癌症基因檢測檔 欄位名稱及資料描述

(Non-squamous NSCLC, Esophageal cancer, Gastric cancer, Gallbladder cancer, Extrahepatic cholangiocarcinoma, Pancreatic cancer)

1 Report_Patient(一名個案僅一筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
1	hash_id	FMI's unique id for this sample (de-identified)	
2	submitted_diagnosis	Patient's diagnosis submitted with sample	Unknown primary melanoma
3	spec_site	Location from which sample was retrieved	Rectum Blood
4	test_type	Name of the test performed on this sample	FoundationOneDx FoundationOneLiquidDx
5	coll_date	Date when sample was collected	20180320
6	received_date	Date when sample was received	20180330

2 Report_genomic findings (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
7	hash_id	FMI's unique id for this sample (de-identified)	
8	gene_name	Name of gene	KDR AR
9	alteration_property_name	Name of alteration property	Amplification L702H

3 Report_biomarker_findings (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
10	hash_id	FMI's unique id for this sample (de-identified)	
11	report_property	Property key	TumorMutationBurdenScore Microsatellite status
12	value	Value of defined property	3 Muts/Mb MS-Stable

4 Variant_short-variants (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
13	hash_id	FMI's unique id for this sample (de-identified)	
14	transcript	The accession/identifier of the reference transcript used as the basis for attributes like cds-effect and protein-effect	NM_002447 NM_000044
15	gene	The name (gene symbol) of the nearest targeted gene associated with this short variant	MST1R AR
16	position	The genomic position (chromosome name and coordinate) of this short variant; coordinate corresponds to position value in VCF representation	chr3:49939820 chrX:66931463
17	cds_effect	The cds-level effect of this short variant for the associated gene (typically the standard CDS mutant syntax excluding the "c." prefix)	1223C>T 2105T>A
18	protein_effect	The protein-level effect of this short variant for the associated gene (typically either the standard protein mutant syntax, excluding the "p." prefix, for coding variants, or special values starting with "splice" or "promoter")	P408L L702H
19	depth	The local sequencing depth at the genomic position of this short variant	802 1675
20	percent_reads	The percent of sequencing reads at the genomic position of this variant that support the variant allele (For short-variants, this is identical to allele-fraction except represented as a percentage.)	70.57 79.22

5 Variant_copy-number-alterations (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
21	hash_id	FMI's unique id for this sample (de-identified)	
22	gene	The name (gene symbol) of the targeted gene associated with this copy number alteration variant	KDR
23	position	The genomic position (chromosome name and coordinate interval) of this copy number alteration variant	chr4:55896637-56038755
24	ratio	The log (base 2) of the normalized	1.68

序號	英文欄位名稱	資料描述	欄位內容範例
		tumor/reference coverage ratio for the copy number alteration	
25	type	The type of copy number alteration	amplification

6 Variant_rearrangements (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
26	hash_id	FMI's unique id for this sample (de-identified)	
27	othe_gene	The non-targeted or secondary targeted gene, if any, involved in this rearrangement	CNTNAP2
28	percent_reads	The percent of sequencing reads at the genomic position of this variant that support the variant allele. (For rearrangements, this is related to, but not equivalent to, the allele-fraction value. The allele-fraction value is typically the value of interest.)	0.39
29	pos1	The genomic position (chromosome name and coordinate interval) corresponding to the target-gene breakpoint of this rearrangement variant	chr10:43615345
30	pos2	The genomic position (chromosome name and coordinate interval) corresponding to the other-gene breakpoint of this rearrangement variant	chr7:146206742
31	targeted_gene	The targeted gene (or primary targeted gene) involved in this rearrangement	RET

7 Variant_non-human-content (一名個案多筆紀錄)

序號	英文欄位名稱	資料描述	欄位內容範例
32	hash_id	FMI's unique id for this sample (de-identified)	
33	organism	The name of the non-human organism detected	HPV-16
34	reads_per_million	The normalized abundance of sequence read data supporting the non-human content call	15