



检测项目

GATK Germline SNP/Indel V2.0

报告编号

7192128156063498240

报告日期

2024-05-07

一、项目信息

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二、样本信息

患者姓名

样本编号

测序设备

SRR9993255

患者性别

样本类型

取样日期

2022-02-21

患者年龄

取样方法

核收日期

2022-02-21

患者编号

6901194220845924353

取样位置

报告日期

2024-05-07

诊断信息

样本来源

临床信息

三、参考基因组版本

参考基因组版本

hg38

项目bed文件

None

四、软件版本



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fastqc

0.12.1

multiqc

1.21

fastp

0.23.2

bwa

0.7.17

samtools

1.16.1

bedtools

2.30.0

sambamba

1.0.1

gatk

4.3.0.0

version_vep

108

version_vep_cache

108

五、数据质控

Sample Number	Total Reads Before Filtering(M)	Q20 Before Filtering(%)	Q30 Before Filtering(%)
SRR9993255	1.36	96.31%	90.9%

五、致病突变

Chr	Start	End	Ref	Alt	DP	VAF	GeneID	
13	20189347	20189347	G	-	239	50.00%	ENSG00000165474	G
16	21736318	21736318	G	T	318	50.00%	ENSG00000155719	O

六、突变文件



CHROM	START	END	ID
chr1	6440984	6440984	rs2311045
chr1	40819415	40819415	rs4660468
chr1	102886895	102886895	rs2229783
chr1	102888582	102888582	rs1676486
chr1	102888872	102888872	rs1763347
chr1	102914362	102914362	rs3753841
chr1	169468154	169468154	rs17847484
chr1	215675245	215675245	rs2797234
chr1	215675299	215675299	rs2797235
chr1	215741484	215741484	rs35309576
chr1	215743221	215743221	rs11120616
chr1	215786825	215786825	rs10864198
chr1	215813880	215813880	rs4129843
chr1	215838019	215838019	rs56032526
chr1	215877783	215877783	rs41277200
chr1	215999038	215999038	rs10864219
chr1	216175422	216175422	rs1805049
chr1	216325569	216325569	rs3767698
chr1	216418661	216418661	rs4253963
chr1	216421964	216421964	rs10779261
chr1	217424175	217424175	rs3806268

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七、突变注释文件



Uploaded_variation	Location	Allele	Gene	Feature
rs2311045	chr1:6440984	G	ENSG00000187017	ENST00000418286
rs2311045	chr1:6440984	G	ENSG00000231868	ENST00000419034
rs2311045	chr1:6440984	G	ENSG00000187017	ENST00000632142
rs2311045	chr1:6440984	G	ENSG00000187017	ENST00000633651
rs2311045	chr1:6440984	G	ENSG00000187017	ENST00000636330
rs2311045	chr1:6440984	G	ENSG00000187017	ENST00000645284
rs2311045	chr1:6440984	G	-	ENSR00000000670
rs2311045	chr1:6440984	G	-	ENSR00000345889
rs4660468	chr1:40819415	C	ENSG00000117013	ENST00000347132
rs4660468	chr1:40819415	C	ENSG00000117013	ENST00000443478
rs4660468	chr1:40819415	C	ENSG00000117013	ENST00000506017
rs4660468	chr1:40819415	C	ENSG00000117013	ENST00000509682
rs4660468	chr1:40819415	C	-	ENSR00000005258
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000353414
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000358392
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000370096
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000512756
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000635193
rs2229783	chr1:102886895	G	ENSG00000060718	ENST00000639098
rs1676486	chr1:102888582	G	ENSG00000060718	ENST00000353414
rs1676486	chr1:102888582	G	ENSG00000060718	ENST00000358392
rs1676486	chr1:102888582	G	ENSG00000060718	ENST00000370096

