OSIRIS minimum clinical data set for collection in local applications Version 1.0						
Item Group	Objective(s)	Item n°	Collection status	Item	Item definition	Expected value
1. Detection methods and technologies	Methods and technology for the detection of genetic anomalies	1.1	Obligatory	Analysis method	OSIRIS code for the molecular analysis method used	OSIRIS:O6-1 : ChIP-on-Chip OSIRIS:O6-2 : ChIP-seq OSIRIS:O6-3 : DNA microarray OSIRIS:O6-4: Targeted genome sequencing OSIRIS:O6-5 : Exome sequencing OSIRIS:O6-6 : Mass spectrometry OSIRIS:O6-7 : Methylated DNA microarray after immunoprecipitation (MeDIPChip) OSIRIS:O6-8 : Methylated DNA sequencing after immunoprecipitation (MeDIP-seq) OSIRIS:O6-9 : Array for methylation study OSIRIS:O6-10 : Transcriptomic array OSIRIS:O6-11 : Targeted transcriptome sequencing OSIRIS:O6-12 : Whole transcriptome sequencing OSIRIS:O6-13 : Reverse phase protein array (RPPA) OSIRIS:O6-14 : Whole genome sequencing after bisulfate treatment OSIRIS:O6-15 : Whole genome sequencing OSIRIS:O6-16 : MicroRNA
		1.2	Obligatory (condition 1.1)	GEO accession number	Accession number of the molecular analysis method in the GEO (Gene Expression Omnibus) database	GEO accession n° (GPLxxx format)
		1.3	Obligatory (condition 1.1)	Name of technology	Name of the technology used allowing the implementation of the analysis method	OSIRIS technology code
		1.4	Optional	Name of gene panel	In the case of targeted sequencing, the name of the gene panel used. It can be a “home-made” panel or a commercial one.	OSIRIS:O10-0 : Not applicable OSIRIS:O10-1 : Ion AmpliSeq Cancer Hotspot Panel v2 OSIRIS:O10-2 : Ion AmpliSeq Colon and Lung Cancer Research Panel v2 OSIRIS:O10-3 : Ion AmpliSeq Comprehensive Cancer Panel OSIRIS:O10-4 : Ion AmpliSeq Oncomine Comprehensive Assay OSIRIS:O10-5 : Ion AmpliSeq Oncomine Focus Assay OSIRIS:O10-6 : Ion AmpliSeq TP53 Research Panel OSIRIS:O10-7 : Lyric: Ion AmpliSeq Profler OSIRIS:O10-8 : QIAGEN GeneRead DNAseq Targeted Panels V2 OSIRIS:O10-9 : SAFIR02 Panel OSIRIS:O10-10 : Mosc3
		1.5	Obligatory	Name of analysis software	The name of the software allowing the analysis of molecular data from the analysis technology. It can be the name of an ad hoc bioinformatics analysis chain or commercial software.	Character string

2. Genetic profile	Qualification and overall assessment of tumor's genetic profile	2.1	Optional	Percentage of tumor cells	Percentage of tumor cells in the tissue sample	Full number between 0 and 100
		2.2	Optional	Ploidy	Number of complete sets of chromosomes	OSIRIS:O13-1 : Haploid OSIRIS:O13-2 : Diploid OSIRIS:O13-3 : Triploid OSIRIS:O13-4 : Tetraploid OSIRIS:O13-0 : not available
		2.3	Optional	Genomic complexity	Number of breakpoints resulting from chromosomal rearrangements	Whole number
3. Catalogue of genetic cancer alterations	Type of genetic alteration	3.1	Obligatory	Alteration type	OSIRIS code for the genetic alternation type	OSIRIS:O15-1 : Gene expression OSIRIS:O15-2 : Protein expression OSIRIS:O15-3 : Fusion OSIRIS:O15-4 : Copy-number variation OSIRIS:O15-5 : Mutation OSIRIS:O15-6 : Structural modification OSIRIS:O15-7 : Transcription factor OSIRIS:O15-8 : Chromatin modification OSIRIS:O15-9 : Transcript isoform OSIRIS:O15-10 : Viral insertion site
	Version of the reference genome	3.2	Obligatory	Version of the reference genome	The version of the reference genome used to reconstruct the tumor genome	OSIRIS:O22-1 for GRCh37 OSIRIS:O22-2 for GRCh38
	Genomic localization	3.3	Obligatory	Chromosome	Chromosome localization	LOINC code (LAxxxxx-x format)
		3.4	Obligatory	Start position	Start position of the genetic alteration on the chromosome	Whole number (1 to n)
		3.5	Obligatory	End position	End position of the genetic alteration on the chromosome	Whole number (1 to n)
		3.6	Optional	Cytogenetic map	Localization of genomic alteration on the cytogenetic map	UCSC code
4. Mutation(s)	Mutations (condition 3.1 = OSIRIS: O15-5)	4.1	Obligatory	Type	Mutation type	LA6692-3 : Deletion LA6686-5 : Duplication LA6687-3 : Insertion LA6688-1 : Insertion/Deletion LA6689-9 : Inversion LA6690-7 : Substitution OSIRIS:O55-1 : Viral insertion site OSIRIS:O55-2 : Delins
		4.2	Obligatory	Cell type	Cell type where the mutation occurs	LA6683-2 : Germinal LA6684-0 : Somatic LA10429-1 : Prenatal LA18197-6 : Unknown
		4.3	Optional	Zygosity	Zygosity of the mutation	LA6703-8 : Heteroplasmic LA6704-6 : Homoplasmic LA6705-3 : Homozygous LA6706-1 : Heterozygous LA6707-9 : Hemizygous
		4.4	Obligatory	Allele of reference	Allele seen on the strand before the reference genome	Character string
		4.5	Obligatory	Alternative allele	Allele resulting from a somatic mutation event during tumorigenesis	Character string

		4.6	Obligatory	Depth	Number of reads at the genomic mutation position	Whole number
		4.7	Obligatory	Depth of the alternative allele	Number of reads containing the alternative allele at the genomic mutation position	Whole number
		4.8	Optional	DNA strand bias	Is there a DNA strand bias?	Yes No
5. Copy-number variability	Copy-number variability (VNC) (condition 3.1 = OSIRIS:O15-4)	5.1	Obligatory	VNC type	The type of copy-number variation	OSIRIS:O49-1 : Amplifcaton OSIRIS:O49-2 : Gain OSIRIS:O49-3 : Normal OSIRIS:O49-4 : Deletion heterozygous OSIRIS:O49-5 : Deletion homozygous OSIRIS:O49-6 : Neutral copy OSIRIS:O49-7 : Isodisomy
		5.2	Optional	Copy-number	Estimation of the DNA copy-numbers	Whole number
		5.3	Obligatory	Signal intensity	Intensity of measured signal	Decimal number
		5.4	Optional	Loss of heterozygosity (LOH)	Is the fragment in a region with loss of heterozygosity?	Yes No
6. Gene expression	Gene expression (condition 3.1 = OSIRIS:O15-1)	6.1	Obligatory	Quantification type	Quantification used to measure gene expression	OSIRIS:O84-1 : Gross expression value OSIRIS:O84-2 : Normalized expression value
		6.2	Obligatory	Value of expression	Value of gene expression	Decimal number
7. Fusion(s)	Fusion(s) (condition 3.1 = OSIRIS: O15-3)	7.1	Optional	Fusion type	Type of fusion	LA26331-1 : Translocation OSIRIS:O281 : Read-through
		7.2	Obligatory	Gene name at 5’	HUGO symbol for the implicated gene at the 5’ end of the fusion	HUGO symbol
		7.3	Optional	Gene region at 5’	Region name for the implicated gene at the 5’ end of the fusion (for example: exon 3)	Character string
		7.4	Obligatory	Transcript identifier at 5’	RefSeq or Ensembl database identifier at the 5’ end of the fusion	Character string
		7.5	Obligatory	DNA strand at 5’	DNA strand at the 3’ end of the fusion	OSIRIS:O34-1 : + OSIRIS:O34-2 : -
		7.6	Obligatory	Position at 3’	Fusion 3’ genomic position	Whole number
		7.7	Obligatory	Gene name at 3’	HUGO symbol of the implicated gene at the 3’ end of the fusion	HUGO symbol
		7.8	Optional	Gene region at 3’	Name of the implicated gene region at the 3’ end of the fusion (for example: intron 5)	Character string

		7.9	Obligatory	Transcript of identifier at 3'	Transcript RefSeq ou Ensembl database identifier at the 3' end of the fusion	Character string
		7.10	Obligatory	DNA strand at 3'	DNA strand at the 3' end of the fusion	OSIRIS:O34-1 : + OSIRIS:O34-2 : -
		7.11	Obligatory	Position at 3'	Fusion at the 3' position	Whole number
		7.12	Optional	Reading frame	Has the fusion moved the DNA reading frame?	Yes No
		7.13	Obligatory	Fusion depth	Number of read pairs covering the fusion	Whole number
		7.14	Obligatory	Depth of the fusion point	Number of read pairs covering the fusion point	Whole number
8. Molecular annotation(s)	Genetic alteration annotation(s) from molecular biology databases	8.1	Obligatory	Molecular entity	Molecular entity OSIRIS code where the genomic alteration is	OSIRIS:O24-1 : gene OSIRIS:O24-2 : miRNA OSIRIS:O24-3 : piRNA OSIRIS:O24-4 : mRNA OSIRIS:O24-5 : ncRNA OSIRIS:O24-6 : ORF
		8.2 (conditional 8.1)	Obligatory	The database indexing the molecular entity	The OSIRIS code for the database used to name the exact molecular entity	OSIRIS:O25-1 : HGNC OSIRIS:O25-2 : miRDB OSIRIS:O25-3 : Entrez OSIRIS:O25-4 : Human lincRNA Catalog
		8.3 (conditional 8.2)	Obligatory	Molecular entity identifier	The molecular entity identifier in the database	Character string
		8.4	Obligatory (condition 8.1 = OSIRIS:O24-1)	Gene name	Gene symbol following the HUGO (Human Genome Organization) nomenclature	HUGO symbol
9. Genomic annotations	Functional annotation(s) of the genetic alteration of the genome	9.1	Obligatory	Genomic base name	Name of the database referencing the gene containing the genetic alteration	OSIRIS:O91-2 : Ensembl
		9.2	Obligatory	Gene identifier	Gene identifier in the database	Character string
		9.3	Optional	Gene region	Name of the functional gene region containing the genetic alteration (example: exon 3)	Character string
		9.4	Optional	Genomic impact	Impact of the genetic alteration according to the HGVS (Human Genome Variation Society) nomenclature (example g.289G>A)	Character string (HGVS documentation)
		9.5	Optional	Impact on the gene coding region	If the genetic alteration is on a gene coding region, indicate the impact on this region according to the HGVS (Human Genome Variation Society) nomenclature (example: c.12G>A)	Character string (HGVS documentation)
10. Transcriptomic annotation(s)	Functional annotation(s) of the genetic alteration on the transcriptome	10.1	Obligatory	Name of the transcriptomic database	Name of the database referencing the transcript containing the genetic alteration	OSIRIS:O91-1 : RefSeq OSIRIS:O91-2 : Ensembl
		10.2	Obligatory	Gene identifier	Transcript identifier in the database	Character string

		10.3	Optional	Impact on the transcript	Impact of the genetic alteration according to the HGVS (Human Genome Variation Society) nomenclature (example : r.67g>u)	Character string (HGVS documentation)
11. Proteomic annotation(s)	Functional annotation(s) of the genetic alteration on the proteome	11.1	Obligatory	Name of the protein database	Name of the database referencing the protein containing the genetic alteration	OSIRIS:O91-3 : SwissProt OSIRIS:O91-4 : UniProt
		11.2	Obligatory	Protein identifier	Protein identifier in the database	Character string
		11.3	Optional	Impact on the protein	Impact of the genetic alteration according to the HGVS (Human Genome Variation Society) (example: p.Arg2322Cys)	Character string (HGVS documentation)
		11.4	Optional	Pfam identifier	Identifier containing the genetic alteration in the protein families database (Pfam)	Pfam identifier
		11.5	Optional	Pfam domain name	Name of the protein domain containing the genetic alteration in the Pfam database	Character string
12. Validation method(s)	Pertinence of a genetic alteration	12.1	Obligatory	Validation type	Validation method used	OSIRIS:O16-1 : Experimentally OSIRIS:O16-2 : By a biologist OSIRIS:O16-3 : Computationally
		12.2	Obligatory	Validation status	Indicates if the alteration was validated or not using the validation method	OSIRIS:O18-1 : Not Validated OSIRIS:O18-2 : Validated
		12.3	Optional	Validation method	Indicates a validation method (example bibliographically)	Character string
13. Genomic profile / Molecular RCP	Identify candidate genomic alterations for a Multidisciplinary Coordination Meeting (RCP)	13.1	Optional	Prediction software	Name of the software used to predict the impact of the alteration on the protein	OSIRIS:O63-1 : SIFT OSIRIS:O63-2 : POLYPHEN2_HDIV
		13.2	Optional	Confidence rating (condition 13.1)	Confidence rating given by the software	
		13.3	Optional	Pathogenicity	Information indicating the pathogenicity of the genetic alteration	OSIRIS:O81-1 : Pathogenic OSIRIS:O81-2 : Probably pathogenic OSIRIS:O81-3 : Benign OSIRIS:O81-4 : Probably benign OSIRIS:O81-5 : Uncertain significance
		13.4	Optional	Impact on patient treatment	Can/could the genetic alteration affect patient response to a treatment?	OSIRIS:O82-1 : Yes OSIRIS:O82-2 : No
		13.5	Optional	Impact on therapeutic choice	Has the genetic alteration been used to orient the patient's treatment choice	Yes No