Module: molecular

Module Contents

msi
1. <u>CENTER_NO</u>
2. <u>PERSON_ID</u>
3. <u>MSI_TEST_LAB</u>
4.COLLECTION CID
5.TEST TISSUE
6.TUMOR NO
7. POLYP NO
8.BLOCK SPEC CID
9.NUC ACID CID TUMOR
10.NUC ACID CID NORMAL
11.METASTASIS
12.MSI DETECTION METHOD
13.MSI ACTC
14.MSI BAT25
15.MSI BAT26
16.MSI BAT34C4
17.MSI BAT40
18.MSI D5S346
19.MSI D10S197
20.MSI D17S250
21.MSI D18S55
22.MSI D2S123
23.MSI MYCL

1	CENTER_	NO		number (2,0)	Required: true
	Center id	entific	cation number.		
		Allo	wable Values		
		11	Sinai Health Systems (formerly Cancer Care Ontario)		
		12	Cedars-Sinai & Cleveland Clinic (formerly USC Consortium)		
		13	University of Melbourne		
		14	University of Hawaii Cancer Center		
		15	Mayo Clinic		
		16	Fred Hutch, Seattle		
		17	UCSF: University of California at San Franscisco (formerly CPIC, originally N	orthern California (N	((222
				1	
2	PERSON	_ID		string (12)	Required: true
	Number t	hat U	niquely Identifies an Individual.		
2	MSI_TES	T_L	AB	number (1,0)	Required: true
5	Code for	type o	of lab that did the MSI testing.		
			Allowable Values		
			1 CFR Lab		
			2 Clinical Lab		
	COLLECT	ION	_CID	string (30)	Required: true
4	Identifier	used	internally by centers to denote a unique surgical event.		

_	TEST	TISSU	E			number (1,0)	Required: true
5	Tissu	ie on whi	ch test was performed.				
				All	lowable Values		
				1	Cancer		
				2	Contiguous (Adjacent) adenoma		
				3	Colon Polyp		
				4	Gastric polyp		
				5	Sebaceous adenoma		
6	TUM	OR_NO				number (2,0)	Required: true
	Numb	bers labe	ling tumors of the individu	ual. Numb	pers are not necessarily sequential.		
		Allowa	ble Values				
		1 to 9	9 Range				
		-	9 NA/Out of scope: Tissu	ue does n	ot currently have a TUMOR_NO and is	not cancer or contiguous	adenoma
		-1	5 Information Unknown				
	POLY	YP_NO				number (2,0)	Required: true
7	Sequ		mber over range of 1 to 9	9 to disti	nguish a polyp removed on a particula	r date. The tuple PERSON	_ID,
	Allowab	ole Value	S				
	1 to -	99 or -9, -1	Range				
ļĻ		-9	NA/Out of scope. Tissue is	not a pol	ур.		
		-1	Polyp has IHC/MSI result to this information is currentle	out center y underw	r is currently unable to locate polyp pa ay.	thology information. Cent	er review to obtain
Q	BLOO	CK_SPE	C_CID			string (40)	Required: true
0	Uniqu	ue local i	dentifier used at a center	to unique	ly identify a block tissue specimen.		
	NUC_	_ACID_				string (40)	Required: true
9	Cente	er's uniqu	ue identifier for the tumor	DNA use	d in this MSI test. Each DNA extractior	n has a unique identifier.	
	NUC	C_ACID	_CID_NORMAL			string (40)	Required: true
10	Cen	iter's unio	que identifier for the norm	al DNA u	sed in this MSI test. Each DNA extract	ion has a unique identifier	
	MET	TASTAS	IS			number (1,0)	Required: true
1	1 Prim	nary or n	netastatic tumor.				
				Allowab	le Values		
				1 Pri	mary		
				2	tastatasis		

10	MSI_DETECTION_METHOD			number (2,0)	Required: true
12	MSI Testing method.				
		Allow	vable Values		
		1	PCR non-radiolabeled		
		2	PCR radiolabeled		
		3	Fluorescent PCR capillary electrophoresis		
		4	Fluorescent PCR slab gel electrophoresis		
		5	Silver-stained gel electrophoresis		
		-13	Not tested: Quantity of DNA or tissue not sufficient	t	
		-15	Method not specified		

10	MSI_ACTC		number (2,0)	Required: true
13	MSI test result at ACTC locus			
	Allo	wable Values		
	1	Stable		
	2	Unstable		
	6	Equivocal (Inconclusive)		
	7	Stable - Normal DNA not used in test		
	8	Unstable - Normal DNA not used in test		
	9	Equivocal (Inconclusive) - Normal DNA not used in te	est	
	-11	No Amplification		
	-12	Not tested, reason not specified		
	-13	Quantity of DNA or tissue not sufficient		
	-14	Tested, outcome not specified		
	-15	Unknown if tested		

14	MSI_BAT25			number (2,0)	Required: true
14	MSI test result at BAT25 locus				
		Allov	vable Values		
		1	Stable		
		2	Unstable		
		6	Equivocal (Inconclusive)		
		7	Stable - Normal DNA not used in test		
		8	Unstable - Normal DNA not used in test		
		9	Equivocal (Inconclusive) - Normal DNA not used in te	est	

-11	No Amplification
-12	Not tested, reason not specified
-13	Quantity of DNA or tissue not sufficient
-14	Tested, outcome not specified
-15	Unknown if tested

1	MSI_BAT26			number (2,0)	Required: true
15	MSI test result at BAT26 locus				
	A	Allow	able Values		
		1	Stable		
	Γ	2	Unstable		
		6	Equivocal (Inconclusive)		
		7	Stable - Normal DNA not used in test		
		8	Unstable - Normal DNA not used in test		
		9	Equivocal (Inconclusive) - Normal DNA not used in te	est	
	-	-11	No Amplification		
	-	-12	Not tested, reason not specified		
	-	-13	Quantity of DNA or tissue not sufficient		
	-	-14	Tested, outcome not specified		
	-	-15	Unknown if tested		

MSI_BAT34C4 number (2,0) Required: true 16 MSI test result at BATC34C4 locus Allowable Values 1 Stable 2 Unstable Equivocal (Inconclusive) 6 7 Stable - Normal DNA not used in test Unstable - Normal DNA not used in test 8 9 Equivocal (Inconclusive) - Normal DNA not used in test -11 No Amplification -12 Not tested, reason not specified -13 Quantity of DNA or tissue not sufficient Tested, outcome not specified -14 -15 Unknown if tested

Required: true

17					
	MSI test result at BA140 locus				
		Allow	vable Values		
		1	Stable		
		2	Unstable		
		6	Equivocal (Inconclusive)		
		7	Stable - Normal DNA not used in test		
		8	Unstable - Normal DNA not used in test		
		9	Equivocal (Inconclusive) - Normal DNA not used in tes	st	
		-11	No Amplification		
		-12	Not tested, reason not specified		
		-13	Quantity of DNA or tissue not sufficient		
		-14	Tested, outcome not specified		
		-15	Unknown if tested		

10	MSI_D5S346			number (2,0)	Required: true
10	MSI test result at D5S346 locus				
		Allow	vable Values		
		1	Stable		
		2	Unstable		
		6	Equivocal (Inconclusive)		
		7	Stable - Normal DNA not used in test		
		8	Unstable - Normal DNA not used in test		
		9	Equivocal (Inconclusive) - Normal DNA not used in t	est	
		-11	No Amplification		
		-12	Not tested, reason not specified		
		-13	Quantity of DNA or tissue not sufficient		

		-15	Unknown if tested		
	MSI_D10S197			number (2,0)	Required: true
19	MSI test result at D10S197 loc	us			
		Allov	vable Values		
		1	Stable		
		2	Unstable		
		6	Equivocal (Inconclusive)		
		7	Stable - Normal DNA not used in test		

Tested, outcome not specified

-14

8	Unstable - Normal DNA not used in test
9	Equivocal (Inconclusive) - Normal DNA not used in test
-11	No Amplification
-12	Not tested, reason not specified
-13	Quantity of DNA or tissue not sufficient
-14	Tested, outcome not specified
-15	Unknown if tested

MSI_D17S250 20

Required: true

number (2,0)

MSI test result at D17S250 locus

Allow	able Values				
1	Stable				
2	Unstable				
6	Equivocal (Inconclusive)				
7	Stable - Normal DNA not used in test				
8	Unstable - Normal DNA not used in test				
9	Equivocal (Inconclusive) - Normal DNA not used in test				
-11	No Amplification				
-12	Not tested, reason not specified				
-13	Quantity of DNA or tissue not sufficient				
-14	Tested, outcome not specified				
-15	Unknown if tested				

Required: true MSI_D18S55 number (2,0) 21 MSI test result at D18S55 locus Allowable Values 1 Stable 2 Unstable t

_	
6	Equivocal (Inconclusive)
7	Stable - Normal DNA not used in test
8	Unstable - Normal DNA not used in test
9	Equivocal (Inconclusive) - Normal DNA not used in test
-11	No Amplification
-12	Not tested, reason not specified
-13	Quantity of DNA or tissue not sufficient
-14	Tested, outcome not specified
-15	Unknown if tested

22	MSI_D2S123			number (2,0)	Required: true		
22	MSI test result at D2S123 locus	SI test result at D2S123 locus					
		Allow	vable Values				
		1	Stable				
	[2	Unstable				
		6	Equivocal (Inconclusive)				
		7	Stable - Normal DNA not used in test				
		8	Unstable - Normal DNA not used in test				
		9	Equivocal (Inconclusive) - Normal DNA not used in te	est			
		-11	No Amplification				
	[-12	Not tested, reason not specified				
	[-13	Quantity of DNA or tissue not sufficient				
	[-14	Tested, outcome not specified				
	[-15	Unknown if tested				

22	MSI_MYCL		number (2,0)	Required: true
23	MSI test result at MyCL locus			
	Allo	wable Values		
	1	Stable		
	2	Unstable		
	6	Equivocal (Inconclusive)		
	7	Stable - Normal DNA not used in test		
	8	Unstable - Normal DNA not used in test		
	9	Equivocal (Inconclusive) - Normal DNA not used in t	est	
	-11	No Amplification		
	-12	Not tested, reason not specified		
	-13	Quantity of DNA or tissue not sufficient		
	-14	Tested, outcome not specified		
	-15	Unknown if tested		

Module: molecular

Module Contents

ihc 1.<u>CENTER_NO</u> 2.<u>PERSON_ID</u> 3.<u>IHC_TEST_LAB</u> 4.<u>COLLECTION_CID</u> 5.<u>TEST_TISSUE</u> 6.<u>TUMOR_NO</u> 7.<u>POLYP_NO</u> 8.<u>BLOCK_SPEC_CID</u> 9.<u>METASTASIS</u> 10.<u>IHC_MLH1</u> 11.<u>IHC_MSH2</u> 12.<u>IHC_MSH6}</u> 13.<u>IHC_PMS2</u>

1	CENTER_	_NO		number (2,0)	Required: true				
1	Center identification number.								
		Allo	vable Values						
		11	Sinai Health Systems (formerly Cancer Care Ontario)						
		12	Cedars-Sinai & Cleveland Clinic (formerly USC Consortium)						
		13	University of Melbourne						
		14	University of Hawaii Cancer Center						
		15	Mayo Clinic						
		16	Fred Hutch, Seattle						
		17	UCSF: University of California at San Franscisco (formerly CPIC, originally No	orthern California (N	CCC))				

2	PERSON_ID string (12) Required: true							
2	Number that Uniquely Identifies an Individua	Number that Uniquely Identifies an Individual.						
	IHC_TEST_LAB			number (1,0)	Required: true			
3	Lab that conducted the results							
			Allowable Values1CFR lab2Clinical lab					
	COLLECTION_CID			string (40)	Required: true			
ŀ	The label assigned by the center to all tissue associated with a single surgical event - resection or biopsy/endoscopy procedure.							
	TEST_TISSUE			number (1,0)	Required: true			
2	Type of tissue tested.							
		All	owable Values					
		1	Cancer					
		2	Contiguous (Adjacent) adenoma					

3	Colon Polyp
4	Gastric polyp
5	Sebaceous adenoma

_	TUMOR_NO)				number (2,0)	Required: true					
0	Numbers lab	peling tum	nors of the individu	ial. N	lumbers are not necessarily sequential.							
Allowable Values												
	1 to 99 or											
		77 10										
		-9	NA/Out of scope:	Tiss	ue does not currently have a TUMOR_NO and	is not cancer or conti	guous adenoma					
		-15	Information Unkn	nown								
	POLYP_NO					number (2,0)	Required: true					
7	Sequential r DATE_COLL	number ov ECTED, ar	ver range of 1 to 99 nd POLYP_NO uniqu	9 to uely	distinguish a polyp removed on a particular da identify a physical polyp globally within the C	ate. The tuple PERSON FR database.	N_ID,					
A	llowable Valu	les										
	1 to 99 or	Range										
	-1, -9											
	-9	NA/Out	of scope. Tissue is	not a	a polyp.							
	-1 Polyp has IHC/MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway.						ter review to obtain					
Q	BLOCK_SP	EC_CID				string (40)	Required: true					
8	BLOCK_SPI	EC_CID identifier	used at a center to	o un	iquely identify a block tissue specimen.	string (40)	Required: true					
8	BLOCK_SPI	EC_CID identifier	used at a center to	o un	iquely identify a block tissue specimen.	string (40) number (1,0)	Required: true					
8 9	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic	used at a center to	o un	iquely identify a block tissue specimen.	string (40) number (1,0)	Required: true					
8	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic	used at a center to tumor.	o un	iquely identify a block tissue specimen.	string (40) number (1,0)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic	used at a center to tumor.	o un Allo	iquely identify a block tissue specimen. wable Values	string (40) number (1,0)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic	used at a center to	Allo	iquely identify a block tissue specimen. wable Values Primary Metastatasis	string (40)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic	used at a center to	Allo	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40) number (1,0)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic	used at a center to	Allc 1 2	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic	used at a center to	Allc 1 2 -9	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40) number (1,0) number (2,0)	Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or n	EC_CID identifier IS netastatic 1 for MSI te	used at a center to tumor.	Allc 1 2 -9	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40) number (1,0) number (2,0)	Required: true Required: true Required: true Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic 1 for MSI te	tumor.	Allc 1 2 -9	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40) number (1,0) number (2,0)	Required: true Required: true Required: true Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or r	IS Inetastatic	est at locus MLH1 wable Values	Alla 1 2 -9	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue	string (40) number (1,0) number (2,0)	Required: true Required: true Required: true Required: true					
8 9 10	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic 1 for MSI te Allo 0	tumor. est at locus MLH1 wable Values Negative - Express	Alla 1 2 -9	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue n absent and abnormal present and normal	string (40) number (1,0) number (2,0)	Required: true Required: true Required: true Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic for MSI te Allo 0 1	used at a center to tumor. est at locus MLH1 wable Values Negative - Expres Positive - Techni	Allco Allco -9 esssion sssion	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue n absent and abnormal present and normal proc. probably positive	string (40) number (1,0) number (2,0)	Required: true Required: true Required: true					
9	BLOCK_SPI Unique local METASTAS Primary or r	EC_CID identifier IS netastatic 1 for MSI te Allo 1 2	used at a center to tumor. est at locus MLH1 wable Values Negative - Expres Positive - Techni Positive - Techni	Allc 1 2 -9 ession ically	iquely identify a block tissue specimen. wable Values Primary Metastatasis Test is not performed on cancerous tissue n absent and abnormal present and normal poor, probably positive (equivocal, pot truly interpretable but if force	d would be positive	Required: true Required: true Required: true					

4	Negative - Technically poor, probably negative
5	Negative - Technically equivocal, not truly interpretable but if forced would be negative
7	Heterogeneous tumor - some areas lack expression and some have expression
8	Reduced - expression reduced but not absent
-11	Technical failure
-12	Not tested
-13	Quantity of tissue not sufficient

IHC_MSH2

11

Required: true

IHC result for MSI test at locus MSH2

Allow	able Values
0	Negative - Expression absent and abnormal
1	Positive - Expression present and normal
2	Positive - Technically poor, probably positive
3	Positive - Technically equivocal, not truly interpretable but if forced would be positive
4	Negative - Technically poor, probably negative
5	Negative - Technically equivocal, not truly interpretable but if forced would be negative
7	Heterogeneous tumor - some areas lack expression and some have expression
8	Reduced - expression reduced but not absent
-11	Technical failure
-12	Not tested
-13	Quantity of tissue not sufficient

IHC_MSH6

12

number (2,0)

number (2,0)

Required: true

IHC result for MSI test at locus MSH6

Allow	vable Values
0	Negative - Expression absent and abnormal
1	Positive - Expression present and normal
2	Positive - Technically poor, probably positive
3	Positive - Technically equivocal, not truly interpretable but if forced would be positive
4	Negative - Technically poor, probably negative
5	Negative - Technically equivocal, not truly interpretable but if forced would be negative
7	Heterogeneous tumor - some areas lack expression and some have expression
8	Reduced - expression reduced but not absent
-11	Technical failure
-12	Not tested
-13	Quantity of tissue not sufficient

IHC_PMS2

13

number (2,0)

Required: true

IHC result for MSI test at locus PMS2

Allow	vable Values
0	Negative - Expression absent and abnormal
1	Positive - Expression present and normal
2	Positive - Technically poor, probably positive
3	Positive - Technically equivocal, not truly interpretable but if forced would be positive
4	Negative - Technically poor, probably negative
5	Negative - Technically equivocal, not truly interpretable but if forced would be negative
7	Heterogeneous tumor - some areas lack expression and some have expression
8	Reduced - expression reduced but not absent
-11	Technical failure
-12	Not tested
-13	Quantity of tissue not sufficient

Module: molecular

Module Contents

mlpa 1.<u>CENTER_NO (*PK)</u> 2.<u>PERSON_ID</u> 3.<u>GENE</u> 4.<u>NUC_ACID_CID (*PK)</u> 5.<u>MLPA_DATE_TESTED</u> 6.<u>MLPA_INVEST_ID</u> 7.<u>MLPA_DETECTION_METHOD</u> 8.<u>MLPA_RESULT</u> 9.<u>MLPA_RESULT_SUB</u> 10.<u>MLPA_EXON_FROM</u> 11.<u>MLPA_EXON_FROM</u> 11.<u>MLPA_EXON_TO</u> 12.<u>MLPA_OTHER_TEST_COMMENT</u> 13.<u>GENOMIC_REGION_TESTED</u> 14.<u>VARIANT_NAME</u>

-	CENTER_	number (2,0)	Required: true						
Center identification number. *CENTER_NO & NUC_ACID CID are the primary key for the table.									
Allowable Values									
11 Sinai Health Systems (formerly Cancer Care Ontario)									
		12	Cedars-Sinai & Cleveland Clinic (former	ly USC Consortiu	m)				
		13	University of Melbourne						
		14	University of Hawaii Cancer Center						
		15	Mayo Clinic						
		16	Fred Hutch, Seattle						
		17	UCSF: University of California at San Fr	anscisco (former	y CPIC, originally No	orthern California (N	ICCC))		
2	2 PERSON_ID Required						Required: true		
Number that Uniquely Identifies an Individual.									
3	GENE					string (20)	Required: true		
	Gene on	which	testing has been performed.						
				Allowable Values	5				
				BRCA1					
				BRCA2					
				MLH1					
				MSH2					
				MSH6					
				PMS2					
				EPCAM					
	NUC_AC	ID_C	ID (*PK)			string (40)	Required: true		
4	I -I + ! <i>C</i> !								

Identifier used internally by centers for a nucleic acid sample from a single extraction. *CENTER_NO & NUC_ACID_CID are the primary key for the table.

_							
	5	MLPA_DATE_TESTED				string (8)	Required: true
		Date of test.					
			1700		nimum year		
	6	MLPA_INVEST_ID				number (1,0)	Required: true
		Laboratory investigator conducting th	ne tests or	n sar	nples.		
				1 2 3 4 5 6	Dr. Steve Thibodeau Dr. Melissa Southey GMP Genetics (Corporation) Joanne Young Clinical Laboratory Dan Buchanan		
	7	MLPA_DETECTION_METHOD				number (1,0)	Required: false
	1	Testing method.					
		All 1 2	lowable Va Multiple> Conversi	alues (liga ion a	s ation-dependent probe amplification (MLI analysis	PA)	
		MLPA_RESULT number (1,0)					
	8	Summary of test findings.					
			All 0 1 2	owa Fai Ne De	ble Values iled test gative (no deletion or duplication) letion		

3 Duplication
4 Equivocal
5 Inversion

0	М	LPA_RESULT_SUBnumber (2,0)Required: false								
9	Ac	ditional specific find	findings of test.							
A	Allowable Values									
	0	0 Failed test								
	1	Confirmed by secon	nd MLPA test only							
	2	Confirmed by South	nern blot							
	3	Confirmatory test n	ot done for positive result							
	4	Confirmed by other	method or other information							
	5	Deletion artifact. DI	NA sequence change under primer found by sequencing (scored as ne	gative)						
	6	Deletion artifact. Re	esults not consistent, normal on repeat (scored as negative)							
	7	Deletion artifact. So	outhern did not confirm deletion							
	8	Deletion artifact. Se based on analysis o	equencing and/or Southern not done to determine cause for discordan f other similar cases)	t result (assumed to	be negative					
	9	Deletion artifact, Of	ther							
1	0	Duplication artifact.	Results not consistent, normal on MLPA repeat (scored as negative)							
1	1	Duplication artifact.	Southern did not confirm duplication							
1	12	Duplication artifact. based on analysis o	Sequencing and/or Southern not done to determine cause for discord for the similar cases)	lant result (assumed	to be negative					
1	13	Duplication artifact,	Other							
1	4	Equivocal. Single ex	kon deletion, unclear whether artifact; Southern could not be performe	ed						
1	15	Equivocal. Multiple	exons deleted, not enough sample to repeat test							
1	6	Equivocal. Single ex	kon duplication, unclear whether artifact; Southern could not be performed	rmed						
1	17	Equivocal. Multiple	exons duplicated, not enough sample to repeat test							
	-7	NA One assay only	for negative result							
			Error Description							
			If MIPA_RESULT = 0 then MIPA_RESULT_SUB = 0							
			If MIPA_RESULT=1 then MIPA_RESULT_SUB_must be in $(-7.1.2)$ or i	in range 4-13						
			If MIDA_RESULT is 2 or 3, then MIDA_RESULT_SUB_must be in range	= 1_1						

If MLPA_RESULT=4, then MLPA_RESULT_SUB must be in range 14-17

10 MLPA_EXON_FROM

number (10,0)

Required: false

Starting exon in range.

Allowable Values

1 to 999999999 or -7 Range

			-7	NA: Negative result or failed test Otherwise num	nber of starting exon	
		Γ		Error Description		
	If MLPA_RESULT is in (0,1), then MLPA_EXON_FROM must be -7 If MLPA_RESULT is in (2,3,4), then MLPA_EXON_FROM must not be -7					
			If GENE = ML	H1 then MLPA_EXON_FROM must be -7 or in ran	ge 1-19	
			If GENE = MS	H2 then MLPA_EXON_FROM must be -7 or in ran	ige 1-16	
			If GENE = MS	SH6 then MLPA_EXON_FROM must be -7 or in ran	ge 1-10	
			If GENE = PM	IS2 then MLPA_EXON_FROM must be -7 or in ran	ge 1-15	
	MLPA_EXON	_то			number (10,0)	Required: false
11	Ending exon i	n range.				1
		Allowable Val	ues			
	1 to 999999999 or -7 Range					-
			-7	NA: Negative result or failed test Otherwise num	nber of starting exon	
				Error Description		
			If MLPA_RE	SULT is 0 or 1, then MLPA_EXON_TO must be -7		
			If MLPA_RE	SULT is in (2,3,4), then MLPA_EXON_TO must no	t be -7	
			If GENE = N	/ILH1 then MLPA_EXON_TO must be -7 or in rang	e 1-19	
			If $GENE = N$	ISH2 then MLPA_EXON_TO must be -7 or in rang	e 1-16	
			If $GENE = N$	ISH6 then MLPA_EXON_TO must be -7 or in rang	e 1-10	
			If GENE = P	MS2 then MLPA_EXON_TO must be -7 or in rang	e 1-15	
	MLPA_OTHE	R_TEST_COM	IMENT		string (400)	Required: false
12	Text field con	taining comme	nts regarding	other test findings.		
	GENOMIC_R	EGION_TEST	ED		string (100)	Required: false
13	Indicates what	t part(s) of the	e gene or othe	er genomic region(s) was tested.		
	VARIANT_N	AME			string (100)	Required: false
14	Proper varian	t name using F	IGVS nomenc	lature.		

Module: molecular

Module Contents

genomics 1. CENTER NO (*PK) 2.PERSON ID 3. GENOMIC CID (*PK) 4.<u>GENE</u> 5. VARIANT ORIGIN 6. SOURCE NUC ACID 7.NUC ACID CID 8. DETECTION METHOD 9. GENOMIC REGION TESTED 10.<u>TEST_RESULT</u> 11. VARIANT NAME RESULT 12.<u>EXON NO</u> 13. INTRON NO 14. OTHER REGION 15.NUC POS START 16.NUC POS END 17. NUCLEOTIDE FROM 18. NUCLEOTIDE TO 19. DELETE SEQ 20.INSERT SEQ 21. TYPE VARIANT 22.<u>CODON</u> 23.<u>AA FROM</u> 24.<u>AA TO</u> 25. CONSEQUENCE 26. ZYGOSITY 27. SEVERITY 28. INSIGHT SEVERITY 29.<u>LAB ID</u> 30.LAB OTHER TEXT 31.DATE TEST

	CENTER_	NO	number (2,0)	Required: true						
	Center identification number. * CENTER_NO & GENOMIC_CID are the primary key for the table.									
Allowable Values										
		11 Sinai Health Systems (formerly Cancer Care Ontario) 12 Cedars-Sinai & Cleveland Clinic (formerly USC Consortium)								
		13 University of Melbourne 14 University of Hawaii Cancer Center								
16 Fred Hutch, Seattle										
		17	UCSF: University of California at San Franscisco (formerly CPIC, originally Northern California (NCCC))							
_										
,	PERSON_	RSON_ID string (12) Requi								
•	Number th	nat U	niquely Identifies an Individual.							
GENOMIC_CID (*PK) string (40)					Required: true					
)	Center identifier for the test. *CENTER_NO & GENOMIC_CID are the primary key for the table.									
	GENE			number (1,0)	Required: true					
÷	Gene on v	vhich	testing has been performed.							

			Allow	able Values			
			1	MLH1			
			2	MSH2			
			3	MSH6			
			4	MUTYH			
			5	PMS2			
			8	EPCAM			
			10	APC			
			11	TP53			
VARIANT_ORIGI	N					number (1,0)	Required: true
Describes if the var	iant was tested as ar	n acquire	d or a germli	ine variant.			
			Allowab	le Values			
			1 Pres	sumed somatic			
			2 Knov	wn somatic			
			3 Gerr	mline			
			Error	Description			
	If GENE is in (6.7)	AND SOU	IRCE NUC A	C(D) = i = (7, 0) +			
			NCL_NOC_A	CID IS IN (7,8), t			
	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is in (7,8), t	B), then VAR	ANT_ORIGIN = 3	
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is in (7,8), t	3), then VAR	ANT_ORIGIN = 3	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is not in (7,8), t	3), then VAR	IANT_ORIGIN = 3	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is not in (7,8), t	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is not in (7,8), t	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU	IRCE_NUC_A	CID is not in (7,8), t CID is not in (7,8	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU AND SOU Allowat 1 bloc 2 Mou 3 Ivm	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal	CID is in (7,8), t CID is not in (7,4 CID is not in (7,4 cells	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU AND SOU Allowat 1 bloc 2 Mou 3 lym 4 nor	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal uphoblastoid mal fresh frc	CID is in (7,8), t CID is not in (7,8 CID is not in (7,8 iva cells ozen tissue	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) if GEN	AND SOU AND SOU Allowat Allowat Doc Mou 3 lym 4 nor 5 oth	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal uphoblastoid mal fresh frc er non-tumo	CID is in (7,8), t CID is not in (7,8 iva cells ozen tissue ur tissue	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7)	AND SOU AND SOU Allowat Allowat Allowat Mou Allowat Mou Allowat Allowa	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal uphoblastoid mal fresh fro er non-tumo yp	CID is in (7,8), t CID is not in (7,8 iva cells ozen tissue ur tissue	3), then VAR	ANT_ORIGIN = 1	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) if If GENE is in (6,7) if tissue used for testin	AND SOU AND SOU Allowat Allowat Allowat Doc Mou 3 lym 4 nor 5 oth 6 poly 7 tum	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal uphoblastoid mal fresh fro er non-tumo yp nour (paraffir	CID is in (7,8), t CID is not in (7,8 iva cells ozen tissue ur tissue n embedded tum	a), then VAR	ANT_ORIGIN = 1 number (1,0)	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) if If GENE is in (6,7) if tissue used for testin	AND SOU AND SOU Allowat 1 bloo 2 Mou 3 lym 4 nor 5 oth 6 poly 7 tur 8 tur	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal uphoblastoid mal fresh fro er non-tumo yp nour (paraffir nor- fresh fro	CID is in (7,8), t CID is not in (7,8 iva cells ozen tissue ur tissue n embedded tum ozen	our- PET)	ANT_ORIGIN = 1 number (1,0)	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) i If GENE is in (6,7) i tissue used for testir	AND SOU AND SOU Allowat 1 bloo 2 Mou 3 lym 4 nor 5 oth 6 poly 7 tur 8 tur 9 unk	IRCE_NUC_A IRCE_NUC_A Dele Values od uth wash/sal nphoblastoid mal fresh fro er non-tumo yp nour (paraffir nour (paraffir nour fresh fro cnown/privat	iva cells ozen tissue ur tissue n embedded tum ozen e lab	our- PET)	ANT_ORIGIN = 1 number (1,0)	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) i If GENE is in (6,7) i tissue used for testin	AND SOU AND SOU Allowat 1 bloo 2 Mou 3 lym 4 nor 5 oth 6 poly 7 tur 8 tur 9 unk	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal nphoblastoid mal fresh fro er non-tumo yp nour (paraffir nour (paraffir nor- fresh fro	iva cells ozen tissue ur tissue n embedded tum ozen e lab	a), then VAR	ANT_ORIGIN = 1 number (1,0)	Required: true
SOURCE_NUC_AC	If GENE is in (6,7) i If GENE is in (6,7) i tissue used for testir	AND SOU AND SOU Allowat Allowat Down Allowat Al	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal phoblastoid mal fresh fro er non-tumo yp nour (paraffir nour (paraffir	iva cells ozen tissue ur tissue n embedded tum ozen e lab	our- PET)	ANT_ORIGIN = 1 number (1,0)	Required: true
SOURCE_NUC_AC Specifies source of NUC_ACID_CID Identifier used inter	If GENE is in (6,7)	AND SOU AND SOU Allowat Allowa	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal phoblastoid mal fresh fro er non-tumo yp nour (paraffir nor- fresh fro snown/privat	iva cells ozen tissue ur tissue n embedded tum ozen e lab	as carried o	ANT_ORIGIN = 1 ANT_ORIGIN = 3 number (1,0)	Required: true
SOURCE_NUC_AC Specifies source of NUC_ACID_CID Identifier used inter	If GENE is in (6,7)	AND SOU AND SOU Allowat 1 blou 2 Mou 3 lym 4 nor 5 oth 6 poly 7 tum 8 tum 9 unk a single e	IRCE_NUC_A IRCE_NUC_A ole Values od uth wash/sal ophoblastoid mal fresh fro er non-tumo yp nour (paraffir nor- fresh fro cnown/privat	CID is in (7,8), t CID is not in (7,8 CID is not in (7,8 cells bzen tissue ur tissue n embedded tum bzen e lab which the test v	as carried or	ANT_ORIGIN = 1 ANT_ORIGIN = 3 number (1,0) string (12)	Required: true

	DETECTION_METHOD			number (2,0)	Required: true
8	Germline or somatic DNA test	ALDI-TOF)			
		Allov	wable Values		
	[1	DNA Sequencing		
	[10	DHPLC (Denaturing High Performance Liquid Chromatog	graphy)	
	[11	Phase 1 protocol DHPLC+SEQ (MLH1 and MSH2)		
	[12	12 SNP panel genotyping by Mass Spec		
	[13 12 SNP panel genotyping, dHPLC with sequencing (MUTYH)			
	[14	9 SNP panel genotyping by Mass Spec		
	[15	9 SNP panel genotyping with sequencing (MUTYH)		
		16	PTT		
		17	SSCP		
		18	Fluorescent ASP (BRAF)		
		19	HRM with sequencing (KRAS)		
		33	Sequencing from Conversion Analysis		
		50	whole exome sequencing		
		60	whole genome sequencing		
		70	targeted panel sequencing		
		99	Unknown/private genetic test results		

_	GENOMIC_REGION_TESTED string (100) Required: true							
9	ndicates what part(s) of the gene or specific mutation or other genomic region(s) was TESTED							
10	TEST_RESULT	number (1,0)	Required: true					
	Result of the test carried out and this is at the test level (e.g. entire gene sequencing or MSH2 exon 2 etc.). This is NOT at person level or at variant level.							

Allowable Values						
1	Change detected					
2	No change detected					
3	Test failed					
9	Equivocal					

	Proper va	riant name using Human Genomic Variant Society (HG	VS) nomenclature.	
1	VARIAN	T_NAME_RESULT	string (100)	Required: false

If TEST_RESULT is not 1 or 9 then VARIANT_NAME_RESULT must be null

	EXON_NO	number (2,0)	Required: false					
12	Number of exon containing variant. In the case the variant spans more than one exon, the affected by the variant.	en this field should in	dicate the first exon					
	Allowable Values 1 to 99 Range							
	Error Description							
lf	TEST_RESULT is not 1 or 9 and DETECTION_METHOD is not 10, then EXON_NO must be nu	11						
lf	DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then EXON_NO must be null							
lf	DETECTION_METHOD = 10 AND CENTER_NO = 11 then must EXON_NO be null							
lf N	INTRON_NO is null AND OTHER_REGION is null and TEST_RESULT = 1 AND LAB_ID not in(JLL	998,999), then EXON	I_NO must not be					
	INTRON_NO	number (2,0)	Required: false					
13	Number of intron containing variant. In the case the variant spans more than one intron, t intron affected by the variant	hen this field should	indicate the first					
	Allowable Values							
	1 to 99 Range							
	Error Description							
lf								
lf	DETECTION METHOD = 10 AND CENTER NO in (11.16) then INTRON NO must be null							
lf	DETECTION METHOD = 10 AND CENTER NO = 11 then INTRON NO must be null							
lf N	EXON_NO is null AND OTHER_REGION is null and TEST_RESULT = 1 AND LAB_ID not in(99 JLL	8,999), then INTRON	I_NO must not be					
	OTHER REGION	number (1 0)	Required false					
14	Region, other than coding exon and intron, containing the variant.							
	Allowable Values							
	2 SUIR							
	3 Other							
	Error Description							
lf	If TEST_RESULT is not 1 or 9, then OTHER_REGION must be null							
lf	DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then OTHER_REGION must be null							
lf	DETECTION_METHOD = 10 AND CENTER_NO = 11 then must OTHER_REGION be null							
lf N	EXON_NO is null AND INTRON_NO is null and TEST_RESULT = 1 AND LAB_ID not in(998,99 JLL	99), then OTHER_REG	GION must not be					

NUC_POS_START

string (20)

Required: false

For exonic mutations, the numeric value of the nucleotide first affected by the mutation relative to the reference sequence. For intronic mutations, the numeric value of the first affected nucleotide relative to the exon number; a (-) indicates the number of nucleotides upstream (5) of the exon, and a (+) indicates the number of nucleotides downstream (3) from the exon. The 5 UTR and 3 UTR are considered to be part of the first andILast Exon respectively but have special notation here. Positions in the 5-UTR have (-) sign. Positions in the 3-UTR have (*) sign.

Error Description

If TEST_RESULT is not 1 or 9, then NUC_POS_START must be null

If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUC_POS_START must be null

If TEST_RESULT=1 and LAB_ID not in(998,999), then NUC_POS_START must not be null

If NUC_POS_START is not null AND if OTHER_REGION = 1, then NUC_POS_START must begin with (-)

If NUC_POS_START is not null AND TYPE_VARIANT = 3 (substitution) AND EXON_NO is not null AND OTHER_REGION is null, then N UC_POS_START must not begin with (+, -)

If NUC_POS_START is not null AND If TYPE_VARIANT = 3 (substitution) AND INTRON_NO is not null AND OTHER_REGION is null the n NUC_POS_START must begin with in (+, -)

If NUC_POS_START is not null AND If TYPE_VARIANT = 3 (substitution) AND INTRON_NO is not null AND OTHER_REGION is null the n NUC_POS_START must begin with in (+, -)

If NUC_POS_START is not null AND if TYPE_VARIANT = 3, then NUC_POS_START = NUC_POS_END

If NUC_POS_START is not null AND if TYPE_VARIANT not in (1,3,9), then NUC_POS_START must not equal NUC_POS_END

NUC_POS_END

Required: false

string (20)

string (1)

For exonic mutations, the numeric value of the nucleotide last affected by the mutation relative to the reference sequence. For intronic mutations, the numeric value of the first affected nucleotide relative to the exon number; a (-) indicates the number of nucleotides upstream (5) of the exon, and a (+) indicates the number of nucleotides downstream (3) from the exon. The 5 UTR and 3 UTR are considered to be part of the first andLast Exon respectively but have special notation here. Positions in the 5-UTR have (-) sign. Positions in the 3-UTR have (*) sign

Error Description

If TEST_RESULT is not 1 or 9, then NUC_POS_END must be null

If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUC_POS_END must be null

If TEST_RESULT=1 and LAB_ID not in(998,999), then NUC_POS_END must not be null

If NUC_POS_END is not null AND if OTHER_REGION = 1, then must begin with (-)

If NUC_POS_END is not null AND If TYPE_VARIANT = 3 (substitution) AND EXON_NO is not null AND OTHER_REGION is null, then N UC_POS_END must not begin with (+, -)

If NUC_POS_END is not null AND If TYPE_VARIANT = 3 (substitution) AND INTRON_NO is not null AND OTHER_REGION is null then NUC_POS_END must begin with in (+, -)

If NUC_POS_END is not null AND if TYPE_VARIANT = 3, then NUC_POS_START = NUC_POS_END

If NUC_POS_END is not null and TYPE_VARIANT not in (1,3,9), then NUC_POS_START must not equal NUC_POS_END

NUCLEOTIDE_FROM

17

Expected nucleotide with respect to the reference sequence.



Required: false

T Thymidine Error Description If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_FROM must be null 18 NUCLEOTIDE_TO 18 Observed (mutated) nucleotide. A Adenine C Cytosine C Cytosine G Guanine T Thymidine Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ string (50) Required: false 19 DELETE_SEQ string (50) Required: false If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null 19 DELETE_SEQ string (50) Required: false If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
Error Description If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_FROM must be null 18 NUCLEOTIDE_TO 0bserved (mutated) nucleotide. Allowable Values A Adenine C C Cytosine G Guanine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null 19 DELETE_SEQ Error Description 19 DELETE_SEQ Error Description 19 DELETE_SEQ If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null 19 DELETE_SEQ If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null
Error Description If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_FROM must be null 18 NUCLEOTIDE_TO 18 Observed (mutated) nucleotide. Allowable Values A Adenine C Cytosine G Guanine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ String (50) Required: false Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_FROM must be null 18 NUCLEOTIDE_TO 18 Observed (mutated) nucleotide. Allowable Values A Adenine C Cytosine G Guanine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ String (50) Required: false 19 DELETE_SEQ If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
18 NUCLEOTIDE_TO string (1) Required: false 0bserved (mutated) nucleotide. Allowable Values A Adenine C C Cytosine G Guanine T Thymidine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null string (50) Required: false 19 DELETE_SEQ string (50) Required: false If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null 19 DELETE_SEQ string (50) Required: false If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
18 Observed (mutated) nucleotide. Allowable Values A Adenine C Cytosine G Guanine T Thymidine T Thymidine If DELETE_SEQ String (50) Required: false String (50) Peletion nucleotide sequence Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null String (50) 19 DELETE_SEQ string (50) 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DELETE_SEQ string (50) Deletion nucleotide sequence
Allowable Values A Adenine C Cytosine G Guanine T Thymidine Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
A Adenine C Cytosine G Guanine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DELETE_SEQ Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DELETE_SEQ Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DELETE_SEQ Deletion nucleotide sequence
C Cytosine G Guanine T Thymidine If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ string (50) Required: false Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
G Guanine T Thymidine Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ String (50) Required: false 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
T Thymidine Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
Error Description If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If TEST_RESULT is not 1 or 9, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ string (50) Required: false Deletion nucleotide sequence If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null 19 DELETE_SEQ 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
19 DELETE_SEQ string (50) Required: false 19 Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null Interest provide the provideth
DELETE_SEQ string (50) Required: false Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
Deletion nucleotide sequence Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
Error Description If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If TEST_RESULT is not 1, then NUCLEOTIDE_TO must be null If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then NUCLEOTIDE_TO must be null
If TEST_RESULT=1 and TYPE_VARIANT in (1,7), then NUCLEOTIDE_TO must not be null
20 string (50) Required:false
Error Description
If TEST_RESULT is not 1, then INSERT_SEQ must be null
If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then INSERT_SEQ must be null
If TEST_RESULT=1 and TYPE_VARIANT in (2,4,7), then INSERT_SEQ must not be null
TYPE VARIANT number (1.0) Dequired false
21 Type of genetic change
Allowable Values
Allowable Values 1 Deletion
Allowable Values 1 Deletion 2 Insertion

4	Duplication
5	Translocation
6	Inversion
7	Complex Rearrangement
9	Uncertain

	Error Description						
	If TEST_RESULT is not 1 or 9, then TYPE_VARIANT must be null						
	If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then TYPE_VARIANT must be null						
	If TEST_RESULT is 1 and LAB_ID is not 998 or 999, then TYPE_VARIANT must not be null						
	If VARIANT_NAME_RESULT is null, AND LAB_ID is not 998 or 999 then TYPE VARIANT must not be null						
CODON number (4,0) Required: false							
Numeric location of first mutated amino acid of reference sequence. A numeric value of 1 to the total number of codons on the reference sequence							
Allowable Values 1 to 9999 Range							
		1 to 9999 Range					
		1 to 9999 Range Error Description					
	If TEST_RESULT is not 1, then CODON	1 to 9999 Range Error Description must be null					
	If TEST_RESULT is not 1, then CODON If DETECTION_METHOD = 10 AND CEN	1 to 9999 Range Error Description must be null TER_NO in (11,16) then CODON must	be null				
	If TEST_RESULT is not 1, then CODON If DETECTION_METHOD = 10 AND CEN If TEST_RESULT=1 and EXON_NO=null	1 to 9999 Range Error Description must be null ITER_NO in (11,16) then CODON must I, then CODON must be null	be null				

23 **AA_FROM**

22

string (1) Required: false

Wild type amino acid value of first affected amino acid on reference sequence

Allowable Values				
Α	Ala			
С	Cys			
D	Asp			
E	Glu			
F	Phe			
G	Gly			
н	His			
I	lle			
к	Lys			
L	Leu			
м	Met			
Ν	Asn			

QGInRArgSSerTThrVValWTrpYTyrXStop	Р	Pro
RArgSSerTThrVValWTrpYTyrXStop	Q	Gln
SerTVVVVYYXStop	R	Arg
TThrVValWTrpYTyrXStop	S	Ser
VValWTrpYTyrXStop	т	Thr
WTrpYTyrXStop	v	Val
Y Tyr X Stop	w	Trp
X Stop	Y	Tyr
	x	Stop

Error Description

If TEST_RESULT is not 1, then AA_FROM must be null

If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then AA_FROM must be null

If TEST_RESULT=1 and EXON_NO=null, then AA_FROM must be null

If TEST_RESULT=1 AND EXON_NO is not null AND TYPE_VARIANT =3 , then AA_FROM must not be null

Required: false

string (1)

24 **AA_TO**

Expressed amino acid value of last affected amino acid on reference sequence

Allowable Values				
Α	Ala			
С	Cys			
D	Asp			
E	Glu			
F	Phe			
G	Gly			
н	His			
I	lle			
к	Lys			
L	Leu			
м	Met			
Ν	Asn			
Р	Pro			
Q	Gln			
R	Arg			
S	Ser			
Т	Thr			
v	Val			
w	Trp			

Y	Tyr
Х	Stop
x	Stop

Error Description

If TEST_RESULT is not 1, then AA_TO must be null

If DETECTION_METHOD = 10 AND CENTER_NO in (11,16) then AA_TO must be null

If TEST_RESULT=1 and CODON=null, then AA_TO must be null

If TEST_RESULT=1 AND EXON_NO is not null AND TYPE_VARIANT =3 , then AA_TO must be not null

CONSEQUENCE 25

number (2,0)

Required: false

Category of mutation- defining outcome of the variant



ZYGOSITY

26 Indicates the heterozygote/homozygote state of a variation, regardless of whether it is a polymorphism, unclassified variant or well established mutation.



	Error Description
If VARIANT_ORIGIN does	not equal 3 (germline), then ZYGOSITY must be null
If DETECTION_METHOD =	10 AND CENTER_NO in (11,16) then ZYGOSITY must be null
If VARIANT_ORIGIN = 3 a	nd TEST_RESULT = 3 (failed), then ZYGOSITY must be null
If VARIANT_ORIGIN = 3 a	nd TEST_RESULT not equals 3, then ZYGOSITY must be not null
If VARIANT_ORIGIN = 3 a	nd TEST_RESULT=9, then ZYGOSITY must be 9

1	SEVERITY				number (1.0)	Required false			
27	Type of mutation					Required.tuise			
All	Allowable Values								
1	Deleterious								
2	Polymorphism, as	Polymorphism, assumed neutral							
3	Unclassified variar	Unclassified variant							
4	Unclassified variant, possibly pathogenic. DNA missense alteration with corresponding loss of expression by tumor IHC; alternatively, missense alteration that cosegregates with the disease in family.								
			-	men Decenintien					
		If TEST PESULT is not 1 or 9, the							
		If DETECTION METHOD $= 10 \text{ AND}$		EVERT I Must be full	ITV must be pull				
		If VAPIANT, OPIGIN in $(1, 2)$ AND		SET PESHIT = 1 then SEVEPITY n					
28 INSIGHT_SEVERITY number (1,0) F						Required: false			
20	This will be the 5-point severity code which InSiGHT use.								
		A	Allo	owable Values					
		5	5	Pathogenic					
	4 Likely Pathogenic								
		3	3	Unclassified					
		2	2	Likely Neutral					
		1	1	Neutral					
	9 Unknown								
	Error Description								
If TEST_RESULT is not 1 or 9, then INSIGHT_SEVERITY must be null									
	 I								
If TEST_RESULT=1 and SEVERITY=null, INSIGHT_SEVERITY then must be null									
	If G	ENE in (6,7), then INSIGHT_SEVER	RIT	Y must be null					
20	LAB_ID				number (3,0)	Required: true			
29	Lab or individual v	ho performed the testing.							

Allowable Values						
119	Graham Casey					
125	Steve Gallinger					
128	Peter Laird					
129	Joanne Young					
141	Steve Thibodeau					
228	Robert Haile					

			200	Den Bushenen		
			300			
			301	Karen Makar		
			998	Other (specify, free text)		
			999	Unknown		
20	LAB_OTHER_	ТЕХТ			string (50)	Required: false
30	Name of other/private lab					
	DATE_TEST				string (8)	Required: false
31	Date on which	test was performed. Format: Y	YYYMM	DD		
Date Value Check The date must follow to the following format: Format YYYYMMDD. Must consist of valid date. Components of date should be right justified and zero filled. MM = 01 - 12, 88, 99 DD = 01 - 31, 88, 99 YYYY = Minimum year - system date year, 8888, 9999 Use 88, 8888 for not currently known, in progress to obtain information. Use 99, 9999 for not known. If century is known, but year is unknown then give an estimate of year or code YYYY = 9999. If MM = 99 then DD must = 99. If century is known, but year is unknown then give an estimate of year or code YYYY = 9999. If YYYY = 9999 then MM and DD must = 99. The following special parameters are used: 2002 Minimum year						

Module: molecular

Module Contents

braf_kras 1. CENTER NO (*PK) 2.SOMATIC CID (*PK) 3. PERSON ID 4.TUMOR NO 5. POLYP NO 6.<u>GENE</u> 7. VARIANT ORIGIN 8. SOURCE NUC ACID 9. NUC ACID CID 10.<u>DETECTION METHOD</u> 11.<u>GENOMIC REGION TESTED</u> 12.<u>TEST_RESULT</u> 13. VARIANT NAME RESULT 14.<u>EXON NO</u> 15.<u>NUC POS START</u> 16.<u>NUC POS END</u> 17. NUCLEOTIDE FROM 18. NUCLEOTIDE TO 19. NUCLEOTIDE TO 19. NUCLEOTIDE TO 20.<u>CODON</u> 21.AA FROM 22.<u>AA TO</u> 23.CONSEQUENCE 24.LAB ID 25. LAB OTHER TEXT 26. DATE TEST

1	CENTER_NO (*PK) number(2,0) Require Center identification number. *CENTER_NO & SOMATIC_CID are the primary key for the table. Require									
1										
Allowable Values										
	11 Sinai Health Systems (formerly Cancer Care Ontario)									
	12 Cedars-Sinai & Cleveland Clinic (formerly USC Consortium)									
	13 University of Melbourne									
	14 University of Hawaii Cancer Center									
		15	Mayo Clinic							
		16	Fred Hutch, Se	eattle						
17 UCSF: University of California at San Franscisco (formerly CPIC, originally Northern California (NCC						NCCC))				
2	SOMATIC_CID (*PK) String (40) Require						Required: TRUE			
	Centre identifier for the test. *CENTER_NO & SOMATIC_CID are the primary key for the table.									
PERSON_ID string (12) Require						Required: TRUE				
	Number t	Number that uniquely identifies an individual.								
TUMOR_NO number (2,0) Required							Required: TRUE			
4	Number la	abelli	ng the tumor of	the i	ndividual. Numbers are not necessarily sequential.					
Allowable Values										
				-9	NA/Out of scope: Tissue is not cancer or contiguous add	enoma.				
				-15	Information Unknown					
			L							

РО	DLYP_NO						number (2,0)	Required: TRUE			
Sec	Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date.										
	Allo	owable Values									
	-9 NA/Out of scope: Tissue is not a polyp.										
	-1	Polyp has MSI/IHC re	esult but cent	ter is c	urrently unabl	e to locate poly	p pathology information	on			
GE	ENE						number (1,0)	Required: TRUE			
Ge	ene on which tes	ting has been performe	ed.								
				Allov	vable Values						
				6	BRAF						
				7	KRAS						
	ARIANT_ORIGI	N					number (1,0)	Required: TRUE			
De	escribes if the va	riant was tested as an	acquired or a	a germ	line variant.						
				Allowal	ole Values						
				1 Pre	sumed somati	с					
				2 Kno	own somatic						
			[:	3 Ger	mline						
				Error	Description						
		If GENE in (6,7) AND	SOURCE_NU	C_ACII	D (5,6,7,8), th	en VARIANT_O	RIGIN must equal 1				
SO	OURCE_NUC_A	CID					number (1,0)	Required: TRUE			
Spe	ecifies source of	tissue used for testing	I								
			Allowable Va	alues							
			4 normal f	resh fr	ozen tissue						
			5 other no	n-tum	our tissue						
			6 polyp								
			7 tumour	(paraff	in embedded t	umour- PET)					
			8 tumor- f	resh fr	ozen						
			9 unknowr	n/priva	te lab						
		·									
9 NU	JC_ACID_CID						string (15)	Required: TRUE			
Ide	entifier used inte	ernally by centers for a	single extrac	tion o	n which the te	st was carried o	put				
				Allov	vable Values						

			-9 unknown		
			Error Description		
	If	SOURCE_I	NUC_ACID=9, then NUC_ACID_CID must be	-9	
	DETECTION_METHOD			number (2,0)	Required: TRUE
10	Somatic DNA testing method.] [
	(Mass Spec = Sequenom MassARRA	Y system,	MALDI-TOF)		
		Allo	wable Values		
		1	DNA Sequencing		
		18	Fluorescent ASP (BRAF)		
		19	HRM with sequencing (KRAS)		
		40	SYBR real time PCR assay		
		99	Unknown/private genetic test results		
	GENOMIC_REGION_TESTED			string (30)	Required: TRUE
11	Indicates what part(s) of the gene of	r specific	mutation or other genomic region(s) was TE	STED	
		Allewahl			
		Allowabl			
			BRAF C. 17991>A p. Valouogiu		
			KPAS codon 61		
	TEST_RESULT			number (1,0)	Required: TRUE
12	Result of the test carried out and the	is is at the	e test level. This is NOT at person level.		
			Allowable Values		
			1 Change detected		
			2 No change detected		
			3 Test failed		
			9 Equivocal		
12	VARIANT_NAME_RESULT			string (30)	Required: FALSE
	Proper variant name using Human G	Genomic V	ariant Society (HGVS) nomenclature.		
			Allowable Values		
			BRAF c.1799T>A p.V600E		
			KRAS c.34G>A		
			KRAS c.34G>C		
			KRAS c.34G>T		
			KRAS c.35G>A		

		KRAS c.35G>C	
		KRAS c.35G>T	
		KRAS c.37G>A	
		KRAS c.37G>C	
		KRAS c.37G>T	
		KRAS c.38G>A	
		KRAS c.38G>T	
		Frror Description	
		If TEST_RESULT does not equal 1, then VARIANT_NAME_RESULT must be null	
		If TEST_RESULT=1, then VARIANT_NAME_RESULT must not be null	
	EXON_NO	number (2,0)	Required: false
14	Number of exon In the case the v	containing variant. ariant spans more than one exon, then this field should indicate the first exon affected by t	he variant.
		Allowable Values	
		2 KRAS	
		15 BRAF	
		Error Description	
	IF TEST PE		
		NO is null AND OTHER REGION is null AND LAB ID not in(998,999), then EXON NO must	not be NUU I
15	NUC_POS_STAI	string (20)	Required: false
	For exonic mutat	ions, the numeric value of the nucleotide first affected by the mutation relative to the reference	ence sequence.
		Error Description	7
		If TEST_RESULT does not equal 1, then NUC_POS_START must be null	
		If TEST_RESULT=1 and LAB_ID not in(998,999), then NUC_POS_START must not be null.	
		If TYPE_VARIANT =3, then NUC_POS_START must equal NUC_POS_END	
		If TYPE_VARIANT does not equal 3, then NUC_POS_START must not equal NUC_POS_END	
	NUC_POS_END	string (20)	Required: false
16	For exonic mutat	ions, the numeric value of the nucleotide last affected by the mutation relative to the refere	ence sequence.
		Error Description	
		If TEST_RESULT does not equal 1, then NUC_POS_END must be null	
		If TEST_RESULT=1 and LAB_ID not in(998,999), then NUC_POS_END must not be null	
1		IF TYPE VARIANT -3 then NUC POS END must equal NUC POS START	

47	NUCLEOTIDE_FROM					string (1)	Required: false	
17	Expected nucleotide with	respect to the reference seq	uen	ce.				
			Alle	owable Values				
			Α	Adenine]			
			С	Cytosine]			
			G	Guanine]			
			Т	Thymidine				
		r			1			
			Erro	or Description	1			
		If TEST_RESULT does not e	equa	I 1, then NUCL	EOTIDE_FROM mu	ist be null		
10	NUCLEOTIDE_TO					string (1)	Required: FALSE	
18	Observed (mutated) nucl	eotide.						
			Alle	owable Values				
			A	Adenine				
			С	Cytosine				
			G	Guanine				
			Т	Thymidine				
								
			Erro	or Description	ו 			
		If TEST_RESULT does not	equ	al 1, then NUC	CLEOTIDE_TO mus	t be null		
10	TYPE_VARIANT					number (1,0)	Required: false	
17	Type of genetic change							
			Alle	owable Values				
			3	Substitution				
			9	Uncertain				
		[1			
			Erro	or Description	ו 			
		If TEST_RESULT does no	t eq	ual 1, then TYI	PE_VARIANT must	be null		
		If TEST_RESULT=1, ther	n TYF	PE_VARIANT m	nust be in (3,9)			
	CODON					number (4,0)	Required: false	
20	Numeric location of first r A numeric value of 1 to the	mutated amino acid of refere he total number of codons or	nce 1 the	sequence. e reference sec	Juence			
			Erro	or Description	1			
	If TEST_RESULT of	does not equal 1, then CODC	N m	lust be null				
	If TEST_RESULT=1 AND EXON_NO does not equal null AND TYPE_VARIANT =3 , then CODON must be not null							

	Wild type amino	acid	value o	of first	affected	amino	acid	on	reference	sec	luence
--	-----------------	------	---------	----------	----------	-------	------	----	-----------	-----	--------

Allowable Values						
A	Ala					
С	Cys					
D	Asp					
E	Glu					
F	Phe					
G	Gly					
н	His					
I	lle					
к	Lys					
L	Leu					
м	Met					
N	Asn					
Р	Pro					
Q	Gln					
R	Arg					
S	Ser					
т	Thr					
v	Val					
w	Trp					
Y	Tyr					
X	Stop					

Error Description

If TEST_RESULT does not equal 1, then AA_FROM must be null

If TEST_RESULT=1 AND CODON is not null AND TYPE_VARIANT =3 , then AA_FROM must not be null

string (1)

Required: FALSE

	AA_TO
22	
~~	

Expressed amino acid value of last affected amino acid on reference sequence

Allowable Values						
A Ala						
С	Cys					
D	Asp					
E	Glu					
F	Phe					
G	Gly					

					н	His			
					<u> </u>	lle			
					К	Lys			
					L	Leu			
					м	Met			
					N	Asn			
					Р	Pro			
					٩	Gln			
					R	Arg			
					S	Ser			
					Т	Thr			
					v	Val			
					w	Trp			
					Y	Tyr			
					X	Stop			
					F F				
				theore					
		IF TEST DESU		, then			2 thon A	A TO must not be	
		II TEST_RESU	LT=T AND CODON T	IS HOL		DITPE_VARIANT	= s , then A	A_TO must not be t	
	CONSEQUE	NCE						number (1,0)	Required: FALSE
23	Category of r	mutation- defini	ng outcome of the v	varian	it				
				Allo	wable V	'alues			
				1	Missens	se			
				2	Premat	ure termination			
				9	Uncerta	iin			
					Error D	escription			
			If TEST_RESULT do	oes no	ot equal	1, then CONSEQ	UENCE must	be null	
			If TEST_RESULT=1	l, ther	ר CONSE	QUNCE must be	not null		
24	LAB_ID							number (3,0)	Required: TRUE
24	Lab or individ	dual who perfor	med the testing.						
				Allowa	able Valu	les			
			-	119	Grahan	n Casey			
			 [·	125	Steve 0	Gallinger			
				128	Peter L	aird			

129 Joanne Young

			141	Steve Thibodeau		
			228	Robert Haile		
			300	Dan Buchanan		
			301	Karen Makar		
			998	Other (specify, free text)		
			999	Unknown		
25	LAB_OTHER_	TEXT			string (30)	Required: false
	Name of other	/private lab				
	DATE_TEST				string (8)	Required: TRUE
26	Date on which	test was performed.				
		Date Value Check The date must follow to the format YYYYMMDD. Must con Components of date should by MM = 01 - 12, 88, 99 DD = 01 - 31, 88, 99 YYYY = Minimum year - syst Use 88, 8888 for not currently Use 99, 9999 for not known. If century is known, but year If MM = 99 then DD must = 9 If century is known, but year If YYYY = 9999 then MM and The following special paramet YYYYMMDD : MM	billowing sist of e right tem da y know is unkr 29. is unkr DD mu ters are 01 - 1	g format: valid date. justified and zero filled. te year, 8888, 9999 n, in progress to obtain inform nown then give an estimate of nown then give an estimate of st = 99. e used: 12, 88, 99 31, 88, 99	nation. ⁷ year or code YYYY = 9999. ⁷ year or code YYYY = 9999.	
		ΥΥΥΥ	2002	- system year, 8888, 9999		
		DD	01 - 3 2002	31, 88, 99 - system year, 8888, 9999		

Module Contents

d_MSJ_IHC_PK_MSI 1.CENTER_NO 2.PERSON_ID 3.COLLECTION_CID 4.TEST_TISSUE 5.TUMOR_NO 6.POLYP_NO 7.BLOCK_SPEC_CID 8.NUC_ACID_CID_TUMOR 9.NUC_ACID_CID_NORMAL 10.MSI_STATUS_DNA

1	CENTER_NO number (2,0)													
	Center Identification Number.													
	Allowable Values													
		11	Sinai Health Systems (formerly Cancer Care Ontario)											
		12	Cedars-Sinai & Cleveland Clinic (formerly USC Consortium)											
		13	University of Melbourne											
		14	University of Hawaii Cancer Center											
	15 Mayo Clinic													
	16 Fred Hutch, Seattle													
		17 UCSF: University of California at San Franscisco (formerly CPIC, originally Northern California (NCCC))												

2	PERSON_ID			string (12)	Required: true				
Number that uniquely identifies an individual.									
	COLLECTION_CID			string (30)	Required: true				
3	Identifier used internally by centers to denote	aι	nique surgical event.						
	This data element is typically used to allow ag	gre	gation of all materials, such as block	s, from a single surgery.					
	TEST_TISSUE			number (1,0)	Required: true				
4	Only values of 1,2,3 link to pathology informat Although values 2 and 3 may represent the sa Adjacent adenomas are recorded on the Color Polyps table. As a result, the pathology inform There may be cases where TEST_TISSUE varia adjacent adenoma present. In these cases, the have been overlooked.	tior ame ect <u>-</u> natio able e Tl	tissue type, their pathology informa _Malig table and so are associated wi on for each will differ somewhat. has level 2 but the ADJ_ADEN varia EST_TISSUE value takes precedence	tion is stored in different th a cancer; Colon polyps ble of Colorect_Malig tabl and the adjacent adenon	database tables. are recorded on the e indicates no a is considered to				
		Allo	owable Values						
	[1	Cancer						
	[2	Contiguous (Adjacent) adenoma	a					
3 Colon polyp									
	[4	Gastric polyp						
	5 Sebaceous adenoma								

Numbers labeling tumors of the Individual. Numbers are not necessarily sequential. No adjustments made when a tumor is detected from the system. Tumor numbers are never reused. PERSON, NO plus TUMOR, NO uniquely identifies each tumor in the contral database. The same tumor number from different tables (including tables outside of the biospecimen module) must always fortor to be same physical tumor. POLYP_NO number (2.0) Required: true Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular dato. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO inquery identify a physical polyp globally within the CFR database. Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular dato. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO inquery identify a physical polyp globally within the CFR database. Corresponding data elements, to PDVP_STER_POLY_PTSEZ, POLY_PTSEZ, POLY_P		TUMOR_NO			number	(2,0)	Required: true	
5 Muchaers are not necessarily sequential. No adjustment 5 made when a tumor is detected from the system. Tumor number from different tables (including tables outside of the biospecimen module) must always refer to the same physical tumor. Image: I		Numbers labeling tumors of	the inc	lividual.				
Allowable Values ••9 NA/Out of scope: Tissue Is not cancer or contiguous adenoma. ••15 Information Unknown POLYP_NO number (2.0) Reguired: true Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CFR database. • The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes compositing data deemonts, i.e. POLYP_STES, POLYP_OTES, LEX, POLYP_CRADEX, where x is an integer between 1 and a escenteed according to the following rules: 1. The largest polyp is reported first, followed by next largest	5	Numbers are not necessarily No adjustment is made when uniquely identifies each tum biospecimen module) must a	PERSON_NO (including t	D plus TUMOR_NO ables outside of the				
-9 NA/Out of scope: Tissue is not cancer or contiguous adenoma. -15 Information Unknown POLYP_NO number (2,0) Required:true Sequential number over range of 1 to 90 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CFR database. 6 The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes corresponding data elements. i.e. POLYP_SITEX, POLYP_SIZEX, POLYP_SIZEX, POLYP_CRADEx, where x is an integer between 1 and 3, inclusive. The physical polyp referenced by x and this data element must be identical. Polyps are described according to the following vulse. 1. The largest poly is reported thirst, Ioliowed by next largest 2. If size is not indicated, adenomas are reported before hyperplastic polyps • 1 bize is not indicated, adenomas are reported before hyperplastic polyps • • 1 bize is not indicated, adenomas are reported before hyperplastic polyps • • 1 bize is not indicated, adenomas are reported before hyperplastic polyps • • 1 bize is not indicated, adenomas are reported before hyperplastic polyps • • 1 bize is not an oligo. • • 1 bize is not an oligo. • • 1 bize is not an oligo. • • 1 bize is not a polyp. • • 1 bixe is not a polyp.			Allov	vable Values				
-15 Information Unknown POLYP_NO number (2.0) Required:true Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLEGTED, and POLYP_NO uniquely identify a physical polyp globally within the CR database. a The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes corresponding data elements. Le. POLYP_SITEX, POLYP_SIZEX, POLYP_SIZEX, POLYP_SRADEX, where x is an integer between 1 and 3, inclusive. The physical polyp referenced by x and this data element must be identical. Polyps are described according to the following rules: 1. The largest polyp is reported off this. (filowode by next largest) 1. The largest polyp is reported off this. (filowode by and this data element must be identical. Polyps are described first. (filowode by and this data element must be identical. 9 NA/Out of scope. Tissue is not a polyp. 1 1 Polyps hit/MS1 result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. 1 BLOCK_SPEC_CID string (40) Required: true 2 Information UNKnown Information UNKnown Information UNKnown Information UNKnown 1 BLOCK_SPEC_CID_TUMOR string (40) Required: true Information UNKnown 2 Ecnter's unique identifier for the tumor DNA used in this MSI t			-9	NA/Out of scope: Tissue is not cancer or contiguous add	enoma.			
POLYP_NO number (2.0) Required true Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CR database. 6 The colon pathology form has three sets of identical fields for recording information en polyps. The POLYPS table includes corresponding data learnents, i.e. POLYP_SITEx, POLYP_SIZEx, POLYP_CRADEX, where x is an integer between 1 and polyps are described according to the following rules: 1. The largest polyp is reported first, followed by nox til argest 2. Horize stopic is reported first, followed by nox til argest 3. Horize throws are reported before hyperplastic polyps 3. Polyps with most severe grade are reported before hyperplastic polyps 3. Polyps with most severe grade are reported Motall polyps present in available tissue have an assigned value, as some are not described. Allowable Values 9 NAVOut of scope. Tissue is not a polyp. 1 Polyp has HE//MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. Information Unknown 8 NUC_ACID_CID_TUMOR Center's unique identifier of the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Indivable Values 15 Information Unknown 16 17 DNA identifying information l			-15	Information Unknown				
POLVP_NO number (2.0) Required:true Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLVP_NO uniquely identify a physical polyp globally within the CR database. 6 The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes corresponding data learners, i.e. POLYP_SITEx, POLYP_SITEx, POLYP_CRADEX, where x is an integer between 1 and 3. Inclusive. The physical polyp preferenced by x and this data element must be identical. 9. Inclusives topins reported first, followed by next largest. 1. The largest polyp is reported first, followed by parts largest. 9. If size is not indicated, adenomas are reported before hyperplastic polyps 3. Inclusive mode according to the following rules: 9. NAVOut of scope. Tissue is not a polyp. 1 Polyp has HE/CNSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. 7 BLOCK_SPEC_CID string (40) Required: true 8 Output first field in not negative, there will be a matching record on the NUC_ACID_CID_TUMOR string (40) Required: true 8 Output field in not negative, there will be a matching record on the NUC_ACID beth at indicates whether remaining function lost/unknown. 14 Center's unique identifier. 1 Indiverse are provided for the same block and tumor then separate identifiers								
Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID. DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CFR database. 6 The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes corresponding data elements. i.e. POLYP_SITEX, POLYP_FIXEX, POLYP_SIZEX, POLYP_GRADEX, where x is an integer between 1 and 3, inclusive. The physical polyp referenced by x and this data element must be identical. Polyps are described according to the following rules: 1. The largest polyp is reported first. followed by next largest 2. If size is not indicated, ademonas are reported before hyperplatic polyps 3. Polyps with most severe grade are reported before hyperplatic polyps 9. NA/Out of scope. Tissue is not a polyp. 1 Polyp has IHC/MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. 7 BLOCK_SPEC_CID string (40) Required: true 1 Information Unknown Allowable Values -15 Information Unknown 8 NUC_ACID_CID_TUMOR string (40) Required: true Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. 8 Unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier.		POLYP_NO			number	(2,0)	Required: true	
Allowable Values -9 NA/Out of scope. Tissue is not a polyp. -1 Polyp has IHC/MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. 7 BLOCK_SPEC_CID string (40) Required: true 7 Unique local identifier used at a center to uniquely identify a block tissue specimen. Allowable Values -15 Information Unknown 8 Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values -15 -15 DNA identifying information lost/unknown. -14 Center does not keep normal tissue DNA identifying information. -13 Quantity of DNA or tissue not sufficient.	6	Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CFR database. The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes corresponding data elements, i.e. POLYP_SITEx, POLYP_TYPEx, POLYP_SIZEx, POLYP_GRADEx, where x is an integer between 1 and 3, inclusive. The physical polyp referenced by x and this data element must be identical. Polyps are described according to the following rules: 1. The largest polyp is reported first, followed by next largest 2. If size is not indicated, adenomas are reported before hyperplastic polyps 3. Polyps with most severe grade are reported Not all polyps present in available tissue have an assigned value, as some are not described						
-9 NA/Out of scope. Tissue is not a polyp. -1 Polyp has IHC/MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. 7 BLOCK_SPEC_CID string (40) Required: true 7 Unique local identifier used at a center to uniquely identify a block tissue specimen. Allowable Values -15 Information Unknown 8 NUC_ACID_CID_TUMOR string (40) Required: true 8 Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, if this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values -15 DNA identifying information lost/unknown. -14 Center does not keep normal tissue DNA identifying information. -13 Quantity of DNA or tissue not sufficient.	4	Allowable Values						
Image: Specific Content is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway. Image: Specific Content is unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Image: Specific Content is unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Image: Specific Content is specific Content is specific to the specific content is specified. Image: Specific Content is specific Content is specific content is specific content is specific. Image: Specific Content is specific content is specific content is specific content is specific. Image: Specific Content is content is specific content is specific content is specific content is specific. Image: Specific Content is content is the point is is content is specific content	[-	-9 NA/Out of scope. Tissue is	s not a	polyp.				
7 BLOCK_SPEC_CID string (40) Required: true 7 Unique local identifier used at a center to uniquely identify a block tissue specimen. Allowable Values -15 Information Unknown 8 NUC_ACID_CID_TUMOR string (40) Required: true Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values • 15 DNA identifying information lost/unknown. • 14 Center does not keep normal tissue DNA identifying information. • 13 Quantity of DNA or tissue not sufficient.		-1 Polyp has IHC/MSI result information is currently u	but ce nderwa	nter is currently unable to locate polyp pathology informa ly.	tion. Cen	ter review to	o obtain this	
7 Unique local identifier used at a center to uniquely identify a block tissue specimen. Allowable Values -15 Information Unknown String (40) Required: true Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values -15 DNA identifying information lost/unknown. -14 Center does not keep normal tissue DNA identifying information. -13 Quantity of DNA or tissue not sufficient. string (40)		BLOCK_SPEC_CID			string (4	40)	Required: true	
Allowable Values -15 Information Unknown 8 String (40) Required: true Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values - 15 DNA identifying information lost/unknown. - 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient.	7	Unique local identifier used a	at a cei	nter to uniquely identify a block tissue specimen.				
NUC_ACID_CID_TUMOR string (40) Required: true 8 Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. 9 Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values - 15 DNA identifying information lost/unknown. - 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient. NUC_ACID_CID_NORMAL string (40)				Allowable Values -15 Information Unknown				
8 Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. 9 Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified. Allowable Values - 15 DNA identifying information lost/unknown. - 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient. string (40)		NUC_ACID_CID_TUMOR			string (4	40)	Required: true	
Allowable Values - 15 DNA identifying information lost/unknown. - 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient.	8	Center's unique identifier for the tumor DNA used in this MSI test. Each DNA extraction has a unique identifier. Unique Identifier, If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining tumor DNA from the extraction used for this test is available for distribution to requestors. If multiple MSI results are provided for the same block and tumor then separate identifiers must be specified.						
- 15 DNA identifying information lost/unknown. - 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient. NUC_ACID_CID_NORMAL string (40)			Allowa	ble Values				
- 14 Center does not keep normal tissue DNA identifying information. - 13 Quantity of DNA or tissue not sufficient. NUC_ACID_CID_NORMAL string (40)			- 15	DNA identifying information lost/unknown.				
- 13 Quantity of DNA or tissue not sufficient. NUC_ACID_CID_NORMAL String (40) Required: true		[- 14	Center does not keep normal tissue DNA identifying info	rmation.			
NUC_ACID_CID_NORMAL string (40) Required: true		[- 13	Quantity of DNA or tissue not sufficient.				
		NUC ACID CID NORMAL			strina (4	40)	Required: true	

Center's unique identifier for the normal DNA used in this MSI test. Each DNA extraction has a unique identifier.

Normal DNA may have been extracted from paraffin embedded normal tissue, blood, mouthwash/buccal cells or fresh frozen normal tissue. Paraffin embedded normal tissue is not necessarily associated with the paraffin block from which the tumor DNA is taken. If this field in not negative, there will be a matching record on the NUC_ACID table that indicates whether remaining normal DNA from the extraction used for this test is available for distribution to requestors.

Allow	able Values
- 15	DNA identifying information lost/unknown.
- 14	Center does not keep normal tissue DNA identifying information.
- 13	Quantity of DNA or tissue not sufficient.

MSI_STATUS_DNA	number (1,0)	Required: true

MSI status of DNA extraction.

9

¹⁰ For each set of scores with a minimum of 4 of 10 markers (excluding D2S123) that are either stable or unstable (Options 1 and 2 only NOT 7 or 8), the percent unstable among stable and unstable markers is calculated and compared to cutpoints to determine if the results are stable (0% unstable), low (1-29% unstable) or high (30-100% unstable). If only 3 markers are scored but they are all unstable then the status is scored as high.

Allowable Values				
-7	NA/Out of scope - Less than 4 marker scores stable or unstable			
1	Stable - 0% unstable			
2	Low - More than 0% but less than 30% unstable			
 2 Low - More than 0% but less than 30% unstable 3 High - At least 30% unstable 				

Module: derived

Module Contents

d_MSI_IHC_PK_IHC 1.PERSON_ID 2.COLLECTION_CID 3.TEST_TISSUE 4.TUMOR_NO 5.POLYP_NO 6.BLOCK_SPEC_CID 7.IHC_STATUS_TISSUE 8.MSI_STATUS_TISSUE 9.MSIIHC_STATUS_TISSUE

	PERSON_ID		string (12)	Required: true			
1	Number that Uniquely Identifies an Individual.						
	COLLECTION_CID		string (40)	Required: true			
2	The label assigned by the center to all tissue as	ne label assigned by the center to all tissue associated with a single surgical event - resection or biopsy/endoscopy procedure.					
	This data element is typically used to allow aggr	regation of all materials, such as blocks,	from a single surgery.				
	TEST_TISSUE		number (1,0)	Required: true			
3	Only values of 1,2,3 link to pathology information. Although values 2 and 3 may represent the same tissue type, their pathology information is stored in different database tables. Adjacent adenomas are recorded on the Colorect_Malig table and so are associated with a cancer; Colon polyps are recorded on the Polyps table. As a result, the pathology information for each will differ somewhat. There may be cases where TEST_TISSUE variable has level 2 but the ADJ_ADEN variable of Colorect_Malig table indicates no adjacent adenoma present. In these cases, the TEST_TISSUE value takes precedence and the adjacent adenoma is considered to have been overlooked.						
	A	llowable Values					
	1	Cancer					
	2	Contiguous (Adjacent) adenoma					
	3	Colon Polyp					
	4	Gastric polyp					
	5	Sebaceous adenoma					
_	TUMOR_NO		number (2,0)	Required: true			
4	Numbers labeling tumors of the individual. Num No adjustment is made when a tumor is deleted uniquely identifies each tumor in the central dat	bers are not necessarily sequential. From the system. Tumor numbers are n abase. The same tumor number from di	never reused. PERSON	_NO plus TUMOR_NC			

 Allowable Values

 1 to 99 or -9, -15
 Range

 -9
 NA/Out of scope: Tissue does not currently have a TUMOR_NO and is not cancer or contiguous adenoma

 -15
 Information Unknown

biospecimen module) must always refer to the same physical tumor.

POLYP_NO

Sequential number over range of 1 to 99 to distinguish a polyp removed on a particular date. The tuple PERSON_ID, DATE_COLLECTED, and POLYP_NO uniquely identify a physical polyp globally within the CFR database.

The colon pathology form has three sets of identical fields for recording information on polyps. The POLYPS table includes

- 5 corresponding data elements, i.e. POLYP_SITEx, POLYP_TYPEx, POLYP_SIZEx, POLYP_GRADEx, where x is an integer between 1 and 3, inclusive. The physical polyp referenced by x and this data element must be identical.
 - Polyps are described according to the following rules:
 - 1. The largest polyp is reported first, followed by next largest
 - 2. If size is not indicated, adenomas are reported before hyperplastic polyps
 - 3. Polyps with most severe grade are reported
 - Not all polyps present in available tissue have an assigned value, as some are not described.

Allowable Valu	les
1 to 99 or -9, -1	Range
-9	NA/Out of scope. Tissue is not a polyp.
-1	Polyp has IHC/MSI result but center is currently unable to locate polyp pathology information. Center review to obtain this information is currently underway.

4	BLOCK_SP	EC_C		string (40)	Required: true	
0	Unique local					
			Allowable Values -15 Information Unknown			
	IHC_STATU	JS_T	TISSUE	number(1,0)	Required: true	
7	ormal if at least one					
	Allowable Values					
	-8 No data transmitted from centers					
		-7	NA/Out of scope: None of IHC_MLH1, IHC_MSH2, IHC_MSH6, IHC_PMS2	recorded values 0,1,	2,4.	
		1	Normal MMR protein expression			

2 Absence of MMR protein expression

MSI_STATUS_TISSUE	number(1,0)	Required: true
-------------------	-------------	----------------

8 MSI status of tissue from tested block.

The maximum of MSI_STATUS_DNA over all DNA extractions from a given tissue-block combination.

Allo	Allowable Values				
-8 No data transmitted from centers					
-7	NA/Out of scope - Less than 4 marker scores stable or unstable				
1	Stable - 0% unstable				
2	Low - More than 0% but less than 30% unstable				
3	High - At least 30% unstable				

	MSIIHC_STATUS_TISSUE			number(1,0)	Required: true
	MMR status considering all MSI DN	JS_TISSUE number(1,0) Required: true sidering all MSI DNA extractions and IHC results on that tissue/block combination. sidering all MSI DNA extractions and IHC results on that tissue/block combination. S_TISSUE has a value for each tissue that has an MSI and/or IHC result within a given block. sidering all MSI_STATUS_TISSUE is high and IHC_STATUS_TISSUE is absent. S_TISSUE is defective 1 if it does not qualify as defective 2 and MSI_STATUS_TISSUE is high or SSUE is absent. SISSUE is proficient 2 if MSI_STATUS_TISSUE is stable or low and IHC_STATUS_TISSUE is normal. S_TISSUE is proficient 1 if it does not qualify as proficient 2 or as defective, and MSI_STATUS_TISSUE is stable or IUS_TISSUE is normal. SISSUE is normal. S_TISSUE otherwise equals -7, corresponding to both MSI_STATUS_TISSUE and IHC_STATUS_TISSUE equaling aling -7 with no record available to merge to on the other table. Allowable Values			
9	MSIIHC_STATUS_TISSUE has a va MSIIHC_STATUS_TISSUE is defect MSIIHC_STATUS_TISSUE is defect IHC_STATUS_TISSUE is absent. MSIIHC_STATUS_TISSUE is profici MSIIHC_STATUS_TISSUE is profici low or IHC_STATUS_TISSUE is nor MSIIHC_STATUS_TISSUE otherwis or one them equaling -7 with no re	ilue f ive 2 ive 1 ient 2 mal. ient 2 mal. ie eq	or each tissue that has an MSI and/or IHC result with if MSI_STATUS_TISSUE is high and IHC_STATUS_TI if it does not qualify as defective 2 and MSI_STATUS 2 if MSI_STATUS_TISSUE is stable or low and IHC_ST 1 if it does not qualify as proficient 2 or as defective, a uals -7, corresponding to both MSI_STATUS_TISSUE I available to merge to on the other table.	in a given block. SSUE is absent. 5_TISSUE is high or ATUS_TISSUE is nor and MSI_STATUS_TI and IHC_STATUS_TI	rmal. SSUE is stable or ISSUE equaling -7
		Allo	wable Values		
		-8	No data transmitted from centers		
		-7	NA/Out of Scope - Both source variables out of scop	e	
		1	Proficient MMR 1		
		2	Proficient MMR 2		
		3	Defective MMR 1		
		4	Defective MMR 2		

Module: derived

Module Contents

d_MSI_IHC

1.<u>PERSON_ID</u> 2.<u>IHC_STATUS_PERSON</u> 3.<u>MSI_STATUS_PERSON</u> 4.<u>MSIIHC_STATUS_PERSON</u>

	PERSON_ID			string (12)	Required: TRUE				
	Number that Uni	Number that Uniquely Identifies an Individual.							
	IHC_STATUS_F	Required: TRUE							
	IHC status of per	IHC status of person tested.							
2	Considers IHC results only for colon cancer tumor types. Excludes all other malignancies, adenomas, metastases and in situ carcinomas. Does not restrict to qualifying tumors. If at least one test is abnormal, then IHC_STATUS_PERSON is abnormal. If there are no abnormal results and at least one normal result, then IHC_STATUS_PERSON is normal. Colon cancer is defined by ICDO3 site codes C180, C181, C182, C183, C184, C185, C186, C187, C188, C189, C199, C209, C218, C260, and a malignant tumor (BEHAV=3). On 1/4/2008 the SITE codes used in deriving this variable were expanded to include C218 and C260.								
		Allo	wable Values						
		-8 No data transmitted from centers							
		-7	NA/Out of scope: Individual has IHC record, but no markers having	values 0,1,2, or 4.					
		1	Normal MMR protein expression						
		2	Absence of MMR protein expression						
	MSI_STATUS_F	PERS	ON	number (1,0)	Required: TRUE				
	MSI status of per	rson 1	ested.						
 Considers MSI results only for colon cancer tumor types. Excludes all other malignancies, adentication carcinomas. Does not restrict to qualifying tumors. If at least one MSI_STATUS_TISSUE result is high, then MSI_STATUS_PERSON is high. If no MSI at least one is low, then MSI_STATUS_PERSON is low. If no MSI_STATUS_TISSUE results are h then MSI_STATUS_PERSON is stable. Colon cancer is defined by ICDO3 site codes C180, C181, C182, C183, C184, C185, C186, C187, C260, and a malignant tumor (BEHAV=3). On 1/4/2008 the SITE codes used in deriving this variable were expanded to include C218 and 				denomas, metastase o MSI_STATUS_TISS re high or low, but a C187, C188, C189, C and C260.	s and in situ GUE result is high bu t least one is stable 199, C209, C218,				

Allo	Allowable Values						
-8	8 No data transmitted from centers						
-7	NA/Out of scope - An MSI record, but no Stable, Low, or High tissue-level result recorded.						
1	Stable - No High or Low tissue-level results, but at least one Stable.						
2	Low - No High tissue-level results, but at least one Low.						
3	High - At least one High tissue-level result						

MSIIHC_STATUS_PERSON

number (1,0) Required: TRUE

MMR status for person tested.

Considers MSI and IHC results only for colon cancer tumor types. Excludes all other malignancies, adenomas, metastases and in situ carcinomas. Does not restrict to qualifying tumors.

MSIIHC_STATUS_PERSON has a value for each each tissue that has an MSI and/or IHC result within a given block.

MSIIHC_STATUS_PERSON is defective 2 if MSI_STATUS_PERSON is High and IHC_STATUS_PERSON is Abnormal. 4 MSIIHC_STATUS_PERSON is defective 1 if it does not qualify as defective 2 and MSI_STATUS_PERSON is High or IHC_STATUS_PERSON is Abnormal. MSIIHC_STATUS_PERSON is proficient 2 if MSI_STATUS_PERSON is Stable or Low and IHC_STATUS_PERSON is Normal. MSIIHC_STATUS_PERSON is proficient 1 if it does not qualify as proficient 2 or as defective, and MSI_STATUS_PERSON is Stable or Low or IHC_STATUS_PERSON is Normal. MSIIHC_STATUS_PERSON otherwise equals -7, corresponding to both MSI_STATUS_PERSON and IHC_STATUS_PERSON equaling -7 or one them equaling -7 with no record available to merge to on the other table. Allowable Values -8 No data transmitted from centers -7 NA/Out of Scope - Both source variables out of scope 1 Proficient MMR 1 2 Proficient MMR 2 Defective MMR 1 3 Defective MMR 2 4

Module: derived

Module Contents

d Genomic 1.PERSON ID 2.<u>C MLH1</u> 3.<u>C MSH2</u> 4.<u>C MSH6</u> 5.<u>C PMS2</u> 6.C MUTYH 7.C EPCAM PERSON_ID string (12) Required: FALSE Identifier for person that is globally unique within the C-CFR central database. 1 FK. Consists of concatenation of CENTER_NO (2 digit; digits 1-2) + (10 digit local unique individual id; digits 3-12). The 10 digit individual number component should be right justified, zero filled. Ex: Local unique individual id 98765 from New York Breast Center should be: 020000098765 or 20000098765. Ex: Individual Id 98765 from Australia Colon Center should be: 130000098765. C_MLH1 number (1,0) Required: FALSE 2 Presence of deleterious mutation or unclassified variant in MLH1 gene Allowable Values -8 No data transmitted from centers -9 Failed or equivocal MLPA result or failed sequencing result One of the tests (Genomic or MLPA) has been run, and no deleterious variant or unclassified variant detected -1 1 Both of the tests (Genomics and MLPA) have been run and no deleterious variant or unclassified variant detected One or more deleterious variants detected (Genomic or MLPA) (If 1 should be homozygous) 2 One or more unclassified variants detected (Genomic or MLPA) 3 One or more deleterious AND one or more unclassifed variants detected (Genomic or MLPA) 4 C_MSH2 Required: FALSE number (1,0)3 Presence of deleterious mutation or unclassified variant in MSH2 gene Allowable Values -8 No data transmitted from centers -9 Failed or equivocal MLPA result or failed sequencing result

-1 One of the tests (Genomic or MLPA) has been run, and no deleterious variant or unclassified variant detected

1 Both of the tests (Genomics and MLPA) have been run and no deleterious variant or unclassified variant detected

- 2 One or more deleterious variants detected (Genomic or MLPA) (If 1 should be homozygous)
- **3** One or more unclassified variants detected (Genomic or MLPA)
- 4 One or more deleterious AND one or more unclassifed variants detected (Genomic or MLPA)

4	C_M	SH6		number (1,0)	Required: FAL	SE
	Presence of deleterious mutation or unclassified variant in MSH6 gene					
		Allowable Values				
	[-8	No data transmitted from centers			

-9	Failed or equivocal MLPA result or failed sequencing result			
-1	One of the tests (Genomic or MLPA) has been run, and no deleterious variant or unclassified variant detected			
1	Both of the tests (Genomics and MLPA) have been run and no deleterious variant or unclassified variant detected			
2	One or more deleterious variants detected (Genomic or MLPA) (If 1 should be homozygous)			
3	One or more unclassified variants detected (Genomic or MLPA)			
4	One or more deleterious AND one or more unclassifed variants detected (Genomic or MLPA)			

E	C_P	MS2		number (1,0)	Required: FALSE	
5	Presence of deleterious mutation or unclassified variant in PMS2 gene					
		Allo	wable Values			
		-8	No data transmitted from centers			
		-9	Failed or equivocal MLPA result or failed sequencing result			
		-1	One of the tests (Genomic or MLPA) has been run, and no deleterious variant or u	inclassified variant de	etected	
		1	1 Both of the tests (Genomics and MLPA) have been run and no deleterious variant or unclassified variant detected			
		2	One or more deleterious variants detected (Genomic or MLPA) (If 1 should be hor	nozygous)		
		3	One or more unclassified variants detected (Genomic or MLPA)			
		4	One or more deleterious AND one or more unclassifed variants detected (Genomic	c or MLPA)		

,	C_M	Ιυτγ	H	number (1,0)	Required: FALSE	
O	Presence of deleterious mutation or unclassified variant in MUTYH gene					
		Allowable Values				
		-8	No data transmitted from centers			
		-9	Failed or equivocal MLPA result or failed sequencing result			
		-1	One of the tests (Genomic or MLPA) has been run, and no deleterious variant or	unclassified variant o	letected	
		1	Both of the tests (Genomics and MLPA) have been run and no deleterious variant	or unclassified varia	nt detected	
		2	One or more deleterious variants detected (Genomic or MLPA) (If 1 should be ho	mozygous)		
		3	One or more unclassified variants detected (Genomic or MLPA)			
		4	One or more deleterious AND one or more unclassifed variants detected (Genom	ic or MLPA)		
					1	
7	C_E	PCAI	Л	number (1,0)	Required: FALSE	
1	Presence of deleterious mutation or unclassified variant in EPCAM gene					
Allowable Values			wable Values			
	-8 No data transmitted from centers					
		-9	Failed or equivocal MLPA result or failed sequencing result			
		-1	One of the tests (Genomic or MLPA) has been run, and no deleterious variant or	unclassified variant o	letected	
		1	Both of the tests (Genomics and MLPA) have been run and no deleterious variant or unclassified variant detected			
		2 One or more deleterious variants detected (Genomic or MLPA) (If 1 should be homozygous)				

3	One or more unclassified variants detected (Genomic or MLPA)					
4	One or more deleterious AND one or more unclassifed variants detected (Genomic or MLPA)					