全外显子基因解读报告

解读医生：王 X 明
医院：吉林大学第三医院
解读时间：2021-01-15

患者信息

姓名	年龄	性别
夏某泽	5	男
药物过敏史		**家族遗传病史**
无		无
病历摘要		
患者因左侧重度鼻衄于 1 月 13 日入院就诊， 鼻腔内无异物，为鼻黏膜出血。使用棉球填塞压迫及局部给药止血略有缓解。20 分钟后取出填塞棉球仍有出血现象。程度同初始强度。 体格检查：T 36.9°C，R：29 次/分，P：100 次/分，BP：12.7/7.9kPa (100/60mmHg)；营养发育良好，神志清醒，哭闹不止。血常规各项指标正常，血小板数量 265.0 x 10^9/L；血小板分布宽度 12.3 fL。血小板形态正常。 凝血功能障碍，怀疑： 1. 血小板无力症 2. 血友病 3. 血管性假血友病 4. 低纤维蛋白原血症		

检测结果

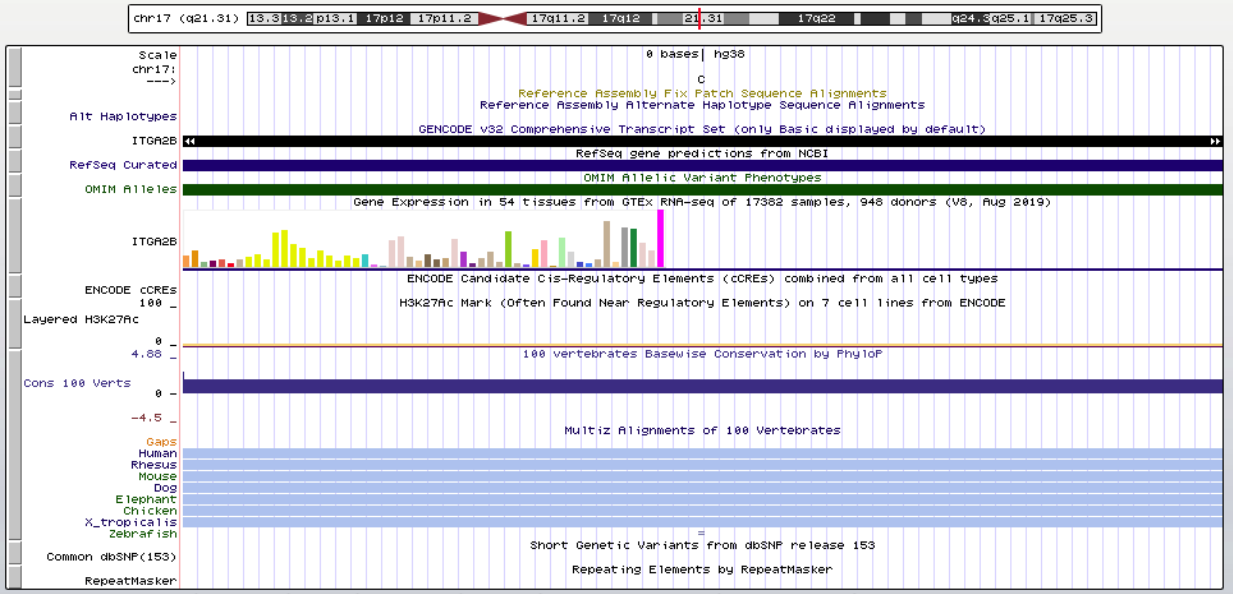
检测到 **3** 个高度关注变异：
17 号染色体上的 **ITGA2B** 基因变异，与 **Glanzmann Thrombasthenia** 高度相关。

检测到 **14** 个中度关注变异。

检测到 **64** 个其他基因变异。

高度关注 | Variants of High Interest

变异位点	染色体位置	关联疾病	参考/患者	基因	MAF（亚洲）	蛋白质变化	依据	文献
rs879255514	chr17:44372407	Glanzmann Thrombasthenia	TT/CC	ITGA2B	0.000002	p.R1025Q	dbNSFP	1-10



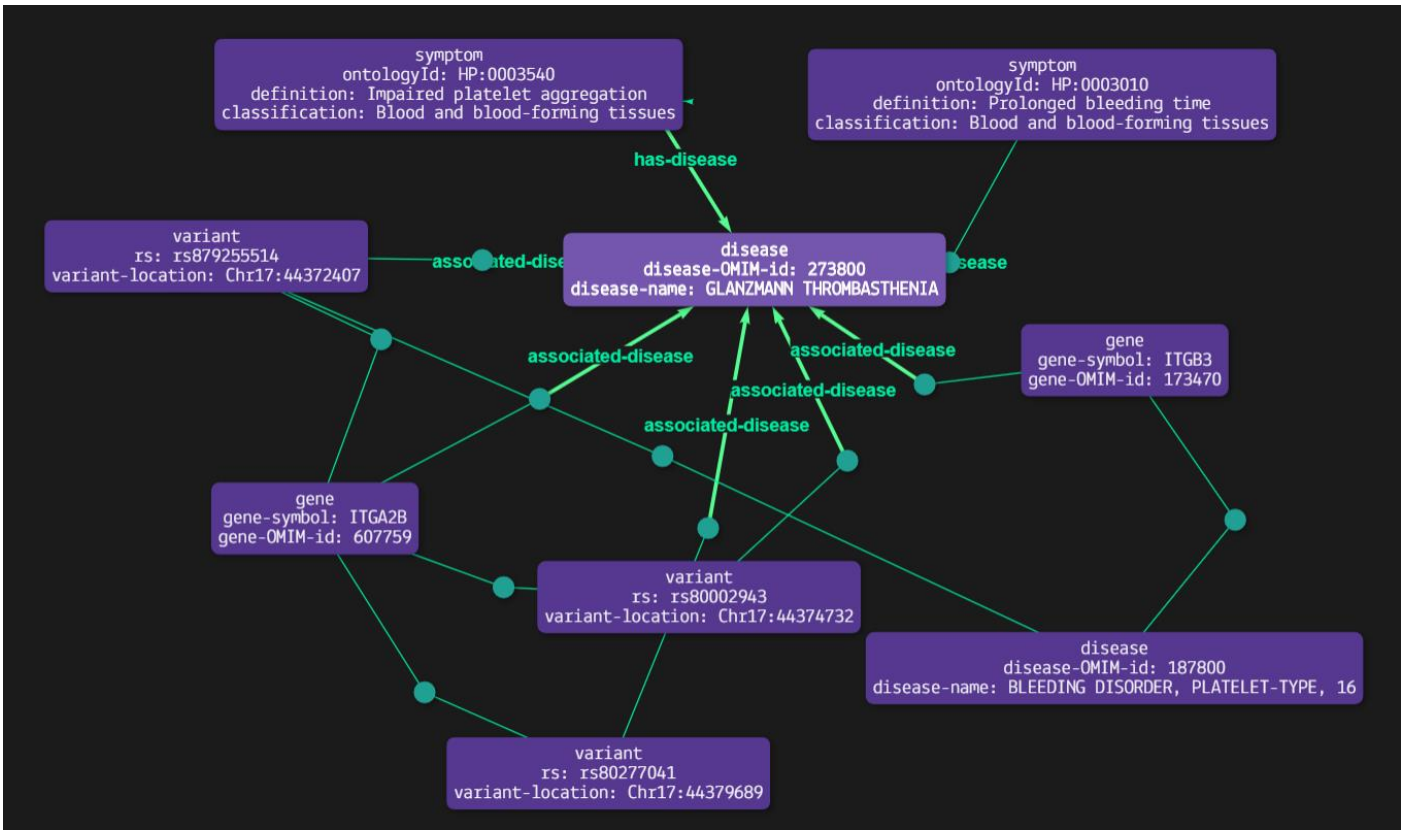
图一、该变异位点在 genome browser 上的展示。更多细节请见网页版报告。

疾病描述 | Disease Description

Glanzmann thrombasthenia is an autosomal recessive bleeding disorder characterized by failure of platelet aggregation and by absent or diminished clot retraction. The abnormalities are related to quantitative or qualitative abnormalities of the GPIIb/IIIa platelet surface fibrinogen receptor complex resulting from mutations in either the GPIIb or GPIIIa genes.

重点关注疾病 | Diseases of High Interest

疾病名	Glanzmann Thrombasthenia
发病年龄	Infancy, Neonatal, Childhood
遗传方式	Autosomal recessive
发病率	1/1000000



图二、该疾病与变异位点所在的基因之间关联的知识图谱展示。

中度关注 | Variants of Moderate Interest

变异位点	染色体位置	关联疾病	参考/患者	基因	MAF（亚洲）	蛋白质变化	依据
rs1186688662	1:91980649	Spermatogenic Failure 21	GG/AA	BRDT	0.003981951	p.A386T	OMIM
rs1380715569	2:231331896	Joubert Syndrome 30	GG/AA	ARMC9	0.008289343	p.G593E	OMIM
rs866378619	5:38881603	Amyloidosis	GG/GT	OSMR	0.006568597	p.S86I	GWAS
rs771613756	5:43609272	Glucocorticoid Deficiency 4	TT/TC	NNT	0.008119438	p.L26P	OMIM
rs1469014555	6:31116246	Peeling Skin Syndrome	GG/AA	CDSN	0.007723356	p.H457Y	LOVD
rs1288534485	7:44114251	Ehlers-Danlos Syndrome	GG/GA	AEBP1	0.005220995	p.G1156E	OMIM
rs1405925342	8:23702587	Conotruncal Heart Malformations	CC/TT	NKX2-6	0.006307472	p.G257D	OMIM
rs766372675	9:33386144	Glycerol Quantitative Trait Locus	GG/GA	AQP7	0.001521132	p.A96V	OMIM
rs1297244083	11:103245300	Short-Rib Thoracic Dysplasia 3	CC/TT	DYNC2H1	0.005903783	p.T3323I	OMIM
rs1393612239	11:118583971	Short Stature Micrognathia	AA/AG	ARCN1	0.0032131	p.I116V	OMIM
rs879334774	18:2740714	Bosma Arhinia Microphthalmia Syndrome	AA/GG	SMCHD1	0.006401085	p.T1176A	OMIM
rs1443923618	X:45069973	Kabuki Syndrome 2	TT/CC	KDM6A	0.002964494	p.L694P	OMIM
rs78848170	11:1017746	Schizophrenia	TT/GG	MUC6	0.004569373	p.L1685F	GWAS
rs200364398	11:1018042	Schizophrenia	AA/AG	MUC6	0.002877227	p.S1587P	GWAS

其它 | Other Variants of Interest

变异	染色体位置	Ref/Alt	MAF（亚洲）	基因	蛋白质变化
rs144490434	1:935833	CC/GG	0.005499761	SAMD11	p.P123A
rs761367418	7:5500610	GG/AA	0.008689522	FBXL18	p.I553I
rs148774441	7:27243238	GG/GA	0.002233892	EVX1	p.V70I
rs765411653	7:31643748	GG/AA	0.00166994	ITPRID1	p.C793Y
rs1487097257	7:39609805	GG/AA	0.002270831	YAE1	p.R147H
rs948775820	7:100550447	CC/CT	0.002506542	AGFG2	p.P123S
rs879868826	7:130023539	CC/TT	0.005792757	ZC3HC1	p.R359Q
rs571832233	7:142166237	GG/AA	0.007579759	MGAM2	p.R931Q
rs1007730288	8:96784901	AA/AG	0.005859888	CPQ	p.K2E
rs1022184348	8:143570921	AA/AG	0.002646293	MROH6	p.W226R
rs377389844	8:144520668	CC/TT	0.001959874	LRRC14	p.R254W
rs780424429	9:4849541	CC/GG	0.004212244	RCL1	p.S135C
rs139792014	9:35612978	GG/AA	0.007157938	CD72	p.S235L
rs186953177	9:136407354	CC/TT	0.007040409	ENTR1	p.G131S
rs1343392202	9:136407360	GG/GA	0.006488262	ENTR1	p.P129S
rs35567200	11:830044	GG/GA	0.004805473	CRACR2B	p.G173S
rs145926870	11:830120	AA/GG	0.0098083	CRACR2B	p.Q198R
rs761217284	11:7509682	GG/AA	0.002756617	OLFML1	p.E99K
rs371164865	11:96092376	CC/CT	0.000113824	MAML2	p.R552H
rs201667884	15:41476465	GG/AA	0.009825904	RTF1	p.R501K
rs778747499	17:4542963	CC/TT	0.009911717	MYBBP1A	p.E948K
rs1309343550	18:3173964	GG/AA	0.007958795	MYOM1	p.A383V
rs139919463	18:13826508	CC/TT	0.007660235	MC5R	p.T248I
rs34708574	19:281468	GG/GA	8.36709E-05	PLPP2	p.R263W
rs1330961569	19:15479367	GG/GA	0.005474043	PGLYRP2	p.A2V
rs147820524	19:18932960	GG/AA	0.00917671	HOMER3	p.T130I
rs375651812	19:19267990	CC/TT	0.006852981	TM6SF2	p.G236D
rs200007011	19:46329438	GG/AA	0.003923494	HIF3A	p.D489N
rs1288031997	6:150840123	GG/AA	0.002287954	PLEKHG1	p.V1090M
rs749066113	22:38822698	CC/CT	0.005110849	NPTXR	p.V472I
rs377029853	6:47707248	GG/AA	0.001867415	ADGRF4	p.M1I
rs1233164249	6:24850715	AA/GG	0.00204071	RIPOR2	p.M256T
rs763158102	1:1916890	CC/TT	0.005101701	CALML6	p.T114I
rs782069935	1:149908174	AA/GG	0.006462967	SV2A	p.M471T
rs372163409	2:46001425	GG/AA	0.00876242	PRKCE	p.R282H
rs1283017731	2:53801491	GG/AA	0.001914884	ERLEC1	p.R207K
rs750191512	2:127631294	CC/TT	0.006645531	MYO7B	p.R1650X
rs1272748573	2:130762653	GG/AA	0.001690122	AMER3	p.G194E
rs1396668991	2:201160699	TT/CC	0.004971572	CFLAR	p.M109T
rs375651741	2:241872597	AA/GG	0.006831949	RTP5	p.I348V
rs765561559	3:52743384	GG/AA	0.007239168	NEK4	p.R569X
rs1230695160	3:58424917	GG/AA	0.008612732	PXK	p.R407K

变异	染色体位置	Ref/Alt	MAF（亚洲）	基因	蛋白质变化
rs143580910	3:73384912	CC/TT	0.006378443	PDZRN3	p.G250S
rs921509869	3:97877334	AA/GG	0.005378614	CRYBG3	p.D2047G
rs1189500747	6:31701303	GG/GA	0.009788571	ABHD16A	p.P76L
rs150691200	3:140404653	AA/GG	0.008346339	CLSTN2	p.Y175C
rs773314504	3:131029865	CC/CT	0.00460541	NEK11	p.R53X
rs1378229397	5:79237489	AA/GG	0.006271904	JMY	p.Q280R
rs1484748624	6:4031998	AA/GG	0.004361617	PRPF4B	p.S161G
rs994927193	6:1312744	GG/AA	0.004609372	FOXQ1	p.D14N
rs201687291	5:178120334	CC/TT	0.009089563	N4BP3	p.R163W
rs542414446	5:154053249	AA/GG	0.003598436	MFAP3	p.I63V
rs201059756	5:140568131	AA/GG	0.008551595	SLC35A4	p.Y321C
rs956482411	3:183951880	TT/TC	0.003269598	ABCC5	p.I459V
rs889418995	22:45931262	CC/CT	0.003843697	WNT7B	p.E136K
rs762609462	5:43292908	CC/AA	0.006447103	HMGCS1	p.G375C
rs763308335	5:1325767	AA/GG	0.008299218	CLPTM1L	p.L377P
rs752054203	4:48535722	CC/CT	0.009592604	FRYL	p.V2167M
rs1048654784	4:39500244	TT/TC	0.007302359	UGDH	p.I395V
rs1462640773	4:27022800	AA/GG	0.009764561	STIM2	p.N682S
rs552041309	3:194341327	GG/AA	0.007705478	CPN2	p.P459L
rs1419816724	1:1287127	GG/AA	0.007010895	SCNN1D	p.D380N
rs765964710	18:2891847	CC/CT	0.007412654	EMILIN2	p.R574C
rs1370067106	9:34723935	CC/TT	0.006693399	FAM205A	p.S1102N

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