rs216304	G	VWF	0.851	0.9107	0.7738	0.9085	0.91
52	NC_000012:6049879-6049879	rs216303	T	VWF	0.8525	0.9107	0.7738	0.9095	0.9018
53	NC_000012:6048856-6048856	rs216302	A	VWF	0.8517	0.9107	0.7758	0.9095	0.9029
54	NC_000012:6048822-6048822	rs216301	G	VWF	0.8517	0.9107	0.7758	0.9095	0.9029
55	NC_000012:6047469-6047469	rs216299	G	VWF	0.8343	0.9049	0.7758	0.9095	0.9029
56	NC_000012:6047311-6047311	rs216298	T	VWF	0.8343	0.9035	0.7758	0.9085	0.9018
57	NC_000012:6045504-6045504	rs216296	G	VWF	0.9614	0.9193	0.7778	0.9095	0.9029
58	NC_000012:6045040-6045040	rs216295	C	VWF	0.8101	0.8977	0.7778	0.9095	0.9029
59	NC_000012:6044584-6044584	rs216294	G	VWF	0.9614	0.9222	0.7778	0.9095	0.9029
60	NC_000012:6044493-6044493	rs216293	T	VWF	0.2171	0.67	0.6865	0.5417	0.684
61	NC_000012:6044098-6044098	rs11064008	G	VWF	0.0023	0.0937	0	0.1521	0.0266
62	NC_000012:6043810-6043810	rs216292	A	VWF	0.1725	0.6599	0.6865	0.5427	0.684
63	NC_000012:6043683-6043683	rs73034873	G	VWF	0.0015	0.0144	0	0.0199	0.001
64	NC_000012:6042707-6042707	rs216291	T	VWF	0.1823	0.6671	0.6855	0.5437	0.7055
65	NC_000012:6042431-6042431	rs216290	A	VWF	0.7511	0.8775	0.7768	0.8757	0.864
66	NC_000012:6041658-6041658	rs183356	A	VWF	0.8533	0.915	0.7768	0.9105	0.9018
67	NC_000012:6040118-6040118	rs216339	C	VWF	0.9735	0.9323	0.7917	0.9135	0.8885
68	NC_000012:6039160-6039160	rs216338	G	VWF	0.7194	0.7219	0.7877	0.6243	0.7587
69	NC_000012:6039143-6039143	rs216337	C	VWF	0.6694	0.7147	0.7877	0.6243	0.7546
70	NC_000012:6039064-6039064	rs216336	G	VWF	0.7171	0.7205	0.7877	0.6233	0.7587
71	NC_000012:6038839-6038839	rs216335	C	VWF	0.9667	0.9265	0.7907	0.9115	0.8824
72	NC_000012:6038445-6038445	rs216334	T	VWF	0.7723	0.745	0.7877	0.6521	0.772
73	NC_000012:6037953-6037953	rs216333	C	VWF	0.7731	0.745	0.7877	0.6541	0.7751
74	NC_000012:6037890-6037890	rs216332	A	VWF	0.7776	0.745	0.7867	0.6541	0.774
75	NC_000012:6037848-6037848	rs216331	T	VWF	0.7776	0.745	0.7867	0.6541	0.773
76	NC_000012:6037464-6037464	rs216330	C	VWF	0.646	0.7421	0.7867	0.6541	0.774
77	NC_000012:6037341-6037341	rs216329	C	VWF	0.8064	0.7507	0.7877	0.6541	0.774
78	NC_000012:6037317-6037317	rs216328	T	VWF	0.8064	0.7507	0.7877	0.6541	0.773
79	NC_000012:6036983-6036983	rs216327	C	VWF	0.8071	0.7507	0.7867	0.6541	0.774
80	NC_000012:6036782-6036782	rs216326	A	VWF	0.5628	0.6744	0.7143	0.5606	0.6155
81	NC_000012:6036363-6036363	rs216325	A	VWF	0.795	0.7507	0.7808	0.6521	0.774
82	NC_000012:6036059-6036059	rs216324	G	VWF	0.9448	0.9265	0.7887	0.9115	0.8824
83	NC_000012:6035943-6035943	rs216323	T	VWF	0.6354	0.7435	0.7867	0.6551	0.774
84	NC_000012:6035731-6035731	rs216322	T	VWF	0.6369	0.7435	0.7867	0.6541	0.7832
85	NC_000012:6035003-6035003	rs669667	G	VWF	0.9448	0.9265	0.7917	0.9145	0.8845
86	NC_000012:6034998-6034998	rs499648	C	VWF	0.9448	0.9265	0.7917	0.9145	0.8845
87	NC_000012:6034818-6034818	rs216321	C	VWF	0.9584	0.9265	0.7907	0.9135	0.8855
88	NC_000012:6034775-6034775	rs756662315	T	VWF	-	-	-	-	-
89	NC_000012:6034190-6034190	rs216320	C	VWF	0.9463	0.9265	0.7907	0.9135	0.8845
90	NC_000012:6032875-6032875	rs796891068	AC	VWF	0.7148	0.7219	0.754	0.6412	0.7546
91	NC_000012:6030095-6030095	rs216318	C	VWF	0.9667	0.9265	0.7956	0.9135	0.8865
92	NC_000012:6027748-6027748	rs216315	G	VWF	0.9743	0.928	0.7956	0.9125	0.8845

93	NC_000012:6027414-6027414	rs216314	C	VWF	0.9667	0.9265	0.7917	0.9135	0.8855
94	NC_000012:6019277-6019277	rs216311	C	VWF	0.9228	0.7752	0.746	0.6014	0.7863
95	NC_000012:6018777-6018777	rs216310	G	VWF	0.9221	0.7752	0.746	0.6014	0.7924
96	NC_000012:6017761-6017761	rs216309	T	VWF	0.326	0.6556	0.6349	0.502	0.6339
97	NC_000012:6015500-6015500	rs216308	C	VWF	0.9145	0.7464	0.75	0.5895	0.7924
98	NC_000012:6011912-6011912	rs216305	T	VWF	0.7005	0.6816	0.6419	0.498	0.6452
99	NC_000012:6009094-6009094	rs216814	T	VWF	0.5696	0.6441	0.7073	0.5129	0.6022
100	NC_000012:6008617-6008617	rs216813	G	VWF	0.6044	0.6542	0.7073	0.5179	0.6094
101	NC_000012:6005478-6005478	rs216810	C	VWF	0.646	0.6542	0.7183	0.5219	0.6043
102	NC_000012:6004278-6004278	rs216808	C	VWF	0.646	0.6542	0.7173	0.5209	0.6002
103	NC_000012:6003290-6003290	rs216807	A	VWF	0.6021	0.6527	0.7173	0.5199	0.5992
104	NC_000012:6002359-6002359	rs216806	G	VWF	0.6467	0.6542	0.7163	0.5219	0.6002
105	NC_000012:6002245-6002245	rs216804	A	VWF	0.646	0.6542	0.7163	0.5209	0.6002
106	NC_000012:6000796-6000796	rs188819154	T	VWF	0.0401	0.2478	0.3651	0.0616	0.138
107	NC_000012:6000790-6000792	rs149367153	-	VWF	0.2769	0.4611	0.7212	0.3678	0.5409
108	NC_000012:6000736-6000736	rs216803	C	VWF	0.6021	0.5965	0.7173	0.5159	0.6207
109	NC_000012:5999937-5999937	rs216802	A	VWF	0.5991	0.5965	0.7262	0.5159	0.6033
110	NC_000012:5999347-5999347	rs216801	A	VWF	0.2814	0.4683	0.7262	0.3708	0.5409
111	NC_000012:5998780-5998780	rs216800	T	VWF	0.2655	0.3674	0.4821	0.2555	0.409
112	NC_000012:5998451-5998451	rs7398722	C	VWF	0.997	1	1	1	1
113	NC_000012:5996852-5996852	rs216904	C	VWF	0.2806	0.4222	0.7262	0.3688	0.545
114	NC_000012:5996333-5996333	rs216903	C	VWF	0.6029	0.5504	0.7272	0.5159	0.6104
115	NC_000012:5996221-5996221	rs216902	A	VWF	0.2784	0.4179	0.7252	0.3648	0.5399
116	NC_000012:5995702-5995702	rs216901	A	VWF	0.4009	0.5259	0.7262	0.5109	0.6053
117	NC_000012:5994832-5994832	rs12099542	T	VWF	0.0408	0.2003	0.375	0.0606	0.1411
118	NC_000012:5993598-5993598	rs216898	A	VWF	0.6437	0.6542	0.7272	0.5119	0.6084
119	NC_000012:5993441-5993441	rs216897	C	VWF	0.4017	0.6311	0.7272	0.5129	0.6104
120	NC_000012:5991630-5991630	rs216896	T	VWF	0.4062	0.5259	0.7262	0.5129	0.6043
121	NC_000012:5991429-5991429	rs216895	T	VWF	0.4092	0.5245	0.7252	0.5139	0.6104
122	NC_000012:5991411-5991411	rs216894	G	VWF	0.41	0.5836	0.7252	0.5139	0.6115
123	NC_000012:5991319-5991319	rs216893	G	VWF	0.4319	0.5245	0.7252	0.5139	0.6094
124	NC_000012:5990909-5990909	rs4348960	A	VWF	-	-	-	-	-
125	NC_000012:5990862-5990862	rs216892	A	VWF	0.3253	0.5115	0.7232	0.5139	0.6125
126	NC_000012:5990683-5990683	rs216891	G	VWF	0.326	0.5187	0.7252	0.5159	0.6309
127	NC_000012:5990372-5990372	rs216890	C	VWF	0.4319	0.5245	0.7252	0.5139	0.6115
128	NC_000012:5990287-5990287	rs216889	C	VWF	0.6346	0.5461	0.7252	0.5149	0.6104
129	NC_000012:5989691-5989691	rs216888	G	VWF	0.4327	0.5245	0.7242	0.5139	0.6115
130	NC_000012:5989286-5989286	rs216887	C	VWF	0.4327	0.5245	0.7252	0.5149	0.6115
131	NC_000012:5988799-5988799	rs216886	T	VWF	0.4289	0.5317	0.7242	0.5119	0.6094
132	NC_000012:5988789-5988789	rs216885	C	VWF	0.3722	0.4741	0.5	0.3986	0.4888
133	NC_000012:5988662-5988662	rs216884	A	VWF	0.3086	0.4568	0.4881	0.3996	0.4775
134	NC_000012:5987650-5987650	rs216882	T	VWF	0.5983	0.879	0.8998	0.8817	0.7965
135	NC_000012:5987546-5987546	rs216881	G	VWF	0.6029	0.8847	0.8998	0.8887	0.7986
136	NC_000012:5987525-5987525	rs216880	A	VWF	0.6014	0.8847	0.8998	0.8867	0.7975
137	NC_000012:5987510-5987510	rs526881	A	VWF	0.6014	0.8862	0.8998	0.8877	0.7975
138	NC_000012:5987507-5987507	rs526861	G	VWF	0.5484	0.8256	0.6647	0.7704	0.6585
139	NC_000012:5987489-5987489	rs216879	T	VWF	0.6014	0.8862	0.8998	0.8867	0.7975

140	NC_000012:5987217-5987217	rs216878	C	VWF	0.6498	0.8862	0.8998	0.8817	0.7965
141	NC_000012:5987128-5987128	rs216877	C	VWF	0.6498	0.8862	0.8998	0.8817	0.7965
142	NC_000012:5987090-5987090	rs216876	A	VWF	0.6498	0.8862	0.8998	0.8817	0.7965
143	NC_000012:5986735-5986735	rs216875	G	VWF	0.6778	0.8862	0.8998	0.8817	0.7965
144	NC_000012:5986641-5986642	rs71450640	TT	VWF	-	-	-	-	-
145	NC_000012:5986591-5986591	rs216874	C	VWF	0.6793	0.8862	0.8998	0.8817	0.7955
146	NC_000012:5986108-5986108	rs216873	C	VWF	0.621	0.8343	0.6617	0.7714	0.6575
147	NC_000012:5985679-5985679	rs177702	A	VWF	0.6399	0.8372	0.6607	0.7694	0.6564
148	NC_000012:5984916-5984916	rs24177	A	VWF	0.1104	0.3602	0.4871	0.2545	0.408
149	NC_000012:5984374-5984374	rs216872	C	VWF	0.1112	0.3588	0.4861	0.2555	0.409
150	NC_000012:5983882-5983882	rs216870	A	VWF	0.4115	0.7305	0.6548	0.6272	0.59
151	NC_000012:5982531-5982531	rs190156617	C	VWF	0.0015	0.0101	0	0.0089	0
152	NC_000012:5981998-5981998	rs216868	A	VWF	0.1029	0.3559	0.4831	0.2535	0.4029
153	NC_000012:5981834-5981834	rs216867	G	VWF	0.708	0.8991	0.9028	0.8817	0.7924
154	NC_000012:5981137-5981137	rs216865	C	VWF	0.4228	0.732	0.6518	0.6272	0.5869
155	NC_000012:5978365-5978365	rs216859	C	VWF	0.6785	0.8991	0.8958	0.8847	0.7904
156	NC_000012:5977875-5977875	rs216858	A	VWF	0.2723	0.6988	0.6389	0.6034	0.5573
157	NC_000012:5977770-5977770	rs216857	T	VWF	0.4614	0.732	0.6528	0.6292	0.5869
158	NC_000012:5977099-5977099	rs216856	C	VWF	0.4228	0.7305	0.6528	0.6272	0.5869
159	NC_000012:5976347-5976347	rs216855	G	VWF	0.4841	0.7824	0.8968	0.7425	0.726
160	NC_000012:5975698-5975698	rs216854	C	VWF	0.5666	0.7435	0.6478	0.6312	0.5869
161	NC_000012:5975547-5975547	rs216853	A	VWF	0.475	0.7334	0.6478	0.6312	0.5859
162	NC_000012:5975157-5975157	rs4764521	G	VWF	0.9629	0.8674	0.9206	0.8519	0.774
163	NC_000012:5975025-5975025	rs216852	C	VWF	0.3048	0.7406	0.6508	0.6491	0.6145
164	NC_000012:5973633-5973633	rs12829271	G	VWF	0.8449	0.5836	0.2956	0.7097	0.5286
165	NC_000012:5973586-5973586	rs10849366	G	VWF	0.7912	0.5807	0.2946	0.7097	0.5286
166	NC_000012:5973392-5973392	rs2239138	T	VWF	0.798	0.5807	0.2946	0.6988	0.5266
167	NC_000012:5972947-5972947	rs917859	G	VWF	0.7973	0.5807	0.2946	0.6988	0.5256
168	NC_000012:5972772-5972772	rs917858	T	VWF	0.7489	0.5735	0.2946	0.6918	0.5256
169	NC_000012:5971429-5971429	rs11063965	A	VWF	0.7958	0.5793	0.2927	0.6998	0.5245
170	NC_000012:5971411-5971411	rs2238110	C	VWF	0.7973	0.5793	0.2927	0.6998	0.5245
171	NC_000012:5971368-5971368	rs10849365	C	VWF	0.7519	0.5764	0.2927	0.6998	0.5245
172	NC_000012:5971124-5971124	rs7301070	A	VWF	0.7474	0.5764	0.2927	0.6988	0.5215
173	NC_000012:5970158-5970158	rs60982029	T	VWF	0.1974	0.1311	0.0159	0.1352	0.1493
174	NC_000012:5969892-5969892	rs12300917	T	VWF	0.1899	0.1686	0.0109	0.2038	0.181
175	NC_000012:5969772-5969772	rs11063964	G	VWF	0.4811	0.2334	0.1319	0.2525	0.2526
176	NC_000012:5969450-5969450	rs2270239	G	VWF	0.9644	0.572	0.3085	0.674	0.5286
177	NC_000012:5969258-5969258	rs35335161	T	VWF	0.0151	0.0346	0.001	0.0517	0.001
178	NC_000012:5968959-5968959	rs4764478	A	VWF	0.261	0.1729	0.0625	0.2316	0.2485
179	NC_000012:5968741-5968741	rs769080	A	VWF	0.9251	0.572	0.244	0.659	0.5153
180	NC_000012:5968688-5968688	rs2012829	C	VWF	0.9614	0.5778	0.245	0.661	0.5174
181	NC_000012:5968656-5968656	rs12307110	C	VWF	0.3533	0.1427	0.0218	0.1402	0.1595
182	NC_000012:5967775-5967775	rs216851	G	VWF	1	1	1	0.996	0.998
183	NC_000012:5965306-5965306	rs73049469	A	VWF	0.1702	0.1225	0.0218	0.1083	0.1973
184	NC_000012:5964354-5964354	rs12369177	G	VWF	0.7837	0.5605	0.2282	0.668	0.5971
185	NC_000012:5964017-5964017	rs12307884	C	VWF	0.3502	0.219	0.0248	0.3052	0.2474
186	NC_000012:5964012-5964012	rs12307883	C	VWF	0.6263	0.4539	0.1766	0.5258	0.4519

187	NC_000012:5962770-5962770	rs11063957	C	VWF	0.9539	0.4712	0.2192	0.6223	0.684
188	NC_000012:5962637-5962645	rs60444970	-	VWF	0.9047	0.3573	0.0367	0.4374	0.5941
189	NC_000012:5961673-5961673	rs2363309	A	VWF	0.7943	0.3444	0.0357	0.4314	0.59
190	NC_000012:5958468-5958468	rs11063956	A	VWF	0.2352	0.1816	0.0149	0.2684	0.3487
191	NC_000012:5958348-5958348	rs11063955	C	VWF	0.6604	0.2781	0.0337	0.3807	0.4693
192	NC_000012:5957986-5957986	rs7964554	T	VWF	0.6536	0.2781	0.0347	0.3807	0.4693
193	NC_000012:5957520-5957520	rs35926634	T	VWF	0.177	0.1744	0.0149	0.2684	0.3497
194	NC_000012:5957191-5957191	rs10849363	G	VWF	0.9062	0.3573	0.0367	0.4384	0.5951
195	NC_000012:5955904-5955904	rs11063953	G	VWF	0.2504	0.1859	0.0149	0.2694	0.3507
196	NC_000012:5955526-5955526	rs12819922	A	VWF	0.2496	0.1859	0.0149	0.2684	0.3497
197	NC_000012:5954793-5954793	rs723189	T	VWF	0.9531	0.4712	0.2163	0.6233	0.684
198	NC_000012:5953976-5953976	rs12317523	T	VWF	0.4047	0.2017	0.0159	0.2684	0.3497
199	NC_000012:5953817-5953817	rs7958883	A	VWF	0.9508	0.4712	0.2173	0.6243	0.684
200	NC_000012:5953723-5953723	rs12297442	G	VWF	0.2731	0.1873	0.0149	0.2684	0.3507
201	NC_000012:5953712-5953712	rs3759321	A	VWF	0.5091	0.3718	0.2004	0.5109	0.5644
202	NC_000012:5953687-5953687	rs3759320	T	VWF	0.8185	0.4568	0.2173	0.6233	0.684
203	NC_000012:5953213-5953213	rs12831864	G	VWF	0.9531	0.4712	0.2173	0.6233	0.684
204	NC_000012:5952781-5952781	rs7133777	C	VWF	0.9531	0.4712	0.2183	0.6233	0.684
205	NC_000012:5948745-5948745	rs1990326	G	VWF ANO2	0.9531	0.4726	0.2173	0.6233	0.684
206	NC_000012:5947502-5947502	rs11063951	C	VWF ANO2	0.1717	0.1744	0.0149	0.2694	0.3497
207	NC_000012:5947326-5947326	rs3809242	C	VWF ANO2	0.7814	0.8689	0.9792	0.838	0.7607
208	NC_000012:5947284-5947284	rs3809241	T	VWF ANO2	0.7769	0.866	0.9792	0.839	0.7607
209	NC_000012:5946627-5946630	rs3842300	- -	VWF ANO2	0.1838	0.1758	0.0149	0.2694	0.3497
210	NC_000012:5945966-5945966	rs6489682	G	VWF ANO2	0.7209	0.8516	0.9792	0.832	0.7505
211	NC_000012:5945874-5945874	rs6416322	T	VWF ANO2	0.7201	0.8516	0.9792	0.832	0.7505
212	NC_000012:5945582-5945582	rs6489681	C	VWF ANO2	0.8616	0.9885	1	1	1
213	NC_000012:5945229-5945229	rs6489680	G	VWF ANO2	0.59	0.9063	0.8383	0.9493	0.8742
214	NC_000012:5945034-5945034	rs117766816	A	VWF ANO2	0.0023	0.0144	0	0.0328	0.001
215	NC_000012:5943702-5943702	rs7314566	T	ANO2	0.1868	0.2767	0.1944	0.4592	0.4264

Ensemble Variant Effect Predictor (VEP) report allele frequency (AF) data for existing variants of the von Willebrand factor (gene) of index patient from genotyping project, the **gnomAD**. AFR (African), AMR (American), EUR (European), EAS (East Asian), SAS (South Asian)