全外显子基因解读报告

解读医生：徐 X 岩
医院：吉林大学第三医院
解读时间：2021-01-08

患者信息

姓名	年龄	性别
石某世	2	男
药物过敏史	**家族遗传病史**	
无	无	

病历摘要

患者因“下肢运动障碍 3 天，行走缓慢，易反复跌倒”于 1 月 6 日入院就诊。
平素健康，无呕吐，腹泻症状，无外伤史。三天前行动不便，上楼梯尤其容易跌倒。跑跳较困难，经常乏力。无吞咽困难，上肢运动正常。既往无类似病史。
体格检查：T 37.1°C，R：28 次/分，P：108 次/分，BP：12.7/8kPa (100/62mmHg)；营养发育良好，神志清醒。神经系统检查： 颅神经未见异常。周围神经：上肢肌力，肌张力，肌容积正常，抓握功能正常。左下肢肌力 III 级，右下肢肌力 IV 级，痛感觉，位置感觉仍存在。骨骼形态，肌肉型态尚未见异常。
诊断怀疑：
1. 杜兴氏肌肉营养不良（Duchenne Muscular Dystrophy）
2. 脊髓性肌肉萎缩（Spinal Muscular Atrophy）

检测结果

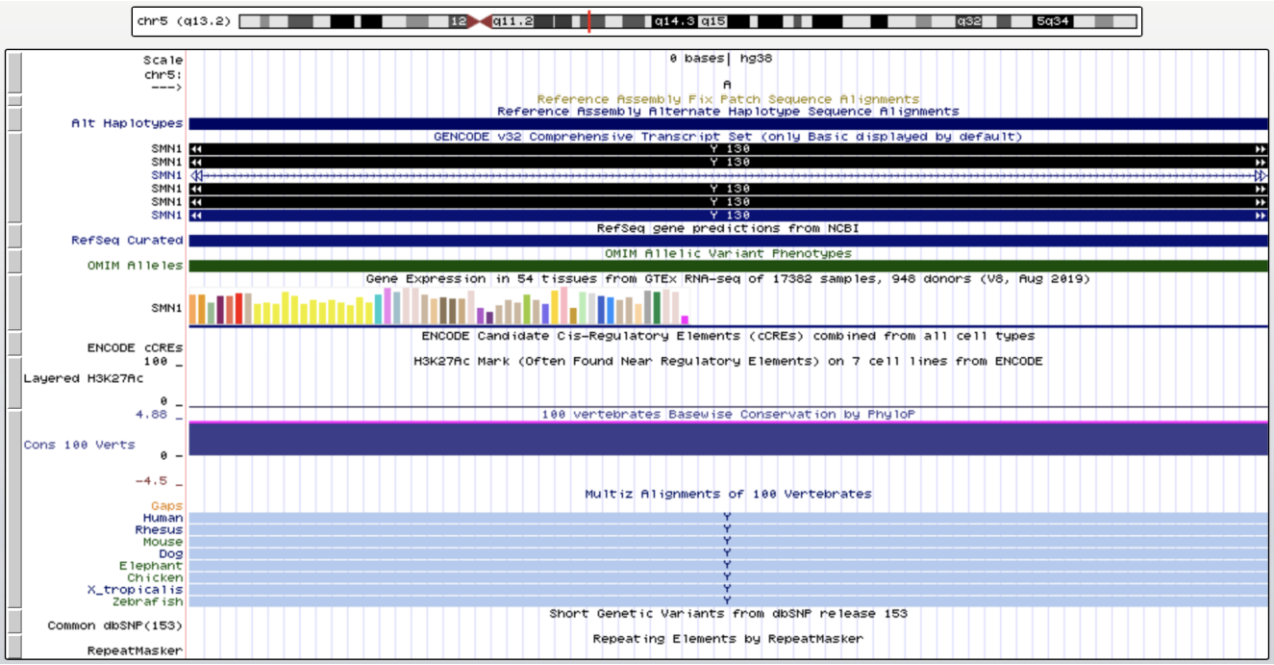
检测到 **1** 个高度关注变异：
5 号染色体上的 **SMN** 和 **SMN2** 基因变异， 与 **Spinal Muscular Atrophies III** 高度相关

检测到 **16** 个中度关注变异。

检测到 **41** 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
rs397514517	chr5:70942473	Spinal Muscular Atrophy III	AG/AA	SMN1,SMN2	0.00002	p.Y130C	OMIM	1-10



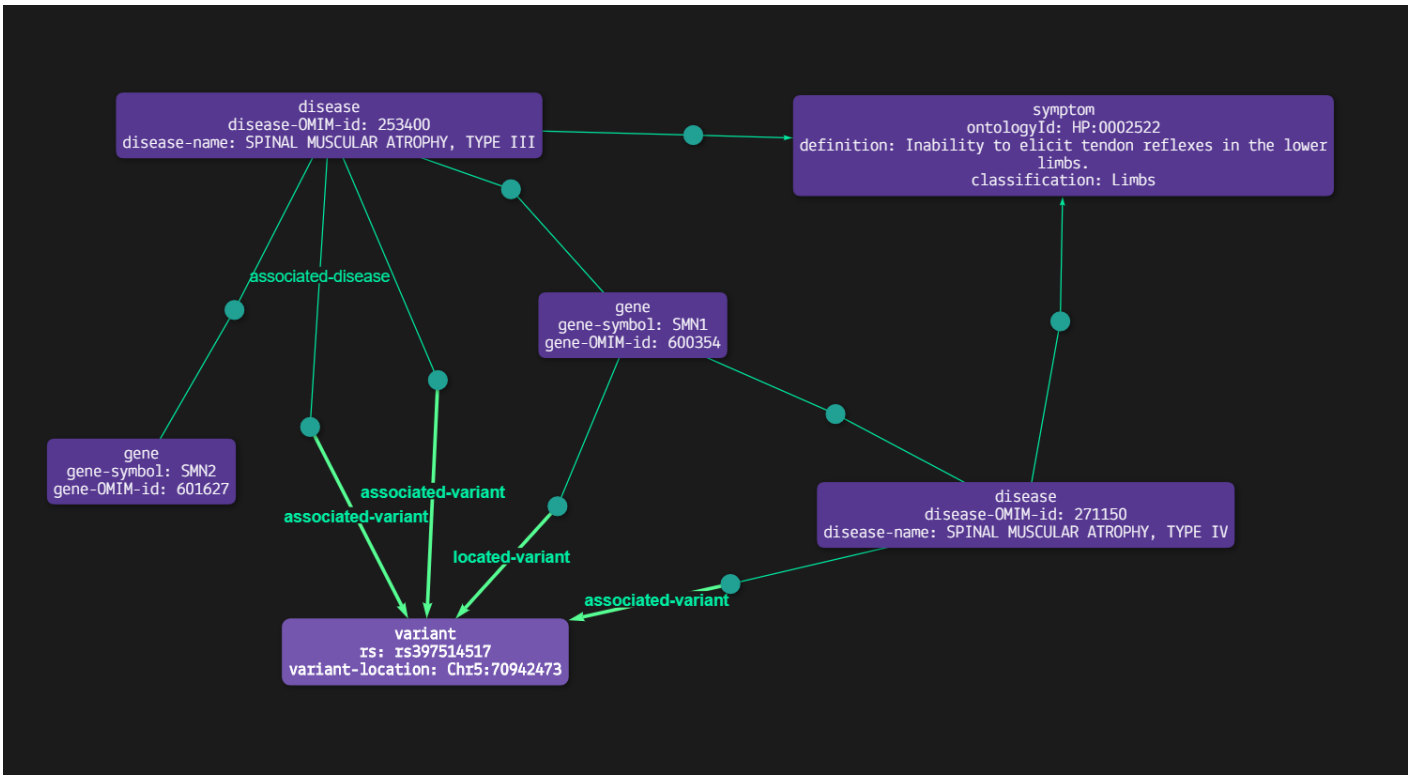
图一、该变异位点在 genome browser 上的展示。更多细节请见网页版报告。

疾病描述 | Disease Description

SMA is an autosomal recessive neuromuscular disorder characterized by progressive proximal muscle weakness and atrophy affecting the upper and lower limbs. By convention, SMA is classified into 4 types: SMA I, SMA II, SMA III, and SMA IV, by increasing age at onset and decreasing clinical severity. SMA I is the most severe form of the disorder and often results in death in early childhood. SMA III, known as the juvenile form, tends to show onset in childhood or adolescence.

重点关注疾病 | Diseases of High Interest

疾病名	Spinal Muscular Atrophy III
发病年龄	Childhood
遗传方式	Autosomal recessive
发病率	0.000045



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
rs1207848794	17:76293190	Spermatogenic Failure 35	GG/AA	QRICH2	0.001514	p.Q347X	OMIM
rs373171844	X:71384985	Mental Retardation	GG/AA	TAF1	0.005518	p.G741E	OMIM
rs756161014	5:1065297	Breast Cancer	CC/CT	SLC12A7	0.00967	p.W808X	GWAS
rs766186198	5:131562015	Creutzfeldt-Jakob Disease	CC/TT	RAPGEF6	0.006893	p.C105Y	GWAS
rs200166093	7:151064305	Creutzfeldt-Jakob Disease	AA/AG	SLC4A2	0.000264	p.Q43R	GWAS
rs912228422	9:34256347	Creutzfeldt-Jakob Disease	CC/CT	KIF24	0.004591	p.G1087D	GWAS
rs748039331	10:102141278	Kaposi Sarcoma	CC/CT	PPRC1	0.005519	p.P924S	NLP
rs767376632	11:4682519	Creutzfeldt-Jakob Disease	GG/GA	OR51E2	0.003916	p.L65F	GWAS
rs773337252	15:40458807	Creutzfeldt-Jakob Disease	CC/CT	BAHD1	0.007029	p.R115C	GWAS
rs1462414343	16:81445429	Gastric Cancer	GG/GA	CMIP	0.004587	p.C63Y	NLP
rs377222562	18:9258531	Alzheimer Disease	CC/TT	ANKRD12	0.004824	p.T1732I	GWAS,
rs765501008	18:63489378	Schizophrenia	CC/TT	SERPINB5	0.007409	p.A113V	GWAS
rs1028072297	19:3162909	Obesity	CC/CT	GNA15	0.007194	p.L339F	GWAS
rs1172291775	19:4511283	Alzheimer Disease	CC/CT	PLIN4	0.001295	p.G893S	GWAS
rs1339531234	19:4511955	Alzheimer Disease	TT/TC	PLIN4	0.004684	p.S669G	GWAS
rs866253502	20:42472272	Obesity	CC/TT	PTPRT	0.006573	p.E482K	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
rs879868826	7:130023539	CC/CT	0.0066039	ZC3HC1	p.R359Q
rs1022184348	8:143570921	AA/GG	0.0099841	MROH6	p.W226R
rs377389844	8:144520668	CC/TT	0.004011575	LRRC14	p.R254W
rs780424429	9:4849541	CC/CG	0.009407099	RCL1	p.S135C
rs1478846720	10:15214177	AA/GG	0.001929161	FAM171A1	p.S471P
rs35567200	11:830044	GG/GA	0.005477289	CRACR2B	p.G173S
rs145926870	11:830120	AA/AG	0.001503249	CRACR2B	p.Q198R
rs761217284	11:7509682	GG/GA	0.006261801	OLFML1	p.E99K
rs1487097257	7:39609805	GG/AA	0.004724392	YAE1	p.R147H
rs371164865	11:96092376	CC/CT	0.00643273	MAML2	p.R552H
rs778747499	17:4542963	CC/CT	0.002329345	MYBBP1A	p.E948K
rs1309343550	18:3173964	GG/GA	0.002768591	MYOM1	p.A383V
rs139919463	18:13826508	CC/TT	0.004051673	MC5R	p.T248I
rs1309343550	18:3173964	GG/GA	0.004936476	MYOM1	p.A383V
rs1309343550	18:3173964	GG/GA	0.006089687	MYOM1	p.A383V
rs1309343550	18:3173964	GG/GA	0.008971924	MYOM1	p.A383V
rs1330961569	19:15479367	GG/AA	0.003967819	PGLYRP2	p.A2V
rs375651812	19:19267990	CC/CT	0.006748945	TM6SF2	p.G236D
rs201667884	15:41476465	GG/GA	0.004014261	RTF1	p.R501K
rs200007011	19:46329438	GG/GA	0.001306437	HIF3A	p.D489N
rs889418995	22:45931262	CC/TT	0.009769024	WNT7B	p.E136K
rs1484748624	6:4031998	AA/AG	0.006150418	PRPF4B	p.S161G
rs763158102	1:1916890	CC/CT	0.007932351	CALML6	p.T114I
rs76597070	1:3752792	GG/AA	0.005846459	CCDC27	p.R104K
rs769259082	1:41583075	GG/GA	0.00355996	HIVEP3	p.H575Y
rs746371650	1:58530619	TT/CC	0.009738717	OMA1	p.I374M
rs746244698	1:1454424	CC/CT	0.00042107	ATAD3C	p.A101V
rs782069935	1:149908174	AA/AG	0.009163632	SV2A	p.M471T
rs782069935	1:149908174	AA/AG	0.002926304	SV2A	p.M471T
rs1233164249	6:24850715	AA/AG	0.008969005	RIPOR2	p.M256T
rs1031030041	1:15546825	AA/GG	0.004553452	DNAJC16	p.H273R
rs782069935	1:149908174	AA/AG	0.0050858	SV2A	p.M471T
rs375651741	2:241872597	AA/GG	0.001514195	RTP5	p.I348V
rs956482411	3:183951880	TT/CC	0.003071656	ABCC5	p.I459V
rs1462640773	4:27022800	AA/GG	0.002908643	STIM2	p.N682S
rs763308335	5:1325767	AA/GG	0.00847634	CLPTM1L	p.L377P
rs201059756	5:140568131	AA/AG	0.009875742	SLC35A4	p.Y321C
rs994927193	6:1312744	GG/AA	0.006865883	FOXQ1	p.D14N
rs1370753468	1:156540754	AA/AG	0.003203552	IQGAP3	p.M898T
rs1419816724	1:1287127	GG/AA	0.006747603	SCNN1D	p.D380N
rs1419816724	1:1287127	GG/AA	0.003467189	SCNN1D	p.D380N

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