

CLEAR FILTERS

NEW CASE

	↓ ID	Accession ID	Genome (ID)	Projects	Test	Assigned	GEM	Status	Flag	Owner	Created	QC	Note
<input type="checkbox"/>	1177282	SFC211110004	Variant Call Data from job 41144 – SFC211110004 (1054762)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177281	SFC2111130001	Variant Call Data from job 41149 – SFC2111130001 (1054752)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177280	SFC2111140004	Variant Call Data from job 41153 – SFC2111140004 (1054750)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177279	SFC2111120004	Variant Call Data from job 41148 – SFC2111120004 (1054745)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177278	SFC2111120001	Variant Call Data from job 41145 – SFC2111120001 (1054757)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177277	SFC2111120002	Variant Call Data from job 41146 – SFC2111120002 (1054748)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177276	SFC2111110003	Variant Call Data from job 41143 – SFC2111110003 (1054746)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177275	SFC2111120003	Variant Call Data from job 41147 – SFC2111120003 (1054753)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177274	SFC2111140003	Variant Call Data from job 41152 – SFC2111140003 (1054751)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177273	SFC2111140001	Variant Call Data from job 41150 – SFC2111140001 (1054747)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	
<input type="checkbox"/>	1177272	SFC2111140006	Variant Call Data from job 41155 – SFC2111140006 (1054761)	11-20-2021-05-08	TruSight Cancer panel - CGX	ACE		Needs Review			a day ago	qc	

INTERPRET VARIANTS (12)

REPORT REVIEW

Case Info [View More](#)

Accession ID: SFC211110004

Status: Needs Review

Clinical Grade: 0

QC Status: [VIEW FILES](#)

Test: TruSight Cancer panel



Patient Sex: Female

Patient Age: 86

+ ADD GEM REPORT

SNVs/INDELS	PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN		ALL SVS										Classification (Condition)
	Gene (Transcript)	Position dbSNP	Change	Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status			
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser	None ▾		Missense	●○	5461.77 99 389 : 151 : 238	0.00004 0.00002			0.937 	56 37	-		Uncertain S Lynch Sync	
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47637246 rs17217772	A → G c.380A>G p.Asn127Ser	None ▾		Missense	●○	7079.77 99 595 : 321 : 274	0.02476 0.02637 0.00699 0.00753			0.571 	0 15	Likely Benign (Jul 30, 2021)		Likely Ben Lynch Sync	
<input type="checkbox"/>	BAP1 (ENST00000460680)	chr3:52437258 rs79014342	T → C c.1786A>G p.Ser596Gly	None ▾		Missense	●○	15081.8 99 1119 : 550 : 569	0.02696 0.02407 0.00715 0.00765			0.051 	0 7	Likely Benign (Jul 30, 2021)		Likely Ben BAP1 tumor	
<input type="checkbox"/>	APC (ENST00000457016)	chr5:112175651 rs111866410	A → G c.4360A>G p.Lys1454Glu	None ▾		Missense	●○	13529.8 99 1199 : 649 : 550	0.0008 0.00246 0.00054 0.00081			0.608 	13 12	-		Likely Ben Familial ad	
<input type="checkbox"/>	PMS2 (ENST00000265849)	chr7:6026864 rs74902811	G → A c.1532C>T p.Thr511Met	None ▾		Missense	●○	12670.8 99 1104 : 560 : 544	0.03694 0.02645 0.00939 0.00984			0.162 	6 10	Likely Benign (Aug 8, 2021)		Likely Ben Lynch Sync	
<input type="checkbox"/>	MET (ENST00000318493)	chr7:116340223 rs77523018	T → C c.1085T>C p.Met362Thr	None ▾		Missense	●○	11235.8 99 1130 : 611 : 519	0.00719 0.00689 0.00198 0.00232			0.776 	7 11	Likely Benign (Sep 12, 2021)		Likely Ben Papillary re	
<input type="checkbox"/>	ATM (ENST00000278616)	chr11:108121733 rs2235000	G → A c.1541G>A p.Gly514Asp	None ▾		Missense	●○	13559.8 99 1092 : 552 : 540	0.01757 0.02116 0.00532 0.00055			0.709 	12 19	Likely Benign (Oct 21, 2021)		Likely Ben Breast can	



INTERPRET VARIANTS (12)

REPORT REVIEW

Case Info [View More](#)

Accession ID: SFC211110004

Status: Needs Review

Clinical Grade: 0

QC Status: [VIEW FILES](#)

SNVS/INDELS PATH/LIKELY PATH BENIGN/LIKELY

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

Score Variant MSH2

← 7 of 28 →

Null variant (nonsense, frameshift, splice sites, initiation codon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code	PVS1	Support	Pathogenic
Section	Effect	Level	Very Strong

Criteria met?

Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ▾

SCORING AUDIT LOG SCORING SHEET

SET CLASSIFICATION

Clinical Grade

Genome

Variant Call Data from job 41144 (1054762) 405

Total Variants



Clinical Grade

Total Coding Variants

203

+ ADD GEM REPORT

The clinical grade measures the quality of your variant file. One bar is earned when any of the below criteria are met. Four bars is the maximum number of bars.

✓ Coverage 900.0

The median coverage of coding variants exceeds 50

✓ Genotype Quality 100.0

More than 95% of the coding variants have a quality above 40

✗ Homozygous / Heterozygous Ratio 0.52

The ratio for the coding variants is between 0.47 and 0.52

✓ Transition / Transversion Ratio 2.79

The ratio for the coding variants is between 2.71 and 3.17



Coding Variants Only

CLOSE

INTERPRET VARIANTS (12)

REPORT REVIEW

 Case Info [View More](#)

Accession ID: SFC211110004

Status: Needs Review

Clinical Grade: 0

QC Status: [VIEW FILES](#)

Test: TruSight Cancer panel

Patient Sex: Female

Patient Age: 86

+ ADD GEM REPORT

SNVS/INDELS	PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status
<input type="checkbox"/>	MSH2 ENST00000233146	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None

X Score Variant | MSH2

← 7 of 28 →

Section Effect

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code PVS1
Section EffectSupport Pathogenic
Level Very Strong

Criteria met?

[Skip](#) [Clear](#)

Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION

SCORING AUDIT LOG SCORING SHEET

SET CLASSIFICATION

Case Info: #1177282

CASE INFORMATION	TEST INFORMATION
Patient SFC211110004 Female, 86 years of age EMMA JAKOBET	Genome: Variant Call Data from job 41144 - SFC211110004 (1054762) DOB: May 19 1935
Edit Patient Information	Close

INTERPRET VARIANTS (12)

REPORT REVIEW

 Case Info [View More](#)

Accession ID: SFC2111110004

Status: Needs Review

Clinical Grade:

QC Status: [VIEW FILES](#)

Test: TruSight Cancer panel

Patient Sex

Patient Age

+ ADD GEM REPORT

SNVs/INDELS PATH/LIKELY PATH BENIGN/LIKELY BENIGN VUS

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None

X Score Variant MSH2

← 7 of 28 →

Section Effect ▾

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code PVS1 Support Pathogenic
Section Effect Level Very StrongCriteria met? [Skip](#) [Clear](#) Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

Case Info: #1177282

CASE INFORMATION

TEST INFORMATION

Case ID 1177282 Test TruSight Cancer panel - CGX
 Patient Accession ID SFC2111110004 Rubric ACMG Mendelian
 Genome Pipeline Version 6.6.9
 Variant Call Data from job 41144 (SFC2111110004)
 Project Assembly Version b37
 Status Include COSMIC False
 Runs of Homozygosity ROH

Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status		Classification (Condition)
0.937	56 37	-		Uncertain Significance Lynch Syndrome

EDIT TEST INFORMATION

CLOSE

VARIANT DESCRIPTION ▾

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

app.fabricgenomics.com/w/2428/clinical_reporter#/report/1177282/interpret[Apps](#) [Google](#) [AMBOSS login](#) [Orthobullets](#) [Curation workspace](#) [OMIM](#) [gnomAD](#) [ClinVar](#) [AA](#) [Sci-Hub](#) [PanelAPP](#) [UniProt](#) [Orphanet](#) [GARD](#) » [Reading List](#)

← Accession ID: SFC2111110004

Sanger - Production



INTERPRET VARIANTS (12) REPORT REVIEW



SNVs/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN ⓘ		ALL SVS													
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF	EVS AF	ExAC AF	gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified)	Confirmation Status		Classification Condition
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None ▾		Missense	● ○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002	0.00004 0.00002	0.00004 0.00002	0.00002		0.937 	56 37	-			Uncertain Significance Lynch Syndrome	

[SCORE VARIANT](#)

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INTERPRET VARIANTS (12)

REPORT REVIEW

SNVs/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN		ALL SVS														
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygoticity	Quality GQ Coverage	1KG AF	EVS AF	ExAC AF	gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status		Classification (Condition)	Scoring Status	Report Status

INTERPRET VARIANTS (12)

REPORT REVIEW

SNVS/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN		ALL SVS									
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygosity	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status		Classification (Condition)
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None		Missense	●○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002			0.937 	56 37	-		Uncertain Significance Lynch Syndrome

INTERPRET VARIANTS (12)

REPORT REVIEW

SNVs/INDELS PATH/LIKELY PATH BENIGN/LIKELY

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

X Score Variant MSH2

← 7 of 28 →

Null variant (nonsense, frameshift, splice sites, initiation codon deletion) in a gene where LOF is a known mechanism of disease.

Caveats

Code	PVS1	Support	Pathogenic
Section	Effect	Level	Very Strong

Criteria met?

 Yes No

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

Configure SOP Columns

Visibility of SOP columns is set in the step configuration and cannot be changed here.

Show	Column	Move
<input checked="" type="checkbox"/>	Gene	↑ ↓
<input checked="" type="checkbox"/>	Position	↑ ↓
<input checked="" type="checkbox"/>	Change	↑ ↓
<input checked="" type="checkbox"/>	Flag	↑ ↓
<input checked="" type="checkbox"/>	Confirmation Status	↑ ↓
<input checked="" type="checkbox"/>	Links	↑ ↓
<input checked="" type="checkbox"/>	Effect	↑ ↓
<input checked="" type="checkbox"/>	Zygoticity	↑ ↓
<input checked="" type="checkbox"/>	Quality / Coverage	↑ ↓
<input checked="" type="checkbox"/>	Frequency	↑ ↓
<input checked="" type="checkbox"/>	Review Priority	↑ ↓
<input checked="" type="checkbox"/>	Evidence	↑ ↓
<input checked="" type="checkbox"/>	Omicia Score	↑ ↓

RESTORE DEFAULTS

CANCEL APPLY

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

INTERPRET VARIANTS (12)

REPORT REVIEW

Case Info [View More](#)

Accession ID: SFC2111110004

Status: Needs Review

Clinical Grade: 0

QC Status: QC [VIEW FILES](#)Test: TruSight Cancer panel ACE

Patient Sex: Female

Patient Age: 86

SNVs/INDELS

PATH/LIKELY PATH

BENIGN/LIKELY BENIGN

VUS/UNCERTAIN i

ALL SVS

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Lates Date Confir
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None		Missense	● ○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002			0.937	56 37	-

Score Variant

MSH2

← 7 of 28 →

Section [Effect](#) ▾

Variant is not a LOF variant

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

[Caveats](#)Code PVS1
Section EffectSupport Pathogenic
Level Very Strong

Criteria met?

[Skip](#) [Clear](#) Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING A

RESET FILTERS

APPLY

X Add Filters

Filtering Protocols

Report Filters

Gene Filters

Zygoty

RSID

Location

Quality

Evidence

Allele Frequency

Impact Scores

Variant Effect

Exclude

Case Info [View More](#)

Accession ID: ASO2111100103

Status: Needs Review

Clinical Grade: 0

QC Status: QC [VIEW FILES](#)

Test: TruSight Cardio panel

ACE

Patient Sex: Male

Patient Age: 61

Add Filters



Variant Type

 Protein Changing Structural Variant Stop Gained/Lost Initiator Codon Variant Frameshift Indel In-Frame Indel Splice Site Splice Region Missense Regulatory

Gene Models

 CCDS RefSeq

rows per page:

[RESET FILTERS](#)

SNVs/INDELS	PATH/LIKELY PATH	BENIGN/LIKELY BENIGN	VUS/UNCERTAIN	ALL SVS	Confirmation Status	Links	Effect	Zygosity	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		None ▾	BAM M	Missense	●○	4708.77 99 415 : 230 : 185	0.02656 0.02845 0.00892 0.00935	●●●	CV
<input type="checkbox"/>	PCSK9 (ENST00000302118)	chr1:55523855 rs28362263	G → A c.1327G>A p.Ala443Thr		None ▾	BAM M	Missense splice site impact	●○	4362.77 99 450 : 248 : 202	0.00439 0.00455 0.00185 0.00133	● ●●●	CV
<input type="checkbox"/>	PCSK9 (ENST00000302118)	chr1:55527222 rs28362277	A → C c.1856A>C p.Gln619Pro		None ▾	BAM M	Missense	●○	408.77 99 510 : 334 : 176	0.01957 0.02114 0.00585 0.00614	● ●●●	CV
<input type="checkbox"/>	NEXN (ENST00000334785)	chr1:78401682	G → C c.1426G>C p.Ala476Pro		None ▾	BAM	Missense	●○	6046.77 99 495 : 251 : 244	0.0004 0.00075 0.00016 0.00017	● ●●●	CV
<input type="checkbox"/>	ACTN2 (ENST00000366578)	chr1:236914923 rs35997569	A → G c.1810A>G p.Met604Val		None ▾	BAM M	Missense	●○	7102.77 99 526 : 242 : 284	0.0028 0.00323 0.00371 0.00394	● ●●●	CV
<input type="checkbox"/>	RYR2 (ENST00000366574)	chr1:237711759 rs202015519	G → T c.2935G>T p.Ala979Ser		None ▾	BAM M	Missense	●○	6898.77 99 548 : 261 : 287	0.0028 0.00323 0.00371 0.00394	●●●	CV
<input type="checkbox"/>	APOB (ENST00000233242)	chr2:21238367 rs12713843	C → T c.3383G>A p.Arg1128His		None ▾	BAM M	Missense	●○	5330.73 99 77 : 1 : 76	- - 0.01289 0.01199	●●●	CV
<input type="checkbox"/>	ALMS1 (ENST00000264448)	chr2:73613031 rs878855000	TGGAGGAGGAGGAGGA → T c.60_74del p.Glu24_Glu28del		None ▾	BAM	Inframe deletion	●○	- - - -	- - - -	●●●	CV





Accession ID: SFC2111110004

Sonoran - Production



INTERPRET VARIANTS (12)

REPORT REVIEW

Case Info [View More](#)

Accession ID: SFC2111110004

Status: Needs Review

Clinical Grade: 0

QC Status: QC [VIEW FILES](#)

Test: TruSight Cancer panel

ACE

Patient Sex: Female

Patient Age: 86



Add Filters

Filtering Protocols

Report Filters

Gene Filters

Zygosity

Proband Zygosity

Any

Any

Heterozygous

Homozygous

Location

Quality

Evidence

Allele Frequency

Impact Scores

Variant Effect

RESET FILTERS

APPLY

SNVs/INDELS	PATH/LIKELY PATH	BENIGN/LIKELY BENIGN	VUS/UNCERTAIN i	ALL SVS										
	Gene (Transcript)	Position dbSNP	Change	Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Lates (Date Conf)
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser	None	BAM	Missense	● ○	5461.77 99 389 : 151 : 238	- - 0.00004 0.00002	● ● ●	CV	0.937 <div style="background-color: red; width: 10px; height: 10px; display: inline-block;"></div> 56 37	-	-

Score Variant | MSH2

← 7 of 28 →

Section Effect

Variant is not a LOF variant

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code PVS1
Section EffectSupport Pathogenic
Level Very Strong

Criteria met?

[Skip](#) [Clear](#)

Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING A

RESET FILTERS

Case Info [View More](#)

Accession ID: SFC211110004

Status: Needs Review

Clinical Grade: 0

QC Status: [VIEW FILES](#)

Test: TruSight Cancer panel

Patient Sex: Female

Patient Age: 86

SNVs/INDELS			PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN		ALL SVS						
	Gene (Transcript)	Position dbSNP	Change	Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CAD	CV	
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser	None		Missense		5461.77 99 389 : 151 : 238	- - 0.00004 0.00002			0.937 	56 37		

Score Variant | MSH2

← 7 of 28 →

Section

Variant is not a LOF variant

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code	PVS1	Support	Pathogenic
Section	Effect	Level	Very Strong

Criteria met?

[Skip](#) [Clear](#) Yes

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION

Advanced Biomedical - Production

Production Workspace for Advanced Biomedical

BioGx - Production

Clinical Services Consultants

Clinical Services consultant workspace

First Choice - Production

First Choice for Sonoran Desert Pathology

GeneDx Pilot

Re-analysis pilot 200 cases

Greystone Labs - Production Workspace

Greystone Labs - Production Workspace

Healthy exome

Healthy exome workspace implementing ACMG73 plus CV P/LP

Probably Genetic - Production

Production workspace for Probably Genetic

Research program _ Fabric Internal

Sonoran - Production

Production workspace for Sonoran

Sophia Genetics pilot

[+ Create new workspace](#)

SCORING AUDIT LOG SCORING SHEET

SET CLASSIFICATION

← Accession ID: SFC211110004

SFC211110004 - Production

INTERPRET VARIANTS (12) REPORT REVIEW

SNVS/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN <small>i</small>		ALL SVS													
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF	EVS AF	ExAC AF	gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status		Classification (Condition)	
<input type="checkbox"/>	MSH2 <small>(ENST00000233146)</small>	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None		Missense		5461.77 99 389 : 151 : 238	- 0.00004 0.00002					0.937		56 37	-			Uncertain Significance Lynch Syndrome
													SCORE VARIANT								

Classifi... rows per page: 100 1 - 1 of 1 items < > >>

INTERPRET VARIANTS (12)

REPORT REVIEW

SNVS/INDELS	PATH/LIKELY PATH	BENIGN/LIKELY	
	Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser
Score Variant	MSH2		
← 1 of 28 →	Se		
Allele frequency is >0.05 (5%) in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium			
Code	BA1	Support	Benign
Section	Frequency	Level	Stand Alone
Criteria met?			
<input type="radio"/>	Yes		
<input checked="" type="radio"/>	No		

Scoring Sheet

SUMMARY CRITERIA ANSWERS SCORING AUDIT LOG

Pathogenic Criteria

<u>PM2</u>	Absent from controls (or at extremely low frequency if recessive) in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium	No
<u>PP2</u>	Missense variant in a gene that has a low rate of benign missense variation and in which missense variants are a common mechanism of disease	No
<u>PVS1</u>	Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease	No
<u>PM4</u>	Protein length changes as a result of in-frame in/del in a nonrepeat region or stop-loss variants	No
<u>PM1</u>	Located in a mutational hot spot and/or critical and well-established functional domain without benign variation	No
<u>PS1</u>	Same amino acid change, but different nucleotide change, as a previously established pathogenic variant	No
<u>PM5</u>	Novel missense change at an amino acid residue where a different missense change determined to be pathogenic has been seen before	No
<u>PP3</u>	Multiple lines of computational evidence support a deleterious effect on the gene or gene product	No
<u>PP5</u>	Reputable source recently reports variant as pathogenic, but the evidence is not available to the laboratory to perform an independent evaluation	No
<u>PS2</u>	The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in	No

CLOSE

28 of 28 complete

Latest Classification (Date Classified)	Confirmation Status	Classification (Condition)
-	⋮	Uncertain Significance Lynch Syndrome

SNVs/INDELS			PATH/LIKELY PATH		BENIGN/LIKELY BENIGN			VUS/UNCERTAIN ⓘ		ALL SVS							
	Gene (Transcript)	Position dbSNP	Change	FLAG	Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status	MESSAGE	Classification (Condition)
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None	<input type="checkbox"/> BAM	Missense	● ○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002	● ● ●	cv	0.937	56 37	-	<input type="checkbox"/> :	Uncertain Significance Lynch Syndrome

X Score Variant | MSH2

← 7 of 28 → Section Effect ▾

Variant is not a LOF variant

Null variant (nonsense, frameshift, splice sites, initiation codon, exon deletion) in a gene where LOF is a known mechanism of disease

Caveats

Code PVS1
Section Effect

Support Pathogenic
Level Very Strong

Criteria met?

Skip Clear

Yes
No

28 of 28 complete

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ▾

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION



INTERPRET VARIANTS (12)

REPORT REVIEW



SNVS/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN <small>i</small>		ALL SVS										
<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change		Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status		Classification (Condition)	
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser		None		Missense		5461.77 99 389 : 151 : 238	- 0.00004 0.00002					0.937 56 37	-		Uncertain Significance Lynch Syndrome

X Score Variant | MSH2



← 1 of 28 →

Set Classification

Classification Uncertain Significance	Report Findings Not Reported
Add Note	
CANCEL DONE	

Allele frequency is >0.05 (5%) in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium

Code BA1
Section Frequency

Support Benign
Level Stand Alone

Criteria met?

[Skip](#) [Clear](#) Yes No

28 of 28 complete

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

INTERPRET VARIANTS (12)

REPORT REVIEW



SNVS/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN <small>i</small>		ALL SVS									
	Gene (Transcript)	Position dbSNP	Change	Confirmation Status	Links	Effect	Zygoty	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status	Classification (Condition)		
<input type="checkbox"/>	MSH2 <small>(ENST00000233146)</small>	chr2:47630487 <small>rs755931648</small>	G → T c.157G>T p.Ala53Ser	None	BAM	Missense	● ○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002	● ● ●	CV	0.937 <small>███████</small>	56 37	-			
<input type="checkbox"/>																⋮	

X Score Variant MSH2

← 1 of 28 →

Section Frequency

Allele frequency is >0.05 (5%) in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium

Code	BA1	Support	Benign
Section	Frequency	Level	Stand Alone

Allele Frequency

1KG AF	-
EVS AF	-
ExAC AF	<u>0.00004</u>
gnomAD AF	<u>0.00002</u>

Description

Criteria met?

Skip Clear Yes No

28 of 28 complete

VARIANT DESCRIPTION

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

INTERPRET VARIANTS (12)

REPORT REVIEW



SNVs/INDELS PATH/LIKELY PATH BENIGN/LIKELY

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

X Score Variant MSH2

← 1 of 28 →

Allele frequency is >0.05 (5%) in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium

Code BA1 Support Benign
Section Frequency Level Stand Alone

Criteria met?

 Yes No

28 of 28 complete

CLOSE

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

Scoring Sheet

SUMMARY CRITERIA ANSWERS SCORING AUDIT LOG

- Nov 24, 2021 scoring started
- Nov 24, 2021 Automatically scored as uncertain significance



Latest Classification (Date Classified) Confirmation Status	Classification (Condition)
-	Uncertain Significance Lynch Syndrome

⋮

SNVS/INDELS		PATH/LIKELY PATH		BENIGN/LIKELY BENIGN		VUS/UNCERTAIN 1		ALL SVS									
		Position dbSNP	Change	Confirmation Status	Links	Effect	Zygosity	Quality GQ Coverage	1KG AF EVS AF ExAC AF gnomAD AF	Review Priority	Evidence	Omicia Score	VVP CADD	Latest Classification (Date Classified) Confirmation Status	Classification (Condition)		
<input type="checkbox"/>	Gene (Transcript)																
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser	None	BAM	Missense	● ○	5461.77 99 389 : 151 : 238	- 0.00004 0.00002	● ● ●	CV	0.937	56 37	-		Uncertain Significance Lynch Syndrome	⋮

X Score Variant | MSH2

← 19 of 28 →

Section Literature

NEW CITATION

Cosegregation with disease in multiple affected family members in a gene definitively known to cause the disease ?

Notes

Code PP1 Support Pathogenic
Section Literature Level Strong, Moderate, Supporting

Criteria met?

[Skip](#) [Clear](#)

- Yes Strong
 Moderate
 Supporting
 No

28 of 28 complete

Curated Citations

No citations have been curated

Search for citations on the web


Search
MSH2 ("157G>T" OR "-30-12G>T" OR "Ala53Ser" OR "A53S" OR "rs755931648")

[GOOGLE SCHOLAR](#) [GOOGLE](#)

Citations from Annotated Evidence and Condition Gene

Citation	Title	Match Type	Source
9843200	MSH2 genomic deletions are a frequent cause of HNPCC.	Overlap	ClinVar CURATE
11809679	A homozygous germ-line mutation in the human MSH2 gene predisposes to hematological malignancy and multiple café-au-lait spots.	Gene	Gene Card CURATE

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

INTERPRET VARIANTS (12)

REPORT REVIEW



SNVS/INDELS PATH/LIKELY PATH BENIGN/LIKELY HARMFUL

<input type="checkbox"/>	Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/>	MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

X Score Variant MSH2

← 19 of 28 →

Cosegregation with disease in multiple affected family members, definitively known to cause the disease

Notes

Code PP1
Section Literature Support Level Pathogenic Strong, Moderate

Criteria met?

- Yes Strong Moderate Supporting No

New Curated Study

Study Type

General Study

Phenotype

PubMed ID

Pubmed Article Abstract
Please enter a valid PubMed ID above.

Assessment

Internal PDF URL

[CANCEL](#) [SAVE](#)

Latest Classification (Date Classified)	Confirmation Status	Classification (Condition)
-		Uncertain Significance
-		Lynch Syndrome
		NEW CITATION
		GOOGLE SCHOLAR GOOGLE
		Match Type Source
		Overlap ClinVar CURATE
		Gene Gene Card CURATE

11809679 A homozygous germ-line mutation in the human MSH2 gene predisposes to hematological malignancy and multiple café-au-lait spots.

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ^

SCORING AUDIT LOG

SCORING SHEET

SET CLASSIFICATION

← Accession ID: SFC211110004

INTERPRET VARIANTS (12)

REPORT REVIEW

SNVs/INDELS PATH/LIKELY PATH BENIGN/LIKELY

<input type="checkbox"/> Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/> MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

X Score Variant | MSH2

← 19 of 28 →

Cosegregation with disease in multiple affected family members
definitively known to cause the disease

Notes

Code	PP1	Support Level	Pathogenicity
Section	Literature		Strong, Moderate

Criteria met?

- Yes
- Strong
 - Moderate
 - Supporting
- No

Assigned Class: Uncertain Significance

Inferred Class: Uncertain Significance

VARIANT DESCRIPTION ▾

SCORING AUDIT LOG SCORING SHEET

SET CLASSIFICATION

New Curated Study

Study Type: Cosegregation Study

Phenotype

PubMed ID:

Pedigree Reference:

Pubmed Article Abstract:
Please enter a valid PubMed ID above.

Alternate variant explains phenotype

Number of Affected with Variant	Number of Affected Without Variant
Number of Unaffected with Variant	Number of Unaffected Without Variant

Assessment:

Internal PDF URL:

CANCEL **SAVE**

Latest Classification (Date Classified)	Classification (Condition)
-	Uncertain Significance
NEW CITATION	
GOOGLE SCHOLAR	GOOGLE
Match Type	Source
Overlap	ClinVar
and multiple	CURATE
Gene	Gene Card
	CURATE



INTERPRET VARIANTS (12)

REPORT REVIEW

SNVS/INDELS PATH/LIKELY PATH BENIGN/LIKELY

<input type="checkbox"/> Gene (Transcript)	Position dbSNP	Change
<input type="checkbox"/> MSH2 (ENST00000233146)	chr2:47630487 rs755931648	G → T c.157G>T p.Ala53Ser

Score Variant MSH2

← 19 of 28 →

Cosegregation with disease in multiple affected family members definitely known to cause the disease

Notes

Code PP1 Support Pathogenic
Section Literature Level Strong, Moderate

Criteria met?

- Yes
- Strong
 - Moderate
 - Supporting
- No

New Curated Study

Study Type

Functional Study

Phenotype

PubMed ID

Pubmed Article Abstract
Please enter a valid PubMed ID above.

Experiment Type

Assessment

Alternate variant explains phenotype

Methodology appropriate

Controls Appropriate

?

CANCEL SAVE

- Home
- Case Queue
- Test Management >
- Upload
- Gene Filters
- Projects
- Switch to Opal™
- Help >

	Sample ID	Projects	Test
	Call Data from job 41144 – SFC211110004 (1054762)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41149 – SFC2111130001 (1054752)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41153 – SFC2111140004 (1054750)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41148 – SFC2111120004 (1054745)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41145 – SFC2111120001 (1054757)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41146 – SFC2111120002 (1054748)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41143 – SFC2111110003 (1054746)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41147 – SFC2111120003 (1054753)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41152 – SFC2111140003 (1054751)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41150 – SFC2111140001 (1054747)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE
	Call Data from job 41155 – SFC2111140006 (1054761)	11-20-2021-05-08	TruSight Cancer panel - CGX ACE



Select Project* ▼

NEW PROJECT

Choose Variant file* ✖

Genome Label* ⓘ

External ID ⓘ

Family ID ⓘ

Type to filter (at least 3 characters)

Sex* ▼

Confirm Assembly Version

GRCh37/hg19* ⓘ

Your workspace PO account will be billed according to your contract terms. Please contact support@fabricgenomics.com if you have any questions regarding the upload or billing process.

RESET UPLOAD

Upload Guidelines

Supported File Formats

- Compression: gzip, bzip2
- Variants: VCF 4.0 and above, samples must provide a GT field

Multi-sample VCFs

- Multi-sample VCF files will be split into separate genome records and annotated individually

Required Fields

- Choose or create a Project
- Choose a VCF file (use compression)
- Specify a label, sex and confirm the assembly version.

153,295,750 bp

153,296,000 bp

153,296,250 bp

153,296,500 bp

153,296,750 bp

153,297,000 bp

Nucleotides

MECP2: NM_004992.3

c.792 c.744

Nucleotides

MECP2: NM_001316337.1

c.1104

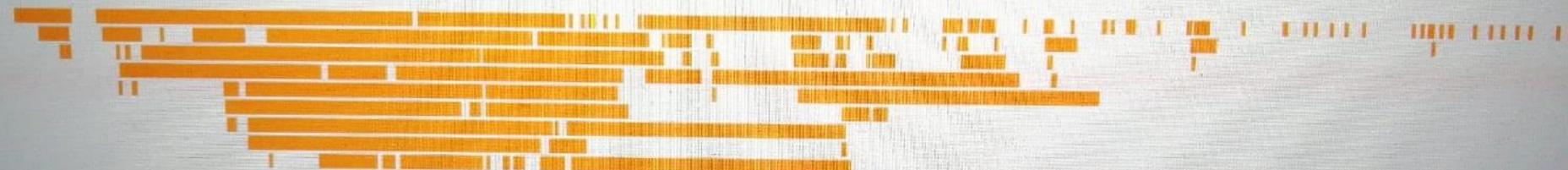
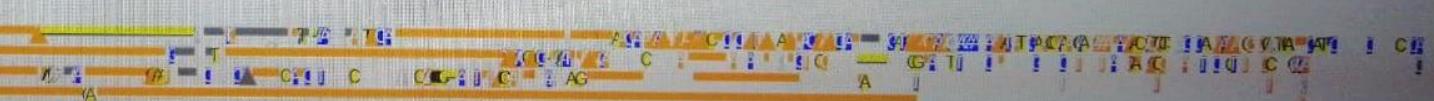
c.600

MECP2: NM_001110792.1

p.210 p.200

c.828 c.780

p.260

HGMD del/delins**HGMD subs****HGMD insert****ClinVar**

G

IGV

hg19

chrX

chrX:153,295,636-153,297,080



Query genome

1,445 bp

MECP2:All

Chr Cytoband

Cursor Guide

Center Line

Track Labels

HGMD insert

ClinVar

G

CentoMD

nt conservation (MECP2)

Splicing Pred. (MECP2)

Protein Domain (MECP2)



INTERPRET VARIANTS (12)

REPORT REVIEW



TEST DETAILS

CASE DETAILS

SAVE

PREVIEW PDF

SET STATUS

Test Name

TruSight Cancer panel

Result

/

Primary Finding

No primary finding for this report.

Secondary Finding

No secondary finding for this report.

Other Finding

No other finding for this report.

Not Reportable

No not reportable for this report.

Variant Section 5



Accession ID: SFC2111110004

Sonoran - Production



INTERPRET VARIANTS (12)

REPORT REVIEW



TEST DETAILS

CASE DETAILS

SAVE PREVIEW PDF

SET STATUS

Recommendations



Regulatory Disclosure



Exceptions



Limitations



References

Lab Contact

