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	FMI_hash	FMI's unique id for this sample (de-identified)	
2	SubmittedDiagnosis	Patient's diagnosis submitted with sample	Unknown primary melanoma
3	SpecSite	Location from which sample was retrieved	Rectum Blood
4	TestType	Name of the test performed on this sample	FoundationOneDx FoundationOneLiquidDx

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
5	FMI_hash	FMI's unique id for this sample (de-identified)	
6	Gene Name	Name of gene	KDR AR
7	Alteration name	Name of alteration	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
8	FMI_hash	FMI's unique id for this sample (de-identified)	
9	reportProperty	Property key	TumorMutationBurdenScore Microsatellite status
10	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
11	FMI_hash	FMI's unique id for this sample (de-identified)	
12	transcript	The accession/identifier of the reference transcript used as the basis for attributes like cds-effect and protein-effect	NM_002447 NM_000044
13	gene	The name (gene symbol) of the nearest targeted gene associated with this short variant	MST1R AR
14	position	The genomic position (chromosome name and coordinate) of this short variant; coordinate corresponds to position value in VCF representation	chr3:49939820 chrX:66931463
15	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
16	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
17	depth	The local sequencing depth at the genomic position of this short variant	802 1675
18	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
19	FMI_hash	FMI's unique id for this sample (de-identified)	
20	gene	The name (gene symbol) of the targeted gene associated with this copy number alteration variant	KDR
21	position	The genomic position (chromosome name and coordinate interval) of this copy number alteration variant	chr4:55896637-56038755
22	ratio	The log (base 2) of the normalized tumor/reference coverage ratio for the copy number alteration	1.68

序號	英文欄位名稱	資料描述	欄位內容範例
23	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
24	FMI_hash	FMI's unique id for this sample (de-identified)	
25	other-gene	The non-targeted or secondary targeted gene, if any, involved in this rearrangement	CNTNAP2
26	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
27	pos1	The genomic position (chromosome name and coordinate interval) corresponding to the target-gene breakpoint of this rearrangement variant	chr10:43615345
28	pos2	The genomic position (chromosome name and coordinate interval) corresponding to the other-gene breakpoint of this rearrangement variant	chr7:146206742
29	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
30	FMI_hash	FMI's unique id for this sample (de-identified)	
31	organism	The name of the non-human organism detected	HPV-16
32	reads-per-million	The normalized abundance of sequence read data supporting the non-human content call	15