# ICGC Data Submission Manual

**Document Version 0.5d**
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# 1. Overview of data submission process

There are three major steps in the data submission process:

a. Submit raw sequence data to the European Genome-phenome Archive (**Appedix C**).
b. Prepare the ICGC submission files according to DCC specifications (**Section 2**).
c. Verify conformity of the submission files (**Section 3**).
d. Submit files to the DCC Secure FTP server (**Section 4**).

NB. All submitted data must be based on human reference genome assembly GRCh37 and Ensembl gene set version 61.

NB. Please include sample EGA accession in the ICGC Sample Data file (page 36, data element *sample_ega_accession*).

Please contact DCC (dcc-support@lists.oicr.on.ca) if you would like to set up an ICGC node or if you have any questions or comments about the data submission process.

# 2. Submission file format

- The submission data are kept in tab-limited text files. Comments may be added to the beginning of the file with a hash ('#') prefixed at beginning of each comment line. The first non-comment line is the header containing the names of the columns. Each column corresponds to a data element defined in DCC Data Element specification (**Appendix A**).

An example file is shown below (note that parts of the lines are omitted for readability):

```
# This is an example of a primary analysis file for simple somatic mutations.
# File name: ssm__ca__01__068__p__8__20090713.txt
#
tumour_sample_id    mutation_id     mutation_type   chromosome  …   note
m124                ssm_3396649     1               1           …   -999
m124                ssm_61023021    1               2           …   -999
m124                ssm_175270973   1               3           …   -999
m124                ssm_72390475    1               4           …   -999
```

- Types of experimental data being supported:
  - simple mutations/variations of ≤ 200 bp
  - copy number mutations/variations
  - structural mutations/variations
  - gene expression

- miRNA
- exon junctions
- DNA methylation

- File Format:

The input file formats are provided below. The files containing experiment results need to comply with the following naming convention (note the use of double underscores ('_') to separate components in the file name):

*featureType__leadJurisdiction__tumourType__institution__fileType__platform__dateFileCreated.txt*

The components of the file name are listed below:

| Components | Description | Values |
|---|---|---|
| *featureType* | Simple somatic mutations including single base substitutions and indels of ≤200 bp | ssm |
| | Simple germline variations including single base substitutions and indels of ≤200 bp | sgv |
| | Copy number somatic mutations | cnsm |
| | Copy number germline variations | cngv |
| | Structural somatic mutations | stsm |
| | Structural germline variations | stgv |
| | Gene and exon expression | exp |
| | miRNA expression | mirna |
| | Exon junction | jcn |
| | Methylation | meth |
| *leadJurisdiction* | Jurisdiction leading the project | Appendix Table B1 |
| *tumourType* | tumour type | Appendix Table B2 |
| *institution* | Institution submitting the data | Appendix Table B3 |
| *fileType* | Primary analysis file | p |
| | Secondary analysis file | s |
| | Metadata file | m |
| | Gene expression file | g |
| | Exon expression file | e |
| *platform* | Platform or technology used in the analysis | Appendix Table B5 |
| *dateFileCreated* | The date on which the file is created | YYYYMMDD |

The file names for donor, diagnosis and sample information follow the convention (note the use of double underscores ('_') to separate components in the file name):

*leadJurisdiction__tumourType__institution__fileType__dateFileCreated.txt*

The components of the file name are listed below:

| Components | Description | Values |
|---|---|---|
| *leadJurisdiction* | Jurisdiction leading the project | Appendix Table B1 |
| *tumourType* | tumour type | Appendix Table B2 |
| *institution* | Institution submitting the data | Appendix Table B3 |
| *fileType* | Donor information | donor |
| | Diagnosis information | diagnosis |
| | Sample information | sample |
| *dateFileCreated* | The date on which the file is created | YYYYMMDD |

For examples of the file names see below:

| Examples | Description |
|---|---|
| ssm__ca__01__068__p__8__20090713.txt | In pancreatic cancer project, the primary analysis file generated on July 13, 2009 by OICR (Canada) for simple somatic mutations analyzed on Affymetrix Genome-Wide Human SNP Array 6.0 |
| cngv__ca__01__068__m__8__20090713.txt | In pancreatic cancer project, the metadata file generated on July 13, 2009 by OICR (Canada) for copy number germline variations analyzed on Affymetrix Genome-Wide Human SNP Array 6.0 |
| ca__01__068__donor__20090713.txt | In pancreatic cancer project, donor information provided by OICR (Canada) on July 13, 2009 |
| ca__01__068__sample__20090713.txt | In pancreatic cancer project, sample information provided by OICR (Canada) on July 13, 2009 |

# 3. Submission file validation

- For the purpose of validating the submission files, download MartLoader (software tool for processing ICGC data) from DCC's SVN server as below (you may change */home/software* to another local path):

```
cd /home/software
svn co https://code.oicr.on.ca/svn/dcc/martloader/branches/release-0_5_i5  martloader
```

- Create a work directory using a name of your choice (e.g. *workdir_testSept10*) for keeping all submission files:

```
cd /home/software/martloader
perl createWorkDir.pl workdir_testSept10
```

- Put all of the submission files into the appropriate subfolders under *'workdir_testSept10/input'* folder. The subfolders are listed as below:

    a. cnv (copy number variation)
    b. exp (expression)
    c. jcn (exon junction)
    d. meth (methylation)
    e. mirna (microRNA)
    f. sample (sample)
    g. snp (simple mutation/variation)
    h. sv (structural mutation/variation)

    A set of example input files can be found under the 'workdir_test/input' directory.

- Run data validation as below:

| **cd */home/software/*martloader/*workdir_testSept10*** |
|---|
| **perl runme.pl -c** |

- When validation finishes, please review the log files under */home/software/*martloader/*workdir_testSept10/logs*. Empty log files (0 bytes) can be safely ignored. Otherwise, review the messages in the log files. After making any necessary changes to the submission files, please rerun data validation.

## 4. File submission

- After the submission files have passed validation check, the files should be compressed and uploaded to DCC's Secure FTP server (data.dcc.icgc.org).

- Contact DCC if you need an SFTP account for file uploading or if you experience any difficulty with the SFTP server.

## 5. Setup BioMart server (optional)

As an alternative to submitting data to DCC, you can setup the data server on your own side by following the steps below:

- Install and configure MySQL database server and create necessary MySQL user account.

- Create a text file named 'dbuser' under /home/software/martloader/bin/, an example file is shown below:

    host=your_host.com
    port=3306

user=your_user_name
pass=password

Please note that this MySQL account needs to have permission to create databases.

- Run data loading as below:

| **cd */home/software/*martloader/*workdir_testSept10*** |
| --- |
| **perl runme.pl -l** |

**Important**: with the above command, martloader will **delete** a MySQL database named *dcc_testSept10* if it exists, and it will create a new *dcc_testSept10* database and populate it with data transformed from submission files.

Once martloader finishes, please review the log files under */home/software/*martloader/*workdir_testSept10/logs*. Empty log files (0 bytes) can be safely ignored. Otherwise, review the messages in the log files. After making any necessary changes to the submission files, please rerun data loading again.

After data successfully loaded in the previous step, please consult the ***Preconfigure Portal Deployment*** section in the ***User Manual*** (available from http://www.biomart.org/rc6_documentation.pdf) of BioMart 0.8 release candidate 6, for configuring and setting up the BioMart server.

# Appendix A: DCC Data Element Specification

Please do not leave any data elements empty in the submission files. Besides the possible values detailed in the tables below, values can also be one of the these codes:

-999 = data not supplied at this time
-888 = not applicable
-777 = data verified to be unknown

Legend: R = required, O = optional

## 1. Simple Somatic Mutations/Simple Germline Variations (SSM/SGV)

SSM and SGV include single base substitutions, multiple base substitutions (> 1bp and ≤200 bp) and short indels of ≤200 bp in length.

**Simple Somatic Mutations (SSM) - Metadata File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | tumour_sample_id | Unique identifier for the tumour sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the control matched to the tumour sample | | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |

| | | | | | |
|---|---|---|---|---|---|
| 16 | O | note | Optional field to leave notes | text | |

## Simple Somatic Mutations (SSM) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |
| 3 | R | mutation_id | Unique identifier for the mutation | | |
| 4 | R | mutation_type | Type of mutation | integer | 1 = single base substitution<br>2 = insertion of <= 200 bp<br>3 = deletion of <= 200 bp<br>4 = multiple base substitution (>= 2bp and <= 200bp) |
| 5 | R | chromosome | Name of the chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 6 | R | chromosome_start | Start position of the mutation/variation on the chromosome | integer | |
| 7 | R | chromosome_end | End position of the mutation/variation on the chromosome | integer | |
| 8 | R | chromosome_strand | Chromosome strand | integer | 1 = 1<br>-1 = -1 |
| 9 | R | refsnp_allele | RefSNP alleles from dbSNP (use a dash for each missing base) | text | e.g. A/T, ---/AAA |
| 10 | O | refsnp_strand | Strand of RefSNP allele | integer | 1 = 1<br>-1 = -1 |
| 11 | R | reference_genome_allele | Allele in the reference genome (use a dash for each missing base) | text | |
| 12 | R | control_genotype | Genotype of the control sample (use a dash for each missing base) | text | |
| 13 | R | tumour_genotype | Genotype of the tumour sample (use a dash for each missing base) | text | |
| 14 | R | mutation | Mutation, e.g. C > G | text | |
| 15 | O | expressed_allele | The expressed allele(s) as revealed by RNA-seq, etc. | text | |
| 16 | O | quality_score | Average quality score for the mutation/variation call | integer | |
| 17 | O | probability | Probability of the mutation/variation call | decimal | |
| 18 | O | read_count | Average number of times the bases are covered by raw reads | decimal | |
| 19 | O | is_annotated | Indicate if the mutation/variation is annotated in dbSNP | integer | 1 = annotated<br>2 = not annotated |
| 20 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | 3 = not valid |
| 21 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 22 | O | xref_ensembl_var_id | Cross-reference: Ensembl Variation ID | text | Variation ID in Ensembl Variation Database: e.g. rs12345; ENSSNP53189 |
| 23 | O | note | Optional field to leave notes | text | |

## Simple Somatic Mutations (SSM) – Secondary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |
| 3 | R | mutation_id | Unique identifier for the mutation | | |
| 4 | R | consequence_type | Functional consequence of the SNP. | integer | Appendix Tables B7 & B8 |
| 5 | O | aa_mutation | Changes at amino acid level. Indicate the reference aa, position and mutation aa. | text | e.g. P234W |
| 6 | O | cds_mutation | Changes in coding sequence. Indicate position, reference base and mutation base. | text | e.g. 12324T>G |
| 7 | O | protein_domain_affected | Protein domain containing the mutation/variation. Use Pfam accession. | text | |
| 8 | O | gene_affected | Gene(s) containing the mutation/variation. | text | |
| 9 | O | transcript_affected | Transcript(s) containing the mutation/variation. Use Ensembl transcript id. | text | |
| 10 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | 55 |
| 11 | O | note | Optional field to leave notes | text | |

**Note:** when a mutation affects more than one transcript, please use multiple rows to record the mutation consequence, one row per transcript.

## Simple Germline Variations (SGV) – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | control_sample_id | Unique identifier for the control sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the tumour matched to the control sample | | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

## Simple Germline Variations (SGV) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | control_sample_id | Unique identifier for the control sample | | |
| 3 | R | variation_id | Unique identifier for the variation | | |
| 4 | R | variation_type | Type of variation | integer | 1 = single base substitution<br>2 = insertion of <= 200 bp<br>3 = deletion of <= 200 bp<br>4 = multiple base substitution (>= 2bp and <= 200bp) |
| 5 | R | chromosome | Name of the chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 6 | R | chromosome_start | Start position of the mutation/variation on the chromosome | integer | |
| 7 | R | chromosome_end | End position of the mutation/variation on the chromosome | integer | |
| 8 | R | chromosome_strand | Chromosome strand | integer | 1 = 1<br>-1 = -1 |
| 9 | R | refsnp_allele | RefSNP alleles from dbSNP (use a dash for each missing base) | text | e.g. A/T, ---/AAA |
| 10 | O | refsnp_strand | Strand of RefSNP allele | integer | 1 = 1 |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | -1 = -1 |
| 11 | R | reference_genome_allele | Allele in the reference genome (use a dash for each missing base) | text | |
| 12 | R | control_genotype | Genotype of the control sample (use a dash for each missing base) | text | |
| 13 | R | tumour_genotype | Genotype of the tumour sample (use a dash for each missing base) | text | |
| 14 | O | expressed_allele | The expressed allele(s) as revealed by RNA-seq, etc. | text | |
| 15 | O | quality_score | Average quality score for the mutation/variation call | integer | |
| 16 | O | probability | Probability of the mutation/variation call | decimal | |
| 17 | O | read_count | Average number of times the bases are covered by raw reads | decimal | |
| 18 | O | is_annotated | Indicate if the mutation/variation is annotated in dbSNP | integer | 1 = annotated<br>2 = not annotated |
| 19 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 20 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 21 | O | xref_ensembl_var_id | Cross-reference: Ensembl Variation ID | text | Variation ID in Ensembl Variation Database: e.g. rs12345; ENSSNP53189 |
| 22 | O | note | Optional field to leave notes | text | |

**Further explanations for the following data elements in the Simple Mutation Dataset:**

**a. chromosome, chromosome_start, chromosome_end**

- Nucleotide position in DNA sequence is expected to start from 1 for the first nucleotide of the forward strand, counting one by one up to the end.
- For any feature on the genome, chromosome_start is always less than or equal to chromosome_end.
- The size of a feature is calculated as: chromosome_end - chromosome_start + 1.
- For single nucleotide substitution, use the coordinate of the mutated nucleotide to report the mutation, e.g. chromosome:chr1, chromosome_start:12345, chromosome_end:12345.
- For multiple nucleotide substitution (≥2bp and ≤200bp), use the start and end coordinates of the mutated fragment, e.g. chromosome: chr1, chromosome_start: 12345, chromosome_end: 12355 for a 11bp substitution.
- For deletion, use the coordinates of the deleted fragment. e.g. chr1:12345-12355 is an 11 bp deletion from 12345 to 12355 on chromosome 1.
- For insertion, use the coordinate of the nucleotide that is immediately after the insertion point. e.g. an insertion at chr1:12345-12345 means that a fragment of DNA sequence is inserted immediately before position 12345 on chromosome 1.

**b. chromosome_strand**

- 'chromosome_strand' is used to record the reference genome strand on which the genotype alleles are located.
- For genotype detected using sequencing platforms, the forward strand sequence is used for genotypes, so chromosome_strand is always forward (i.e. 1).
- For genotype that is called using array based platforms, chromosome_strand can be either forward or reverse depending on what is reported by the assay.
- 'chromosome_strand' does not have anything to do with the strandness of the gene that contains the simple mutation.

**c. mutation_type**

- 1 = single base substitution
- 2 = insertion of <= 200 bp
- 3 = deletion of <= 200 bp
- 4 = multiple base substitution (>= 2bp and <= 200bp)

**d. control_genotype, tumour_genotype**

- Genotype is presented as nucleotide sequence all allele(s). For example, in a diploid genome at chr1:12345-12345, if one allele on the forward strand is A and the other is G, then the genotype is presented as A/G and 'chromosome_strand' being '1' (i.e. forward strand). It may also be presented as T/C with 'chromosome_strand' being '-1' (i.e. reverse strand).
- In the case that the genotype is hemizygous (e.g. G allele is missing), it can be presented as A/-.
- 'control_genotype' and 'tumour_genotype' are used to record genotype for the matched control sample and the primary tumour sample, respectively. Both genotypes must be presented using the same strand of the reference genome.
- Usually, genotypes in control samples are homozygous, and the nucleotides are the same as the reference genome. For example, at chr1:456789-456789, both alleles are A as in the reference genome, so the control genotype should be A/A.
- Due to aneuploidy and normal tissue contamination, it can be difficult to determine zygosity of tumour samples. In the previous example, the genotype of the tumour sample may be G/G but may appear as A/G when the sample is contaminated. If the tumour genotype can not be determined, please use -777 to indicate 'data verified to be unknown'.

**e. mutation**

- 'mutation' records the somatic mutation in the tumour sample.
- For mutation on a single allele, provide the control and tumour sample alleles separated by '>'. For example, 'A>G' indicates that one allele has an A to G mutation (single nucleotide substitution) in the tumour sample.
- In the case that both alleles are mutated, provide the control genotype and tumour genotype separated by '>', e.g. 'A/T>C/G'.
- For multiple nucleotide substitution (≥2bp and ≤200bp), provide the nucleotide sequences in the control and tumour sample alleles separated by '>', e.g. 'CTGAG>AGCCT'.

- For deletions, '-' is expected to represent each missing nucleotide, for example, at chr1:1234-1236, three nucleotides ATG are missing in the tumour sample, it is expressed as 'ATG>---'.
- For insertions, e.g. a DNA fragment 'CTGAG' inserted before nucleotide 'T' at chr1:12345-12345 can be presented as '->CTGAG'.

**f. reference_genome_allele**

- 'reference_genome_allele' is the forward strand nucleotide(s) at the corresponding location on the reference genome where the somatic mutation is detected in the tumour sample.

**g. refsnp_alleles**

- At the genomic location of a somatic mutation, if a refSNP entry is found in dbSNP database, the alleles described in that refSNP should be presented in 'refsnp_alleles'.
- When no refSNP is presented in dbSNP, use '-777' to indicate 'data verified to be unknown'.

**h. refsnp_strand**

- If a refSNP is presented, its strandness compared with reference genome assembly should be recorded in 'refsnp_strand'. For example, rs72466451 is located at chr2:198363487-198363487, the alleles are presented using reserve strand (i.e. "-1").

**i. is_annotated**

- 'is_annotated' indicates whether a SNP is known at the location of the reported mutation.
- If a SNP is present in dbSNP, please use 'annotated', otherwise use 'not annotated'.
- For mutation detected using array based platforms, the SNP should be 'annotated' since the microarray probes are designed from known SNPs.

## 2. Copy Number Somatic Mutations/Copy Number Germline Variations (CNSM/CNGV)

**Copy Number Somatic Mutations (CNSM) – Metadata File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | tumour_sample_id | Unique identifier for the tumour sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the control matched to the tumour sample | | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | O | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | O | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | O | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

## Copy Number Somatic Mutations (CNSM) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |
| 3 | R | mutation_id | Unique identifier for the mutation | | |
| 4 | R | mutation_type | Type of mutation | integer | 1 = gain<br>2 = loss<br>3 = copy neutral LOH |
| 5 | R | chromosome | Name of the chromosome containing the mutation/variation (#) | integer | Appendix Table B6 |
| 6 | R | chromosome_start | Start position of the mutation/variation on the chromosome | integer | |
| 7 | R | chromosome_end | End position of the mutation/variation on the chromosome | integer | |
| 8 | O | chromosome_start_range | Number of bases around chromosome_start that may contain the start position | integer | 0 if start position is exactly at chromosome_start;<br>positive integer for +/- number of bases around chromosome_start |

| | | | | | |
|---|---|---|---|---|---|
| 9 | O | chromosome_end_range | Number of bases around chromosome_end that may contain the end position | integer | 0 if end position is exactly at chromosome_end ;<br>positive integer for +/- number of bases around chromosome_end |
| 10 | O | start_probe_id | Probe id containing the chromosome_start if array platform was used | text | |
| 11 | O | end_probe_id | Probe id containing the chromosome_end if array platform was used | text | |
| 12 | O | copy_number | DNA copy number estimated | decimal | |
| 13 | O | segment_mean | Mean LRR per segment | decimal | |
| 14 | O | segment_median | Median LRR per segment | decimal | |
| 15 | O | quality_score | Quality score for the mutation/variation call | decimal | |
| 16 | O | probability | Probability of the mutation/variation call | decimal | |
| 17 | O | is_annotated | Indicate if the mutation/variation is annotated in the Database of Genomic Variants | integer | 1 = annotated<br>2 = not annotated |
| 18 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 19 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 20 | O | note | Optional field to leave notes | text | |

## Copy Number Somatic Mutations (CNSM) – Secondary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |
| 3 | R | mutation_id | Unique identifier for the mutation | | |
| 4 | R | gene_affected | Gene(s) containing the mutation/variation. Use Ensembl gene id. Separate multiple genes with vertical bars in the form of geneA\|geneB\|geneC. If no gene is affected, use -888 (not applicable). | text | |

| | | | | | |
|---|---|---|---|---|---|
| 5 | O | transcript_affected | Transcript(s) containing the mutation/variation. Use Ensembl transcript id. Separate multiple transcripts from the same gene with commas, and separate transcripts from different genes with vertical bars. eg. transcriptA1, transcriptA2\|transcriptB1\|transcriptC1,transcriptC2,transcriptC3. If no transcript is affected, use -888 (not applicable). | text | |
| 6 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | |
| 7 | O | note | Optional field to leave notes | text | |

## Copy Number Germline Variations (CNGV) – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | control_sample_id | Unique identifier for the control sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the tumour matched to the control sample | | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | O | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | O | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | O | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

## Copy Number Germline Variations (CNGV) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | control_sample_id | Unique identifier for the control sample | | |
| 3 | R | variation_id | Unique identifier for the variation | | |
| 4 | R | variation_type | Type of variation | integer | 1 = gain<br>2 = loss<br>3 = copy neutral LOH |
| 5 | R | chromosome | Name of the chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 6 | R | chromosome_start | Start position of the mutation/variation on the chromosome | integer | |
| 7 | R | chromosome_end | End position of the mutation/variation on the chromosome | integer | |
| 8 | O | chromosome_start_range | Number of bases around chromosome_start that may contain the start position | integer | 0 if start position is exactly at chromosome_start;<br>positive integer for +/- number of bases around chromosome_start |
| 9 | O | chromosome_end_range | Number of bases around chromosome_end that may contain the end position | integer | 0 if end position is exactly at chromosome_end;<br>positive integer for +/- number of bases around chromosome_end |
| 10 | O | start_probe_id | Probe id containing the chromosome_start if array platform was used | text | |
| 11 | O | end_probe_id | Probe id containing the chromosome_end if array platform was used | text | |
| 12 | O | copy_number | DNA copy number estimated | decimal | |
| 13 | O | segment_mean | Mean LRR per segment | decimal | |
| 14 | O | segment_median | Median LRR per segment | decimal | |
| 15 | O | quality_score | Quality score for the mutation/variation call | decimal | |
| 16 | O | probability | Probability of the mutation/variation call | decimal | |
| 17 | O | is_annotated | Indicate if the mutation/variation is annotated in the Database of Genomic Variants | integer | 1 = annotated<br>2 = not annotated |

| | | | | | |
|---|---|---|---|---|---|
| 18 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 19 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 20 | O | note | Optional field to leave notes | text | |

## 3. Structural Somatic Mutations/Structural Germline Variations (StSM/StGV)

### Structural Somatic Mutations (StSM) – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | text | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | text | |
| 4 | R | tumour_sample_id | Unique identifier for the tumour sample donated by the donor | text | |
| 5 | R | matched_sample_id | Unique identifier for the control matched to the tumour sample | text | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

### Structural Somatic Mutations (StSM) – Primary Analysis File

| Order | O/R | Data element | Description | Data | Values |
|---|---|---|---|---|---|

| | | | | type | |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | text | |
| 3 | R | sv_id | Unique variant id (institute wide). One id per single event | text | |
| 4 | R | placement | Ordering of breakpoint pairs within a single structural mutation/variation event | integer | |
| 5 | R | annotation | Annotation describing sequence mutation/variation based on breakpoint pairs | text | |
| 6 | O | interpreted_annotation | HGVS nomenclature for description of sequence mutation/variation. E.g. chr3:g.1234567-2345678inv. | text | |
| 7 | R | variant_type | Type of mutation/variation | integer | Appendix Table B9 |
| 8 | R | chr_from | Name of the donor chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 9 | R | chr_from_bkpt | Breakpoint position of the mutation/variation on the donor chromosome | integer | |
| 10 | R | chr_from_strand | Donor chromosome strand | integer | 1 = 1<br>-1 = -1 |
| 11 | O | chr_from_range | Number of bases around chr_from_bkpt that may contain the real breakpoint | integer | |
| 12 | O | chr_from_flanking_seq | Flanking sequences that are 200bp upstream and 200bp downstream to the chr_from_bkpt position. | text | |
| 13 | R | chr_to | Name of the acceptor chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 14 | R | chr_to_bkpt | Breakpoint position of the mutation/variation on the acceptor chromosome | integer | |
| 15 | R | chr_to_strand | Acceptor chromosome strand | integer | 1 = 1<br>-1 = -1 |
| 16 | O | chr_to_range | Number of bases around chr_to_bkpt that may contain the real breakpoint | integer | |
| 17 | O | chr_to_flanking_seq | Flanking sequences that are 200bp upstream and 200bp downstream to the chr_to_bkpt position. | text | |
| 18 | O | microhomology_sequence | If a microhomology is inserted, provide sequence | text | |
| 19 | O | non_templated_sequence | If non-templated DNA is inserted, provide sequence | text | |
| 20 | O | evidence | Evidence supporting a structural mutation/variation | integer | 1 = Copy number change<br>2 = FISH<br>3 = Flow-sorted chromosome |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | evidence<br>4 = Paired sequence either side of breakpoint<br>5 = Partner breakpoint found<br>6 = PCR product across breakpoint<br>7 = Protein evidence<br>8 = Seen in multiple samples<br>9 = Sequence across breakpoint |
| 21 | O | quality_score | Quality score for the mutation/variation call | integer | |
| 22 | O | probability | Probability of the mutation/variation call | decimal | |
| 23 | O | zygosity | Zygosity | integer | 1 = homozygous<br>2 = heterozygous<br>3 = hemizygous<br>4 = nullizygous |
| 24 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 25 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 26 | O | db_xref | Value code of cross-reference database:ID of the mutation in the cross-reference database. Separate multiple entries by commas. | text | |
| 27 | O | note | Optional field to leave notes | text | |

## Structural Somatic Mutatiosn (StSM) – Secondary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | text | |
| 3 | R | sv_id | Unique variant id (institute wide). One id per single event | text | |
| 4 | R | placement | Ordering of breakpoint pairs within a single structural change event | integer | |
| 5 | O | bkpt_from_context | Contextual description of the first break location (Exonic, Intronic, Intergenic) | text | |
| 6 | O | gene_affected_by_bkpt_from | Gene(s) affected by the breakpoints. Use Ensembl gene id. Separate multiple genes with vertical bars in the form of geneA\|geneB\|geneC. If both breakpoints | text | |

| | | | | | |
|---|---|---|---|---|---|
| | | | affect genes, then use "\|" to separate them. If no gene is affected, use -888 (not applicable). | | |
| 7 | O | transcript_affected_by_bkpt_from | Transcript(s) affected by the breakpoints. Use Ensembl transcript id. Separate multiple transcripts from the same gene with commas, and separate transcripts from different genes with vertical bars. eg. transcriptA1, transcriptA2\|transcriptB1\|transcriptC1 | text | |
| 8 | O | bkpt_to_context | Contextual description of the second break location (Exonic, Intronic, Intergenic) | text | |
| 9 | O | gene_affected_by_bkpt_to | Gene(s) affected by the breakpoints. Use Ensembl gene id. Separate multiple genes with vertical bars in the form of geneA\|geneB\|geneC. If both breakpoints affect genes, then use "\|" to separate them. If no gene is affected, use -888 (not applicable). | text | |
| 10 | O | transcript_affected_by_bkpt_to | Transcript(s) affected by the breakpoints. Use Ensembl transcript id. Separate multiple transcripts from the same gene with commas, and separate transcripts from different genes with vertical bars. eg. transcriptA1, transcriptA2\|transcriptB1\|transcriptC1 | text | |
| 11 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | |
| 12 | O | note | Optional field to leave notes | text | |

## Structural Germline Variations (StGV) – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | control_sample_id | Unique identifier for the control sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the tumour matched to the control sample | | |
| 6 | R | assembly_version | Version of reference genome assembly (#) | integer | Appendix Table B10 |
| 7 | R | platform | Platform or technology used in detecting the mutation/variation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and | text/url | |

| | | | | | |
|---|---|---|---|---|---|
| | | | URL to written protocol | | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA<br>2 = dbSNP |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

## Structural Germline Variations (StGV) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | control_sample_id | Unique identifier for the control sample | text | |
| 3 | R | sv_id | Unique variant id (institute wide). One id per single event | text | |
| 4 | R | placement | Ordering of breakpoint pairs within a single structural mutation/variation event | integer | |
| 5 | R | annotation | Annotation describing sequence mutation/variation based on breakpoint pairs | text | |
| 6 | O | interpreted_annotation | HGVS nomenclature for description of sequence mutation/variation. E.g. chr3:g.1234567-2345678inv. | text | |
| 7 | R | variant_type | Type of mutation/variation | integer | Appendix Table B9 |
| 8 | R | chr_from | Name of the donor chromosome containing the mutation/variation | integer | Appendix Table B6 |
| 9 | R | chr_from_bkpt | Breakpoint position of the mutation/variation on the donor chromosome | integer | |
| 10 | R | chr_from_strand | Donor chromome strand | integer | 1 = 1<br>-1 = -1 |
| 11 | O | chr_from_range | Number of bases around chr_from_bkpt that may contain the real breakpoint | integer | |
| 12 | O | chr_from_flanking_seq | Flanking sequences that are 200bp upstream and 200bp downstream to the chr_from_bkpt position. | text | |
| 13 | R | chr_to | Name of the acceptor chromosome containing the mutation/variation | integer | Appendix Table B6 |

| | | | | | |
|---|---|---|---|---|---|
| 14 | R | chr_to_bkpt | Breakpoint position of the mutation/variation on the acceptor chromosome | integer | |
| 15 | R | chr_to_strand | Acceptor chromome strand | integer | 1 = 1<br>-1 = -1 |
| 16 | O | chr_to_range | Number of bases around chr_to_bkpt that may contain the real breakpoint | integer | |
| 17 | O | chr_to_flanking_seq | Flanking sequences that are 200bp upstream and 200bp downstream to the chr_to_bkpt position. | text | |
| 18 | O | microhomology_sequence | If a microhomology is inserted, provide sequence | text | |
| 19 | O | non_templated_sequence | If non-templated DNA is inserted, provide sequence | text | |
| 20 | O | evidence | Evidence supporting a structural mutation/variation | integer | 1 = Copy number change<br>2 = FISH<br>3 = Flow-sorted chromosome evidence<br>4 = Paired sequence either side of breakpoint<br>5 = Partner breakpoint found<br>6 = PCR product across breakpoint<br>7 = Protein evidence<br>8 = Seen in multiple samples<br>9 = Sequence across breakpoint |
| 21 | O | quality_score | Quality score for the mutation/variation call | integer | |
| 22 | O | probability | Probability of the mutation/variation call | decimal | |
| 23 | O | zygosity | Zygosity | integer | 1 = homozygous<br>2 = heterozygous<br>3 = hemizygous<br>4 = nullizygous |
| 25 | R | validation_status | Indicate if the mutation/variation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 26 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 27 | O | db_xref | Value code of cross-reference database:ID of the mutation in the cross-reference database. Separate multiple entries by commas. | text | |
| 28 | O | note | Optional field to leave notes | text | |

## 4. Gene Expression

### Expression – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 5 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table B10 |
| 6 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | |
| 7 | R | platform | Platform or technology used in detecting the expression | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | normalization_algorithm | Name of normalization algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

### Expression – Gene File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 3 | R | gene_stable_id | For annotated gene, use Ensembl gene | text | |

| | | | | | |
|---|---|---|---|---|---|
| | | | ID. Otherwise, use assemblyBuild_chr_start_end where assemblyBuild is hg18 or hg19. | | |
| 4 | R | gene_chromosome | Name of the chromosome containing the mRNA | integer | Appendix Table B6 |
| 5 | R | gene_strand | Strand of the chromosome | integer | 1 = 1<br>-1 = -1 |
| 6 | R | gene_start | Start position of the gene on the chromosome | integer | |
| 7 | R | gene_end | End position of the transcript on the chromosome | integer | |
| 8 | R | normalized_read_count | Normalized count of sequencing reads if analyzed by sequencing platforms | decimal | |
| 9 | R | raw_read_count | Raw count of sequencing reads if analyzed by sequencing platforms | integer | |
| 10 | O | normalized_expression_level | Normalized value of expression level if analyzed by microarray platforms | decimal | |
| 11 | O | fold_change | Expressed fold change if differential expression is measured | decimal | |
| 12 | O | reference_sample | ID of the reference sample if differential expression is measured | text | |
| 13 | O | quality_score | Quality score for the expression call | integer | |
| 14 | O | probability | Probability of the expression call | decimal | |
| 15 | O | is_annotated | Indicate if the expressed fragment is annotated in Ensembl | integer | 1 = annotated<br>2 = not annotated |
| 16 | R | validation_status | Indicate if the expressed fragment has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 17 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 18 | O | probeset_id | ID of the probeset used in microarray | test | |
| 19 | O | note | Optional field to leave notes | text | |

## 5. miRNA

### miRNA – Metadata File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identifier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 5 | R | assembly_version | Version of reference genome assembly (#) | integer | Appendix Table B10 |
| 6 | R | gene_build_version | Version of Ensembl gene build used for | integer | |

| | | | | | |
|---|---|---|---|---|---|
| | | | annotation | | |
| 7 | R | platform | Platform or technology used in detecting the expression | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | normalization_algorithm | Name of normalization algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by sequencing platforms | decimal | |
| 14 | O | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA |
| 15 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

**miRNA – Expression File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identifier for the analysis performed for a particular group of samples | | |
| 2 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 3 | R | mirna_seq | Sequence of the miRNA | text | |
| 4 | R | normalized_read_count | Normalized count of sequencing reads if analyzed by sequencing platforms | decimal | |
| 5 | R | raw_read_count | Raw count of sequencing reads if analyzed by sequencing platforms | integer | |
| 6 | O | normalized_expression_level | Normalized value of expression level if analyzed by microarray platforms | decimal | |
| 7 | O | fold_change | Expressed fold change if differential expression is measured | decimal | |
| 8 | O | reference_sample | ID of the reference sample if differential expression is measured | text | |
| 9 | O | quality_score | Quality score for the call | integer | |
| 10 | O | probability | Probability of the call | decimal | |
| 11 | O | is_annotated | Indicate if the fragment is annotated | integer | 1 = annotated<br>2 = not annotated |
| 12 | R | validation_status | Indicate if the fragment has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 13 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |

| | | | | | |
|---|---|---|---|---|---|
| 14 | O | note | Optional field to leave notes | text | |

**miRNA – Mapping Information File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | mirna_seq | Sequence of the miRNA | text | |
| 2 | R | chromosome | Name of the chromosome expressing the fragment (#) | integer | Appendix Table B6 |
| 3 | R | chromosome_start | Start position on the chromosome | integer | |
| 4 | R | chromosome_end | End position on the chromosome | integer | |
| 5 | O | chromosome_strand | Strand of the chromosome | integer | 1 = 1<br>-1 = -1 |
| 6 | O | xref_mirbase_id | Cross-reference to miRBase ID (e.g. has-let-7c) if available | text | |
| 7 | O | note | Optional field to leave notes | text | |

## 6. Exon Junction

**Exon Junction – Metadata File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 5 | R | assembly_version | Version of reference genome assembly (#) | integer | Appendix Table B10 |
| 6 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | |
| 7 | R | platform | Platform or technology used in detecting the expression | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |
| 11 | R | normalization_algorithm | Name of normalization algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | text/url | |
| 13 | O | seq_coverage | Sequence coverage if analyzed by | decimal | |

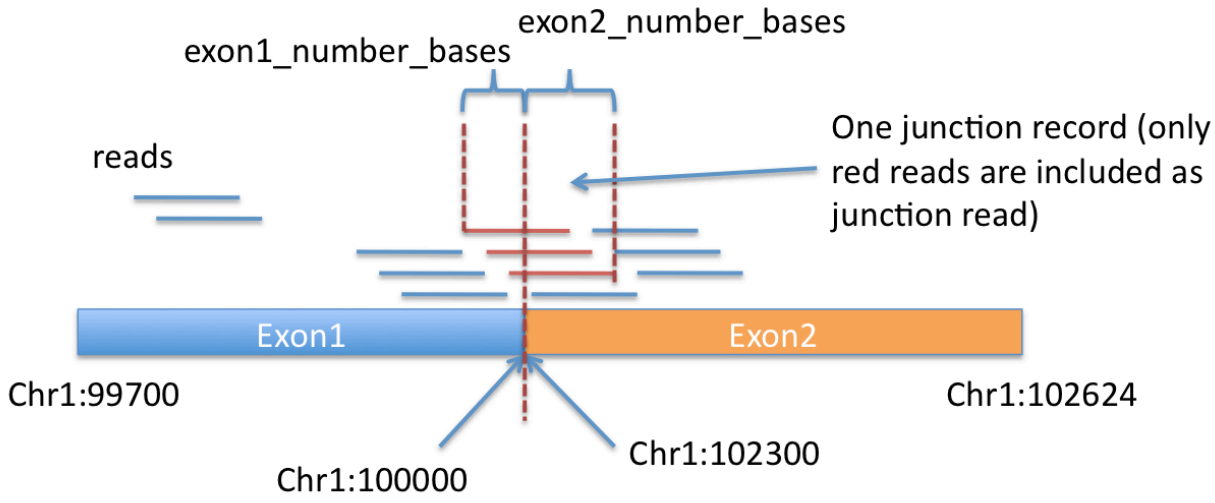
| | | | | | |
|---|---|---|---|---|---|
| | | | sequencing platforms | | |
| 14 | R | raw_data_repository | Public repository where raw data is submitted (#) | integer | 1 = EGA |
| 15 | R | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 16 | O | note | Optional field to leave notes | text | |

## Exon Junction – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | sample_id | Unique identifier for the sample being analyzed | | |
| 3 | R | junction_id | For known exons, use exonID1_exonID2 where exonID1 and exonID2 are Ensembl IDs of the 5' and 3' exons, respectively. For novel or putative exons, use assemblyBuild_exon1chr_exon1end_exon2chr_exon2start where assemblyBuild is hg18 or hg19; exon1chr and exone2chr are the chromosomes of the 5' and 3' exons, respectively; exon1end is the end position of the 5'exon; exon2start is the start position of the 3'exon. | text | |
| 4 | R | gene_stable_id | Stable ID of the gene containing the 5' exon at the junction. For annotated gene, use Ensembl gene ID. For putative and novel gene, use assemblyBuild_chr_start_end where assemblyBuild can be hg18 or hg19. | text | |
| 5 | R | gene_chromosome | Name of the chromosome containing the above gene. | integer | Appendix Table B6 |
| 6 | R | gene_strand | Strand of the chromosome | integer | 1 = 1<br>-1 = -1 |
| 7 | R | gene_start | Start position of the entire gene on the chromosome as annotated in Ensembl | integer | |
| 8 | R | gene_end | End position of the entire gene on the chromosome as annotated in Ensembl | integer | |
| 9 | O | second_gene_stable_id | In the case of a fusion gene, provide the Stable ID of the gene containing the 3' exon at the junction. For annotated genes, use Ensembl gene ID. For putative and novel genes, use assemblyBuild_chr_start_end where assemblyBuild can be hg18 or hg19. | text | |
| 10 | R | exon1_chromosome | Name of the chromosome containing | integer | Appendix Table |

| | | | | | |
|---|---|---|---|---|---|
| | | | the 5' exon (#) | | B6 |
| 11 | R | exon1_number_bases | Number of bases from 5' exon | integer | |
| 12 | R | exon1_end | End position of the 5' exon on the chromosome | integer | |
| 13 | O | exon1_strand | Chromsome strand of the 5' exon | integer | 1 = 1<br>-1 = -1 |
| 14 | R | exon2_chromosome | Name of the chromosome containing the 3' exon (#) | integer | Appendix Table B6 |
| 15 | R | exon2_number_bases | Number of bases from 3' exon | integer | |
| 16 | R | exon2_start | Start position of the 3' exon on the chromosome | integer | |
| 17 | O | exon2_strand | Chromsome strand of the 3' exon | integer | 1 = 1<br>-1 = -1 |
| 18 | O | is_fusion_gene | Indicate if the function is the result of a fusion gene | integer | 1 = yes<br>2 = no |
| 19 | O | is_novel_splice_form | Indicate if the splice form is novel | integer | 1 = yes<br>2 = no |
| 20 | O | junction_seq | Provide junction sequence if either is_fusion_gene or is_novel_splice_form is true | text | |
| 21 | O | junction_type | Type of junction | integer | 1 = canonical<br>2 = non-canonical<br>3 = U12 |
| 22 | R | junction_read_count | Count of sequencing reads that span across exons | decimal | |
| 23 | O | quality_score | Quality score for the junction call | integer | |
| 24 | O | probability | Probability of the junction call | decimal | |
| 25 | R | validation_status | Indicate if the junction has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 26 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 27 | O | note | Optional field to leave notes | text | |

**The following diagram illustrates how junction_id is assigned, how junction_read_count, exon1_number_bases and exon2_number_bases are calculated:**




- junction_id is: hg19_1_100000_1_102300
- junction read count is: 3

## 7. DNA Methylation

**Methylation (METH) – Metadata File**

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | donor_id | Unique identifier for the donor | | |
| 3 | R | diagnosis_id | Unique identifier for the diagnosis record for the donor | | |
| 4 | R | tumour_sample_id | Unique identifier for the tumour sample donated by the donor | | |
| 5 | R | matched_sample_id | Unique identifier for the control matched to the tumour sample | | |
| 6 | R | assembly_version | Version of reference genome assembly | integer | Appendix Table B10 |
| 7 | R | platform | Platform or technology used in detecting the methylation | integer | Appendix Table B5 |
| 8 | O | experimental_protocol | Name of experimental protocol and URL to written protocol | text/url | |
| 9 | R | base_calling_algorithm | Name of base calling algorithm and URL to written protocol | text/url | |
| 10 | R | alignment_algorithm | Name of alignment algorithm and URL to written protocol | text/url | |

| | | | | | |
|---|---|---|---|---|---|
| 11 | R | variation_calling_algorithm | Name of variation calling algorithm and URL to written protocol | text/url | |
| 12 | O | other_analysis_algorithm | Names of other analysis algorithms. Separate multiple algorithms by commas. | | |
| 13 | O | raw_data_repository | Public repository where raw data is submitted | integer | 1 = EGA<br>2 = dbSNP |
| 14 | O | raw_data_accession | Accession and URL for referencing the raw data at the public repository | text/url | |
| 15 | O | note | Optional field to leave notes | text | |

## Methylation (METH) – Primary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |
| 3 | R | methylated_fragment_id | Unique identifier for the methylated fragment | | |
| 4 | R | chromosome | Name of the chromosome containing the methylation | integer | Appendix Table B6 |
| 5 | R | chromosome_start | Start position of the methylation on the chromosome | integer | |
| 6 | R | chromosome_end | End position of the methylation on the chromosome | integer | |
| 7 | O | chromosome_strand | Chromosome strand | integer | 1 = 1<br>-1 = 1 |
| 8 | R | percent_methylation_1 | Percent methylation or beta value for probe 1 | decimal | |
| 9 | R | percent_methylation_2 | Percent methylation or beta value for probe 2 | decimal | |
| 10 | O | quality_score | Quality score for the methylation call | integer | |
| 11 | O | probability | Probability of the methylation call | decimal | |
| 12 | R | validation_status | Indicate if the methylation has been validated | integer | 1 = validated<br>2 = not tested<br>3 = not valid |
| 13 | O | validation_platform | Platform or technology used in validation | integer | Appendix Table B5 |
| 14 | O | note | Optional field to leave notes | text | |

## Methylation (METH) – Secondary Analysis File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | analysis_id | Unique identfier for the analysis performed for a particular group of samples | | |
| 2 | R | tumour_sample_id | Unique identifier for the tumour sample | | |

| | | | | | |
|---|---|---|---|---|---|
| 3 | R | methylated_fragment_id | Unique identifier for the methylation | | |
| 4 | R | gene_affected | Gene(s) containing the methylation. Use Ensembl gene id. Separate multiple genes with vertical bars in the form of geneA\|geneB\|geneC. If no gene is affected, use -888 (not applicable). | text | |
| 5 | R | gene_build_version | Version of Ensembl gene build used for annotation | integer | |
| 6 | O | note | Optional field to leave notes | text | |

## 8. Clinical and Sample Annotation

### Donor Data File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | donor_id | Unique identifier for a donor. It should be a de-identified code that does not link explicitly to the particular individual. | text | |
| 2 | O | biobank_id | Unique identifier for a biobank | text | |
| 3 | R | gender | Gender of the donor (others: Turner syndrome, hermaphrodites, etc) | integer | 1 = male<br>2 = female<br>3 = other |
| 4 | O | ethnicity | Ethnicity of the donor (others: khoisanid, australoid, etc) | text | 1 = negroid<br>2 = mongoloid<br>3 = caucasoid<br>4 = other |
| 5 | O | country_of_residence | Country of residence | text | au = Australia<br>ca = Canada<br>cn = China<br>fr = France<br>de = Germany<br>in = India<br>jp = Japan<br>es = Spain<br>uk = UK<br>us = USA |
| 6 | O | city_and_state_of_residence | City and state/province of residence | text | |
| 7 | O | vital_status | Indicate if the donor is alive or deceased. This element is updated every 6 months. | integer | 1 = alive<br>2 = deceased |
| 8 | O | age_at_recruitment | Age of the donor at the time of recruitment (years) | integer | |
| 9 | R | age_at_last_follow_up | Age of the donor at last follow up (years) | integer | |
| 10 | O | age_at_death | Age of the donor at death (years) | integer | |
| 11 | O | age_at_relapse | Age of the donor at relapse (years) | integer | |
| 12 | O | relapse_type | Type of relapse | integer | 1 = localized |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | 2 = distant |
| 13 | O | disease_outcome | Disease outcome | integer | 1 = progression<br>2 = treatment free survival |
| 14 | O | post_diagnosis_survival | Number of months the donor survived after diagnosis (not applicable if the donor is still alive). This element is updated every 6 months | integer | |
| 15 | O | quality_of_life_karnofsky | Quality of life based on Karnofsky Performance Scale Index (0-100) | integer | |
| 16 | O | quality_of_life_ecog | Quality of life based on Eastern Cooperative Oncology Group (ECOG) Performance Status | integer | |
| 17 | O | family_history_of_cancer | Indicate if family history is available | integer | 1 = Yes<br>2 = No |
| 18 | O | exposure_to_risk_factors | Indicate if donor has exposure to risk factors such as tobacco, alcohol and others | integer | 1 = Yes<br>2 = No |
| 19 | O | tobacco | Indicate if there is tobacco use | integer | 1 = Yes<br>2 = No<br>3 = in the past |
| 20 | O | alcohol | Indicate frequency of alcohol consumption | integer | 1 = regular<br>2 = occasional<br>3 = none |
| 21 | O | environmental_exposure | Indicate if environmental exposure was recorded for the donor | integer | 1 = Yes<br>2 = No |
| 22 | O | clinical_trial | Name of trial if donor is involved in clinical trials or cohort studies | text | |
| 23 | O | donor_record_release_date | Date of record released to DCC | date (YYYY MMDD) | |
| 24 | O | donor_record_created_date | Date of record created | date (YYYY MMDD) | |
| 25 | O | donor_record_last_update_date | Date of last update | date (YYYY MMDD) | |
| 26 | O | donor_record_notes | Optional field to leave notes | text | |

## Diagnosis Data File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | donor_id | Unique identifier for a donor. It should be a de-identified code that does not link explicitly to the particular individual. | text | |
| 2 | R | diagnosis_id | Unique identifier for a diagnosis record for the donor. It should be de-identified. | text | |
| 3 | O | consent | Indicate if consent was obtained | integer | 1 = Yes |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | 2 = No |
| 4 | R | icd_10 | Primary site of diagnosis (ICD-10 code) | text | Appendix Table B4 |
| 5 | O | icd_o3 | Morphology of cancer (ICD-O 3rd edition) | text | |
| 6 | O | therapy_type | Broad category of therapy received by the donor (*) | integer | 1 = biologic response modifier<br>2 = chemotherapy - multiple agent<br>3 = chemotherapy - single agent<br>4 = cryotherapy<br>5 = hormonal therapy<br>6 = immuno therapy<br>7 = radiation - external<br>8 = radiation - internal<br>9 = surgical biopsy<br>10 = surgical resection - cancer directed<br>11 = surgical resection - non cancer directed<br>12 = other |
| 7 | O | therapy_response | Response of donor to the therapy (^). This element is updated every 6 months. | integer | 1 = complete response<br>2 = partial response<br>3 = disease progression<br>4 = stable disease<br>5 = not evaluable |
| 8 | O | therapy_start_date | Start date of therapy | date (YYYY MMDD) | |
| 9 | O | therapy_end_date | End date of therapy | date (YYYY MMDD) | |
| 10 | O | date_of_examination | Date of examination | date (YYYY MMDD) | |
| 11 | R | date_of_diagnosis | Date of diagnosis | date (YYYY MMDD) | |
| 12 | R | age_at_diagnosis | Age of the donor at the time of diagnosis (years) | integer | |
| 13 | O | clinical_staging | Clinical staging using WHO system. | text | 1 = I<br>2 = IA |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | 3 = IB<br>4 = IC<br>5 = II<br>6 = IIA<br>7 = IIB<br>8 = IIC<br>9 = III<br>10 = IIIA<br>11 = IIIB<br>12 = IIIC<br>13 = IV<br>14 = IVA<br>15 = IVB<br>16 = IVC |
| 14 | R | clinical_t | tumour status based on clinical examination | text | 1 = T0<br>2 = T1<br>3 = T2<br>4 = T3<br>5 = T4<br>6 = TX<br>7 = Tis |
| 15 | R | clinical_n | Lymph node status based on clinical examination | text | 1 = N0<br>2 = N1<br>3 = N2<br>4 = N3<br>5 = N4<br>6 = NX |
| 16 | R | clinical_m | Distant metastasis status based on clinical examination | text | 1 = M0<br>2 = M1<br>3 = M2<br>4 = M3<br>5 = M4<br>6 = MX |
| 17 | O | tumour_staging_other | Alternative classification if TNM is not applicable (e.g. Binet/Rai for CLL, Ann Arbor for lymphomas, etc) | | |
| 18 | O | tumour_progress | Indicate if tumour progress occurs | integer | 1 = Yes<br>2 = No |
| 19 | O | concomitant_disease | Indicate if concomitant disease | integer | 1 = Yes<br>2 = No |
| 20 | O | diagnosis_record_release_date | Date of record released to DCC | date (YYYYMMDD) | |
| 21 | O | diagnosis_record_created_date | Date of record created | date (YYYYMMDD) | |
| 22 | O | diagnosis_record_last_update_date | Date of last update | date (YYYYMMDD) | |
| 23 | O | diagnosis_record_notes | Optional field to leave notes | text | |

## Sample Data File

| Order | O/R | Data element | Description | Data type | Values |
|---|---|---|---|---|---|
| 1 | R | donor_id | Unique identifier for a donor. It should be a de-identified code that does not link explicitly to the particular individual. | text | |
| 2 | R | diagnosis_id | Unique identifier for a diagnosis record for the donor. It should be de-identified. | text | |
| 3 | R | sample_id | Unique identifier for the sample as assigned by data provider | text | |
| 4 | O | sample_id_provided_by_central_repo | Unique identifier for the sample as provided by central repository such as biobank | text | |
| 5 | O | sample_name | Name of the sample | text | |
| 6 | O | sample_ega_accession | Sample EGA accession | text | |
| 7 | O | primary_secondary | Indicate if the tumour is primary or secondary | integer | 1 = primary<br>2 = secondary |
| 8 | R | recurrent | Indicate if the tumour recurrent | integer | 1 = Yes<br>2 = No |
| 9 | R | sample_type | Type of sample (#) | integer | 1 = tumour tissue<br>2 = tumour xenograft<br>3 = matched control<br>4 = site-matched control<br>5 = blood<br>6 = buffy coat<br>7 = plasma<br>8 = serum<br>9 = saliva<br>10 = urine<br>11 = cell line<br>12 = cell line - tumour<br>13 = cell line - matched control |
| 10 | R | sample_collection_date | Date of sample collection or storage | date (YYYY MMDD) | |
| 11 | O | sample_collection_procedure | Procedure for collecting the sample | text | |
| 12 | R | sample_freezing_method | Method for freezing the sample | integer | 1 = liquid nitrogen<br>2 = dry ice<br>3 = cyro-preservation<br>4 = others |
| 13 | R | tissue_fixation_protocol | Protocol for fixing the tissue | integer | 1 = formalin<br>2 = formalin |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | buffered<br>3 = embedding |
| 14 | R | time_between_tissue_removal_and_fixation_or_freezing | Time between tissue removal and fixation or cryo-preservation in hours and minutes (hhmm) | integer | |
| 15 | O | time_between_vascular_clamping_and_tissue_removal | Time between vascular clamping and tissue removal in hours and (hhmm) | integer | |
| 16 | O | duration_of_transport | Duration of transport in days, hours and minutes (ddhhmm) | integer | |
| 17 | O | temperature_during_transport | Temperature during transport (Celsius) | integer | |
| 18 | R | storage_method | Type of storage methods used for the sample | integer | 1 = culture<br>2 = frozen<br>3 = liquid frozen<br>4 = parafin block<br>5 = RNA later frozen<br>6 = slide<br>7 = tissue array |
| 19 | O | initial_temperature_at_storage | Initial temperature at storage (Celsius) | integer | |
| 20 | O | temperature_during_storage | Temperature during storage (Celsius) | integer | |
| 21 | O | history_of_freezing_thawing | History of freezing/thawing | text | |
| 22 | R | quantity_on_hand | Amount of sample available (e.g. 3 aliquots, 5 mg, 2 tissue pieces) | text | |
| 23 | R | grading_system_used | Name of grading system used | text | |
| 24 | R | tumour_grading | Pathologist assigned grade | text | |
| 25 | R | digital_image_of_stained_section | Linkout to digital image of stained section | URL | |
| 26 | R | percent_intact_tumour_cells | Percentage of intact ("viable") tumour cells within sample | integer | |
| 27 | O | percent_necrotic_tissue | Percentage of necrotic tissue | integer | |
| 28 | O | percent_inflammatory_tissue | Percentage of inflammatory tissue | integer | |
| 29 | O | percent_debris | Percentage of debris | integer | |
| 30 | O | molecular_genetics_diagnostics | Flow cytometry charts as alternative | text | |
| 31 | O | name_of_pathologist | Initial and reference pathologist(s) | text | |
| 32 | O | sample_record_release_date | Date of record released to DCC | date (YYYYMMDD) | |
| 33 | R | sample_record_created_date | Date of record created | date (YYYYMMDD) | |
| 34 | R | sample_record_last_update_date | Date of last update | date (YYYYMMDD) | |
| 35 | O | sample_record_notes | Optional field to leave notes | text | |

# Appendix B: Value Codes for DEs with Controlled Vocabulary

***Value codes or controlled vocabulary will be added as the projects evolve. Please contact DCC to provide suggestions.***

**Appendix Table B1. Lead Jurisdiction ID**

B1

| Lead Jurisdiction | ID |
|---|---|
| Australia | au |
| Canada | ca |
| China | cn |
| France | fr |
| Germany | de |
| India | in |
| Japan | jp |
| Spain | es |
| UK | uk |
| USA | us |

**Appendix Table B2. ID for Types of Primary Tumours**

B2

| Primary Tumour Type | ID |
|---|---|
| Pancreatic cancer | 01 |
| Breast cancer | 02 |
| Brain cancer | 03 |
| Colorectal cancer | 04 |
| Ovarian cancer | 05 |
| Gastric cancer | 06 |
| Liver cancer | 07 |
| Pediatric brain tumours | 08 |
| Oral cancer | 09 |
| Chronic lymphocytic leukemia | 10 |
| Lung cancer | 11 |
| Melanoma | 12 |
| Kidney renal clear cell cancinoma | 13 |
| Kidney renal papillary cell carcinoma | 14 |
| Acute Myeloid Leukemia | 15 |
| Head and Neck squamous cell carcinoma | 16 |
| Lung adenocaracinoma | 17 |
| Lung squamous cell carcinoma | 18 |
| Rectum adenocarcinoma | 19 |
| Stomach adenocarcinoma | 20 |
| Uterine Corpus Endometrioid Carcinoma | 21 |

| | |
|---|---|
| Colon adenocarcinoma | 22 |

**Appendix Table B3. Institute ID**

B3

| Institution | ID |
|---|---|
| Advanced Centre for Treatment, Research and Education in Cancer (Mumbai) | 001 |
| AMC Medical Research BV (Netherlands) | 002 |
| Applied Biosystems Inc. | 003 |
| Australian Pancreatic Cancer Network | 004 |
| Barcelona Supercomputer Center (BSC-Barcelona) | 005 |
| Baylor College of Medicine (Houston, TX) | 006 |
| BCCA (Canada) | 007 |
| Beijing Cancer Hospital/Insititute | 008 |
| Beijing Genome Institute/Shenzhen | 009 |
| Bioquant (Heidelberg) | 010 |
| British Columbia Cancer Agency (Vancouver, Canada) | 011 |
| Broad Institute (Cambridge, MA) | 012 |
| Catalan Institute of Oncology | 013 |
| Center for Cancer Research (CICSalamanca) and University Hospital | 014 |
| Center for Genomic Regulation (CRG) and Pompeu Fabra University (UPF) | 015 |
| Centre Leon Berard (Lyon, France) | 016 |
| Centre National de Génotypage (France) | 017 |
| Centre Val d'Aurelle (Montpellier, France) | 019 |
| Commissariat à l'Energie Atomique | 020 |
| CRUK (UK) | 021 |
| Dana-Farber Cancer Institute | 022 |
| DFCI (USA) | 023 |
| EMBL-EBI (Hinxton) | 024 |
| Erasmus (Netherlands) | 025 |
| European Molecular Biology Laboratory (EMBL), Heidelberg | 026 |
| Fondation Jean Dausset CEPH | 027 |
| Fondation Synergie-Lyon-Cancer | 028 |
| Garvan Institute of Medical Research | 029 |
| German Cancer Research Center (DKFZ), Heidelberg | 030 |
| Harvard Medical School and Brigham and Women's Hospital (Cambridge, MA) | 031 |
| Hiroshima University, Faculty of Medicine | 032 |
| Hospital Clinic, University of Barcelona | 033 |
| Hospital-University : AP-HP Paris (Beaujon, H. Mondor, A. Béclère and P. Brousse hospitals), Bordeaux, Rennes, Toulouse, Grenoble | 034 |
| HudsonAlpha Institute for Biotechnology (Huntsville, AL) | 035 |
| Human Genome Center, Institute of Medical Science, University of Tokyo | 036 |
| ICR (UK) | 037 |
| INCa (France) | 038 |
| Institut Curie (France) | 039 |
| Institut Génomique | 041 |

| | |
|---|---|
| Institut National de la Santé et de la Recherche Médicale | 042 |
| Institut National du Cancer (Boulogne-Billancourt, France) | 044 |
| Institut Paoli-Calmettes (Marseille, France) | 045 |
| Institute for Molecular Bioscience (Brisbane) | 046 |
| Institute for System Biology (Seattle, WA) | 047 |
| International Breast Cancer Genome Consortium (UK) | 048 |
| International Genome Consortium (Phoenix, AZ) | 049 |
| Johns Hopkins University (Baltimore, MD) | 050 |
| Lawrence Berkeley National Laboratory (Berkeley, CA) | 051 |
| Lund University (Sweden) | 052 |
| Massachusetts General Hospital | 053 |
| Max-Planck-Institut for Molecular Genetics (Berlin) | 054 |
| Mayo Clinic | 055 |
| Memorial Sloan-Kettering Cancer Center (New York, NY) | 056 |
| Mount Sinai Hospital (Toronto) | 057 |
| National Bioinformatics Institute | 058 |
| National Cancer Center | 059 |
| National Center for tumour Diseases (Heidelberg) | 060 |
| National DNA and tumour Bank Networks | 061 |
| National Institute of Biomedical Genomics (Kalyani) | 062 |
| National Institutes of Health; National Cancer Institute, National Human Genome Research Institute | 063 |
| National Sequencing Center (Barcelona) | 064 |
| NCI Bari (Italy) | 065 |
| Norwegian Radium Hospital (Norway) | 066 |
| Ontario Institute for Cancer Research | 067 |
| Osaka Medical Center for Cancer & Cardiovascular Diseases | 068 |
| Peking University School of Oncology | 069 |
| Peter MacCallum Cancer Centre | 070 |
| Queensland Centre for Medical Genomics | 018 |
| Queensland Institute of Medical Research | 071 |
| Radboud University (Netherlands) | 072 |
| Research Center for Advanced Science and Technology, University of Tokyo | 073 |
| RIKEN | 074 |
| Silicon Graphics Inc. | 075 |
| Singapore General Hospital (Hong Kong) | 076 |
| Spanish Cancer Research Network | 077 |
| Spanish National Cancer Research Centre (CNIO-Madrid) | 078 |
| UCSF | 079 |
| University Health Network (Toronto) | 080 |
| University of California (Santa Cruz, CA) | 081 |
| University of Cambridge (UK) | 082 |
| University of Deusto | 083 |
| University of Düsseldorf | 084 |
| University of Heidelberg | 085 |
| University of North Carolina (Chapel Hill, NC) | 086 |

| | |
|---|---|
| University of Oviedo | 087 |
| University of Queensland (Australia) | 088 |
| University of Southern California (Los Angeles, CA) | 089 |
| University of Tromsø (Norway) | 090 |
| University of Verona | 091 |
| Wakayama Medical University | 092 |
| Wellcome Trust Sanger Institute | 093 |
| Westmead Institute for Cancer Research | 094 |
| Washington University Genome Sequencing Center (St. Louis, MO) | 095 |
| The Cancer Genome Atlas | 096 |

*Please contact DCC if your institute is not listed or wish to modify the identifier*

**Appendix Table B4. ICD10 Codes for Disease Sites**

**B4**

| Disease Site | ICD10 Code |
|---|---|
| Pancreas | C25 |
| Breast | C50 |
| Brain | C71 |
| Colon | C18 |
| Rectum | C20 |
| Ovary | C56 |
| Liver | C22 |
| Lung | C30-C39 |
| Skin | C43-C44 |
| Kidney | C64 |
| Stomach | C16 |
| Uterus | C54 |
| Myeloid leukaemia | C92 |
| Prostate | C61 |
| Bladder | C67 |

**Appendix Table B5: Value Codes for Platform or Validation Platform**

**B5**

| Platform or Validation Platform | Values |
|---|---|
| PCR | 1 |
| qPCR | 2 |
| capillary sequencing | 3 |
| SOLiD sequencing | 4 |
| GA sequencing | 5 |
| 454 sequencing | 6 |
| Helicos sequencing | 7 |
| Affymetrix Genome-Wide Human SNP Array 6.0 | 8 |
| Affymetrix Genome-Wide Human SNP Array 5.0 | 9 |
| Affymetrix Mapping 100K Array Set | 10 |
| Affymetrix Mapping 500K Array Set | 11 |
| Affymetrix Mapping 10K 2.0 Array Set | 12 |

| | |
|---|---|
| Affymetrix EMET Plus Premier Pack | 13 |
| Agilent Whole Human Genome Oligo Microarray Kit | 14 |
| Agilent Human Genome 244A | 15 |
| Agilent Human Genome 105A | 16 |
| Agilent Human CNV Association 2x105K | 17 |
| Agilent Human Genome 44K | 18 |
| Agilent Human CGH 1x1M | 19 |
| Agilent Human CGH 2x400K | 20 |
| Agilent Human CGH 4x180K | 21 |
| Agilent Human CGH 8x60K | 22 |
| Agilent Human CNV 2x400K | 23 |
| Agilent Human miRNA Microarray Kit (v2) | 24 |
| Agilent Human CpG Island Microarray Kit | 25 |
| Agilent Human Promoter ChIP-on-chip Microarray Set | 26 |
| Agilent Human SpliceArray | 27 |
| Illumina human1m-duo | 28 |
| Illumina human660w-quad | 29 |
| Illumina humancytosnp-12 | 30 |
| Illumina human510s-duo | 31 |
| Illumina humanmethylation27 | 32 |
| Illumina goldengate methylation | 33 |
| Illumina HumanHT-12 v4.0 beadchip | 34 |
| Illumina HumanWG-6 v3.0 beadchip | 35 |
| Illumina HumanRef-8 v3.0 beadchip | 36 |
| Illumina microRNA Expression Profiling Panel | 37 |
| Illumina humanht-16 | 38 |
| Illumina humanht-17 | 39 |
| Nimblegen Human CGH 3x720 Whole-Genome v3.0 Array | 40 |
| Nimblegen Human CGH 2.1M Whole-Genome v2.0D Array | 41 |
| Nimblegen Gene Expression 385K | 42 |
| Nimblegen Gene Expression 4x72K | 43 |
| Nimblegen Gene Expression 12x135K | 44 |
| Nimblegen Human Methylation 2.1M Whole-Genome sets | 45 |
| Nimblegen Human Methylation 385K Whole-Genome sets | 46 |
| Nimblegen CGS | 47 |
| Illumina Human1M OmniQuad chip | 48 |
| PCR and capillary sequencing | 49 |
| Custom-designed gene expression array | 50 |
| Affymetrix HT Human Genome U133A Array Plate Set | 51 |
| Agilent 244K Custom Gene Expression G4502A-07-1 | 52 |
| Agilent 244K Custom Gene Expression G4502A-07-2 | 53 |
| Agilent 244K Custom Gene Expression G4502A-07-3 | 54 |
| Agilent Human Genome CGH Custom Microaary 2x415K | 55 |
| Affymetrix Human U133 Plus PM | 56 |
| Affymetrix Human U133 Plus 2.0 | 57 |

| | |
|---|---|
| Affymetrix Human Exon 1.0 ST | 58 |
| Almac Human CRC | 59 |
| Illumina HiSeq | 60 |
| Affymetrix Human MIP 330K | 61 |
| Affymetrix Human Gene 1.0 ST | 62 |
| Illumina Human Omni1-Quad beadchip | 63 |
| Sequenom MassARRAY | 64 |
| Custom-designed cDNA array | 65 |

*Please contact DCC if your platform/technology is not listed here*

**Appendix Table B6. Chromosome Names for Reference Genomes NCB136 and GRCh37**

**B6**

| Chromosome Name | Values | Reference Genome | Gene Annotation |
|---|---|---|---|
| 1 | 1 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 2 | 2 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 3 | 3 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 4 | 4 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 5 | 5 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 6 | 6 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 7 | 7 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 8 | 8 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 9 | 9 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 10 | 10 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 11 | 11 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 12 | 12 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 13 | 13 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 14 | 14 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 15 | 15 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 16 | 16 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 17 | 17 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 18 | 18 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 19 | 19 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 20 | 20 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 21 | 21 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| 22 | 22 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| X | 23 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| Y | 24 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| MT | 25 | NCBI36 & GRCh37 | Ensembl 53 & 55 |
| c5_H2 | 26 | NCBI36 | Ensembl 53 |
| c6_COX | 27 | NCBI36 | Ensembl 53 |
| c6_QBL | 28 | NCBI36 | Ensembl 53 |
| NT_113870 | 29 | NCBI36 | Ensembl 53 |
| NT_113871 | 30 | NCBI36 | Ensembl 53 |
| NT_113872 | 31 | NCBI36 | Ensembl 53 |
| NT_113874 | 32 | NCBI36 | Ensembl 53 |
| NT_113878 | 33 | NCBI36 | Ensembl 53 |
| NT_113880 | 34 | NCBI36 | Ensembl 53 |
| NT_113881 | 35 | NCBI36 | Ensembl 53 |
| NT_113884 | 36 | NCBI36 | Ensembl 53 |
| NT_113885 | 37 | NCBI36 | Ensembl 53 |
| NT_113886 | 38 | NCBI36 | Ensembl 53 |

| | | | |
|---|---|---|---|
| NT_113888 | 39 | NCBI36 | Ensembl 53 |
| NT_113889 | 40 | NCBI36 | Ensembl 53 |
| NT_113890 | 41 | NCBI36 | Ensembl 53 |
| NT_113898 | 42 | NCBI36 | Ensembl 53 |
| NT_113899 | 43 | NCBI36 | Ensembl 53 |
| NT_113901 | 44 | NCBI36 | Ensembl 53 |
| NT_113902 | 45 | NCBI36 | Ensembl 53 |
| NT_113903 | 46 | NCBI36 | Ensembl 53 |
| NT_113906 | 47 | NCBI36 | Ensembl 53 |
| NT_113908 | 48 | NCBI36 | Ensembl 53 |
| NT_113909 | 49 | NCBI36 | Ensembl 53 |
| NT_113910 | 50 | NCBI36 | Ensembl 53 |
| NT_113911 | 51 | NCBI36 | Ensembl 53 |
| NT_113912 | 52 | NCBI36 | Ensembl 53 |
| NT_113915 | 53 | NCBI36 | Ensembl 53 |
| NT_113916 | 54 | NCBI36 | Ensembl 53 |
| NT_113917 | 55 | NCBI36 | Ensembl 53 |
| NT_113923 | 56 | NCBI36 | Ensembl 53 |
| NT_113924 | 57 | NCBI36 | Ensembl 53 |
| NT_113925 | 58 | NCBI36 | Ensembl 53 |
| NT_113926 | 59 | NCBI36 | Ensembl 53 |
| NT_113927 | 60 | NCBI36 | Ensembl 53 |
| NT_113929 | 61 | NCBI36 | Ensembl 53 |
| NT_113930 | 62 | NCBI36 | Ensembl 53 |
| NT_113931 | 63 | NCBI36 | Ensembl 53 |
| NT_113932 | 64 | NCBI36 | Ensembl 53 |
| NT_113933 | 65 | NCBI36 | Ensembl 53 |
| NT_113934 | 66 | NCBI36 | Ensembl 53 |
| NT_113935 | 67 | NCBI36 | Ensembl 53 |
| NT_113936 | 68 | NCBI36 | Ensembl 53 |
| NT_113937 | 69 | NCBI36 | Ensembl 53 |
| NT_113939 | 70 | NCBI36 | Ensembl 53 |
| NT_113943 | 71 | NCBI36 | Ensembl 53 |
| NT_113944 | 72 | NCBI36 | Ensembl 53 |
| NT_113946 | 73 | NCBI36 | Ensembl 53 |
| NT_113949 | 74 | NCBI36 | Ensembl 53 |
| NT_113951 | 75 | NCBI36 | Ensembl 53 |
| NT_113953 | 76 | NCBI36 | Ensembl 53 |
| NT_113954 | 77 | NCBI36 | Ensembl 53 |
| NT_113956 | 78 | NCBI36 | Ensembl 53 |
| NT_113957 | 79 | NCBI36 | Ensembl 53 |
| NT_113958 | 80 | NCBI36 | Ensembl 53 |
| NT_113960 | 81 | NCBI36 | Ensembl 53 |
| NT_113961 | 82 | NCBI36 | Ensembl 53 |
| NT_113962 | 83 | NCBI36 | Ensembl 53 |
| NT_113963 | 84 | NCBI36 | Ensembl 53 |
| NT_113964 | 85 | NCBI36 | Ensembl 53 |
| NT_113965 | 86 | NCBI36 | Ensembl 53 |
| NT_113966 | 87 | NCBI36 | Ensembl 53 |
| HSCHR17_1 | 88 | GRCh37 | Ensembl 55 |
| HSCHR17_RANDOM_CTG2 | 89 | GRCh37 | Ensembl 55 |
| HSCHR17_RANDOM_CTG3 | 90 | GRCh37 | Ensembl 55 |
| HSCHR19_RANDOM_CTG2 | 91 | GRCh37 | Ensembl 55 |

| | | | |
|---|---|---|---|
| HSCHR1_RANDOM_CTG12 | 92 | GRCh37 | Ensembl 55 |
| HSCHR1_RANDOM_CTG5 | 93 | GRCh37 | Ensembl 55 |
| HSCHR4_RANDOM_CTG2 | 94 | GRCh37 | Ensembl 55 |
| HSCHR4_RANDOM_CTG3 | 95 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_APD | 96 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_COX | 97 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_DBB | 98 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_MANN | 99 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_MCF | 100 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_QBL | 101 | GRCh37 | Ensembl 55 |
| HSCHR6_MHC_SSTO | 102 | GRCh37 | Ensembl 55 |
| HSCHR7_RANDOM_CTG1 | 103 | GRCh37 | Ensembl 55 |
| HSCHR8_RANDOM_CTG1 | 104 | GRCh37 | Ensembl 55 |
| HSCHR8_RANDOM_CTG4 | 105 | GRCh37 | Ensembl 55 |
| HSCHR9_RANDOM_CTG2 | 106 | GRCh37 | Ensembl 55 |
| HSCHR9_RANDOM_CTG4 | 107 | GRCh37 | Ensembl 55 |
| HSCHR9_RANDOM_CTG5 | 108 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG1 | 109 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG10 | 110 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG11 | 111 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG13 | 112 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG14 | 113 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG15 | 114 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG16 | 115 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG17 | 116 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG2 | 117 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG20 | 118 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG21 | 119 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG22 | 120 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG23 | 121 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG26 | 122 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG29 | 123 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG3 | 124 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG30 | 125 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG31 | 126 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG32 | 127 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG33 | 128 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG34 | 129 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG35 | 130 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG36 | 131 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG4 | 132 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG40 | 133 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG5 | 134 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG6 | 135 | GRCh37 | Ensembl 55 |
| HSCHRUN_RANDOM_CTG9 | 136 | GRCh37 | Ensembl 55 |

**Appendix Table B7. Values for Consequences from SSM/SGV**
Controlled vocabulary adopted from Ensembl Release 55

**B7**

| Consequence | Value |
|---|---|
| 3prime_utr | 1 |
| 5prime_utr | 2 |

| | |
|---|---|
| upstream | 3 |
| downstream | 4 |
| essential_splice_site,3prime_utr | 5 |
| essential_splice_site,5prime_utr | 6 |
| essential_splice_site,intronic | 7 |
| essential_splice_site,non_synonymous_coding | 8 |
| essential_splice_site,stop_lost | 9 |
| essential_splice_site,synonymous_coding | 10 |
| frameshift_coding | 11 |
| frameshift_coding,splice_site | 12 |
| intergenic | 13 |
| intronic | 14 |
| non_synonymous_coding | 15 |
| non_synonymous_coding,splice_site | 16 |
| splice_site,3prime_utr | 17 |
| splice_site,5prime_utr | 18 |
| splice_site,intronic | 19 |
| splice_site,synonymous_coding | 20 |
| stop_gained | 21 |
| stop_gained,splice_site | 22 |
| stop_lost | 23 |
| stop_lost,splice_site | 24 |
| synonymous_coding | 25 |
| utr | 26 |
| splice_site | 27 |
| noncoding_rna | 28 |
| complex_indel | 29 |
| regulatory_region | 30 |
| inframe_indel | 31 |
| start_lost | 32 |
| ambiguous | 33 |
| complex_substitution | 34 |

**Appendix Table B8. Description of Consequences from SSM/SGV**
Description adopted from Ensembl Release 55

**B8**

| Consequence | Description |
|---|---|
| 3' UTR | In 3' UTR |
| 5' UTR | In 5' UTR |
| Upstream | Within 5 kb upstream of the 5'-end of a transcript |
| Splice site | 1-3 bps into an exon or 3-8 bps into an intron |
| Downstream | Within 5 kb downstream of the 3'-end of a transcript |
| Essential splice site | In the first 2 or the last 2 basepairs of an intron |
| Frameshift | In coding sequence, resulting in a frameshift |
| Intronic | In intron |
| Non-synonymous | In coding sequence, resulting in an aa change |

| | |
|---|---|
| Synonymous | In coding sequence, not resulting in an aa change |
| Start lost | In coding sequence, resulting in the loss of a start codon |
| Stop lost | In coding sequence, resulting in the loss of a stop codon |
| Stop gained | In coding sequence, resulting in the gain of a stop codon |
| Regulatory region | In regulatory region annotated by Ensembl |
| Intergenic | More than 5 kb away from a transcript |
| Ambiguous | In coding sequence, resulting in unpredictable effect on amino acid due to ambiguous nucleotide change |
| Complex InDel | Insertion or deletion that spans an exon/intron border or a coding sequence/UTR border. |
| Complex substitution | Substitution that is 2bps or longer |

**Appendix Table B9. Values for Types of StSM/StGV**
Controlled vocabulary adpted from ICGC DCM WG

**B9**

| Type of StSM/StGV | Subtype | Value |
|---|---|---|
| intrachromosomal rearrangement | deletion | 1 |
| | tandem duplication | 2 |
| | inversion | 3 |
| | inverted duplication - head-to-head | 4 |
| | inverted duplication - tail-to-tail | 5 |
| | insertion | 6 |
| | intrachromosomal rearrangement with inverted orientation | 7 |
| | intrachromosomal rearrangement with non-inverted orientation | 8 |
| | fold-back inversion | 9 |
| | complex intrachromosomal rearrangement | 10 |
| interchromosomal rearrangement | reciprocal translocation | 11 |
| | unbalanced translocation | 12 |
| | interchromosomal insertion | 13 |
| | interchromosomal rearrangement - unknown type | 14 |
| | complex interchromosomal rearrangement | 15 |
| rearrangements involving amplicons | intrachromosomal amplicon-to-amplicon | 16 |
| | intrachromosomal amplicon-to-nonamplified dna | 17 |
| | interchromosomal amplicon-to-amplicon | 18 |
| | interchromosomal amplicon-to-nonamplified dna | 19 |
| | extrachromosomal | 20 |

**Appendix Table B10. Value Codes for Reference Genome Assembly Version**

**B10**

| Reference Genome Assembly Version | Values |
|---|---|
| GRCh37 | 1 |
| NCBI36 | 2 |
| GRCh37.p1 | 3 |
| GRCh37.p2 | 4 |

**Appendix Table B11. ISO 3166-1-alpha-2 Country Codes (for Clinical Data Encoding)**
The following table contains ISO standard two-letter country codes for use in encoding country of residence of donors, etc. when submitting clinical data.
(Reproduced here from http://www.iso.org/iso/english_country_names_and_code_elements.)

B11

| Country names | ISO 3166-1-alpha-2 code |
|---|---|
| AFGHANISTAN | AF |
| ALAND ISLANDS | AX |
| ALBANIA | AL |
| ALGERIA | DZ |
| AMERICAN SAMOA | AS |
| ANDORRA | AD |
| ANGOLA | AO |
| ANGUILLA | AI |
| ANTARCTICA | AQ |
| ANTIGUA AND BARBUDA | AG |
| ARGENTINA | AR |
| ARMENIA | AM |
| ARUBA | AW |
| AUSTRALIA | AU |
| AUSTRIA | AT |
| AZERBAIJAN | AZ |
| BAHAMAS | BS |
| BAHRAIN | BH |
| BANGLADESH | BD |
| BARBADOS | BB |
| BELARUS | BY |
| BELGIUM | BE |
| BELIZE | BZ |
| BENIN | BJ |
| BERMUDA | BM |
| BHUTAN | BT |
| BOLIVIA, PLURINATIONAL STATE OF | BO |
| BONAIRE, SINT EUSTATIUS AND SABA | BQ |
| BOSNIA AND HERZEGOVINA | BA |
| BOTSWANA | BW |
| BOUVET ISLAND | BV |
| BRAZIL | BR |
| BRITISH INDIAN OCEAN TERRITORY | IO |

| | |
|---|---|
| BRUNEI DARUSSALAM | BN |
| BULGARIA | BG |
| BURKINA FASO | BF |
| BURUNDI | BI |
| CAMBODIA | KH |
| CAMEROON | CM |
| CANADA | CA |
| CAPE VERDE | CV |
| CAYMAN ISLANDS | KY |
| CENTRAL AFRICAN REPUBLIC | CF |
| CHAD | TD |
| CHILE | CL |
| CHINA | CN |
| CHRISTMAS ISLAND | CX |
| COCOS (KEELING) ISLANDS | CC |
| COLOMBIA | CO |
| COMOROS | KM |
| CONGO | CG |
| CONGO, THE DEMOCRATIC REPUBLIC OF THE | CD |
| COOK ISLANDS | CK |
| COSTA RICA | CR |
| COTE D'IVOIRE | CI |
| CROATIA | HR |
| CUBA | CU |
| CURACAO | CW |
| CYPRUS | CY |
| CZECH REPUBLIC | CZ |
| DENMARK | DK |
| DJIBOUTI | DJ |
| DOMINICA | DM |
| DOMINICAN REPUBLIC | DO |
| ECUADOR | EC |
| EGYPT | EG |
| EL SALVADOR | SV |
| EQUATORIAL GUINEA | GQ |
| ERITREA | ER |
| ESTONIA | EE |
| ETHIOPIA | ET |
| FALKLAND ISLANDS (MALVINAS) | FK |
| FAROE ISLANDS | FO |
| FIJI | FJ |
| FINLAND | FI |

| | |
|---|---|
| FRANCE | FR |
| FRENCH GUIANA | GF |
| FRENCH POLYNESIA | PF |
| FRENCH SOUTHERN TERRITORIES | TF |
| GABON | GA |
| GAMBIA | GM |
| GEORGIA | GE |
| GERMANY | DE |
| GHANA | GH |
| GIBRALTAR | GI |
| GREECE | GR |
| GREENLAND | GL |
| GRENADA | GD |
| GUADELOUPE | GP |
| GUAM | GU |
| GUATEMALA | GT |
| GUERNSEY | GG |
| GUINEA | GN |
| GUINEA-BISSAU | GW |
| GUYANA | GY |
| HAITI | HT |
| HEARD ISLAND AND MCDONALD ISLANDS | HM |
| HOLY SEE (VATICAN CITY STATE) | VA |
| HONDURAS | HN |
| HONG KONG | HK |
| HUNGARY | HU |
| ICELAND | IS |
| INDIA | IN |
| INDONESIA | ID |
| IRAN, ISLAMIC REPUBLIC OF | IR |
| IRAQ | IQ |
| IRELAND | IE |
| ISLE OF MAN | IM |
| ISRAEL | IL |
| ITALY | IT |
| JAMAICA | JM |
| JAPAN | JP |
| JERSEY | JE |
| JORDAN | JO |
| KAZAKHSTAN | KZ |
| KENYA | KE |
| KIRIBATI | KI |

| | |
|---|---|
| KOREA, DEMOCRATIC PEOPLE'S REPUBLIC OF | KP |
| KOREA, REPUBLIC OF | KR |
| KUWAIT | KW |
| KYRGYZSTAN | KG |
| LAO PEOPLE'S DEMOCRATIC REPUBLIC | LA |
| LATVIA | LV |
| LEBANON | LB |
| LESOTHO | LS |
| LIBERIA | LR |
| LIBYAN ARAB JAMAHIRIYA | LY |
| LIECHTENSTEIN | LI |
| LITHUANIA | LT |
| LUXEMBOURG | LU |
| MACAO | MO |
| MACEDONIA, THE FORMER YUGOSLAV REPUBLIC OF | MK |
| MADAGASCAR | MG |
| MALAWI | MW |
| MALAYSIA | MY |
| MALDIVES | MV |
| MALI | ML |
| MALTA | MT |
| MARSHALL ISLANDS | MH |
| MARTINIQUE | MQ |
| MAURITANIA | MR |
| MAURITIUS | MU |
| MAYOTTE | YT |
| MEXICO | MX |
| MICRONESIA, FEDERATED STATES OF | FM |
| MOLDOVA, REPUBLIC OF | MD |
| MONACO | MC |
| MONGOLIA | MN |
| MONTENEGRO | ME |
| MONTSERRAT | MS |
| MOROCCO | MA |
| MOZAMBIQUE | MZ |
| MYANMAR | MM |
| NAMIBIA | NA |
| NAURU | NR |
| NEPAL | NP |
| NETHERLANDS | NL |
| NEW CALEDONIA | NC |
| NEW ZEALAND | NZ |

| | |
|---|---|
| NICARAGUA | NI |
| NIGER | NE |
| NIGERIA | NG |
| NIUE | NU |
| NORFOLK ISLAND | NF |
| NORTHERN MARIANA ISLANDS | MP |
| NORWAY | NO |
| OMAN | OM |
| PAKISTAN | PK |
| PALAU | PW |
| PALESTINIAN TERRITORY, OCCUPIED | PS |
| PANAMA | PA |
| PAPUA NEW GUINEA | PG |
| PARAGUAY | PY |
| PERU | PE |
| PHILIPPINES | PH |
| PITCAIRN | PN |
| POLAND | PL |
| PORTUGAL | PT |
| PUERTO RICO | PR |
| QATAR | QA |
| REUNION | RE |
| ROMANIA | RO |
| RUSSIAN FEDERATION | RU |
| RWANDA | RW |
| SAINT BARTHELEMY | BL |
| SAINT HELENA, ASCENSION AND TRISTAN DA CUNHA | SH |
| SAINT KITTS AND NEVIS | KN |
| SAINT LUCIA | LC |
| SAINT MARTIN (FRENCH PART) | MF |
| SAINT PIERRE AND MIQUELON | PM |
| SAINT VINCENT AND THE GRENADINES | VC |
| SAMOA | WS |
| SAN MARINO | SM |
| SAO TOME AND PRINCIPE | ST |
| SAUDI ARABIA | SA |
| SENEGAL | SN |
| SERBIA | RS |
| SEYCHELLES | SC |
| SIERRA LEONE | SL |
| SINGAPORE | SG |
| SINT MAARTEN (DUTCH PART) | SX |

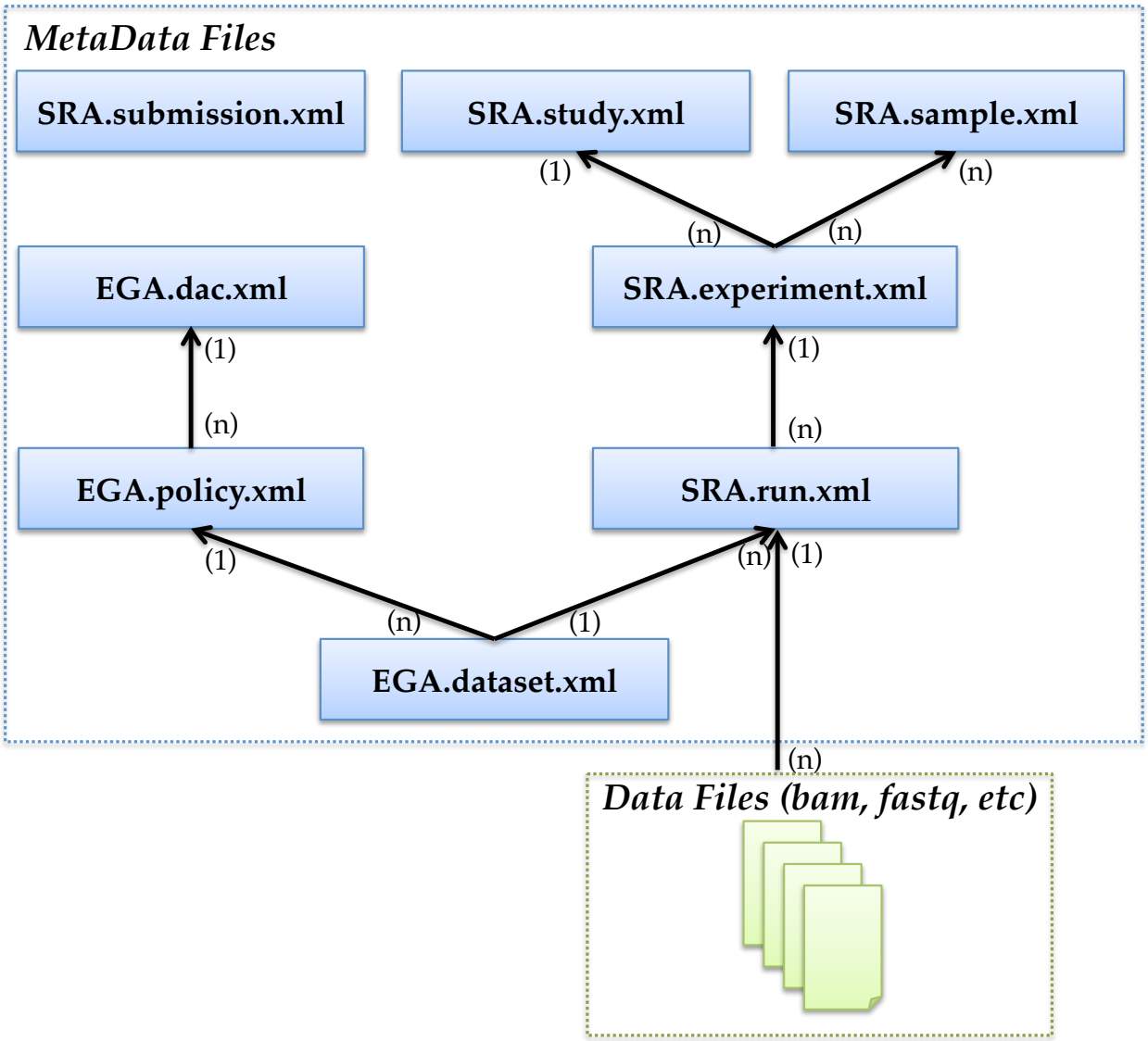
| | |
|---|---|
| SLOVAKIA | SK |
| SLOVENIA | SI |
| SOLOMON ISLANDS | SB |
| SOMALIA | SO |
| SOUTH AFRICA | ZA |
| SOUTH GEORGIA AND THE SOUTH SANDWICH ISLANDS | GS |
| SPAIN | ES |
| SRI LANKA | LK |
| SUDAN | SD |
| SURINAME | SR |
| SVALBARD AND JAN MAYEN | SJ |
| SWAZILAND | SZ |
| SWEDEN | SE |
| SWITZERLAND | CH |
| SYRIAN ARAB REPUBLIC | SY |
| TAIWAN, PROVINCE OF CHINA | TW |
| TAJIKISTAN | TJ |
| TANZANIA, UNITED REPUBLIC OF | TZ |
| THAILAND | TH |
| TIMOR-LESTE | TL |
| TOGO | TG |
| TOKELAU | TK |
| TONGA | TO |
| TRINIDAD AND TOBAGO | TT |
| TUNISIA | TN |
| TURKEY | TR |
| TURKMENISTAN | TM |
| TURKS AND CAICOS ISLANDS | TC |
| TUVALU | TV |
| UGANDA | UG |
| UKRAINE | UA |
| UNITED ARAB EMIRATES | AE |
| UNITED KINGDOM | GB |
| UNITED STATES | US |
| UNITED STATES MINOR OUTLYING ISLANDS | UM |
| URUGUAY | UY |
| UZBEKISTAN | UZ |
| VANUATU | VU |
| VATICAN CITY STATE | see HOLY SEE |
| VENEZUELA, BOLIVARIAN REPUBLIC OF | VE |
| VIET NAM | VN |
| VIRGIN ISLANDS, BRITISH | VG |

| | |
|---|---|
| VIRGIN ISLANDS, U.S. | VI |
| WALLIS AND FUTUNA | WF |
| WESTERN SAHARA | EH |
| YEMEN | YE |
| ZAMBIA | ZM |
| ZIMBABWE | ZW |

# Appendix C: EGA sequence data submission guide

The following instructions are meant to provide ICGC members with guidance on submitting raw sequence data to the European Genome-Phenome Archive (EGA). ICGC members are encouraged to consult the EGA guidelines prior to data submission. Detailed instructions on data submission are available EGA website at http://www.ebi.ac.uk/ega/page.php?page=data_submission.

### C1. Overview of the SRA metadata xml files




### C2. Examples of EGA submission templates

Below are example template files for DAC, POLICY, STUDY, DATASET, SAMPLE, EXPERIMENT, and RUN metadata. Further information regarding XML preparation can be found at: http://www.ebi.ac.uk/ena/about/sra_preparing_metadata

### C2.1 DAC and POLICY xml files

The DAC and POLICY xml files are written as per EGA's specifications and can be used by all ICGC members in their data submission without any further modifications.

- **DAC xml file**

```
<?xml version = '1.0' encoding = 'UTF-8'?>
<DAC_SET>
<DAC alias="ICGC Cancer Genome Projects" center_name="ICGC"
broker_name="">
 <TITLE>ICGC Data Access Compliance Office</TITLE>
 <CONTACTS>
   <CONTACT name="helpdesk" email="info@icgc.org" organisation="ICGC"
telephone_number=""/>
 </CONTACTS>
</DAC>
</DAC_SET>
```

- **POLICY xml file**

```
<?xml version = '1.0' encoding = 'UTF-8'?>
<POLICY_SET>
<POLICY alias="ICGC Data Access Agreements" center_name="ICGC"
broker_name="">
 <TITLE>ICGC Data Access</TITLE>
 <DAC_REF refname="ICGC Cancer Genome Projects" refcenter="ICGC"/>
 <POLICY_TEXT>Please use the ICGC website for applying access to the
data</POLICY_TEXT>
 <POLICY_LINKS>
   <POLICY_LINK>
     <URL_LINK>
       <LABEL>ICGC Data Access Agreements</LABEL>
       <URL>http://www.icgc.org </URL>
     </URL_LINK>
   </POLICY_LINK>
 </POLICY_LINKS>
</POLICY>
</POLICY_SET>
```

### C2.2 DATASET and STUDY xml files

The following examples of DATASET and STUDY xml files are written as per EGA's specifications with key items required for all ICGC submissions highlighted in yellow.

- **DATASET xml files**

```
<?xml version = '1.0' encoding = 'UTF-8'?>
<DATASETS>
<DATASET alias="EGAS00010000006-ega-20110311" center_name="OICR"
broker_name="">
 <TITLE>Pancreatic Cancer Genome Sequencing</TITLE>
 <RUN_REF refname="SC_RUN_4050_1"/>
 <RUN_REF refname="SC_RUN_4000_2"/>
 <POLICY_REF refname="ICGC Data Access Agreements" refcenter="ICGC"/>
 <DATASET_LINKS>
  <DATASET_LINK>
   <URL_LINK>
    <LABEL>ICGC Data Portal</LABEL>
    <URL>http://dcc.icgc.org</URL>
   </URL_LINK>
  </DATASET_LINK>
 </DATASET_LINKS>
</DATASET>
</DATASETS>
```

- **STUDY xml file**

```
<?xml version="1.0" encoding="UTF-8"?>
<STUDY_SET>
<STUDY alias="Pancreatic Cancer Genome Sequencing" center_name="OICR">
 <DESCRIPTOR>
  <STUDY_TITLE>Title of publication</STUDY_TITLE>
  <STUDY_TYPE existing_study_type="Whole Genome Sequencing"/>
  <STUDY_ABSTRACT> STUDY ABSTRACT AS IT COULD APPEAR IN A
PUBLICATION</STUDY_ABSTRACT>
  <CENTER_PROJECT_NAME>Pancreatic Cancer Sequencing
Initiative</CENTER_PROJECT_NAME>
 </DESCRIPTOR>
 <STUDY_ATTRIBUTES>
  <STUDY_ATTRIBUTE>
   <TAG>Consortium</TAG>
   <VALUE>ICGC</VALUE>
  </STUDY_ATTRIBUTE>
  <STUDY_ATTRIBUTE>
   <TAG>Consortium Project</TAG>
   <VALUE>ICGC Cancer Genome Projects</VALUE>
  </STUDY_ATTRIBUTE>
 </STUDY_ATTRIBUTES>
</STUDY>
```

</STUDY_SET>

### C2.3 SAMPLE, EXPERIMENT and RUN xml files

For SAMPLE, EXPERIMENT and RUN metadata, only fragments of the xml files are provided to illustrate how certain IDs, shown in red, are referenced among those files. Key items required for all ICGC submissions are highlighted in yellow.

- **Fragment of the SAMPLE xml file**

```
<SAMPLE alias="CLLS0123" ….>
  <SAMPLE_ATTRIBUTES>
    <SAMPLE_ATTRIBUTE>
      <TAG>Sample ID</TAG>
      <VALUE>CLLS0123</VALUE>
    </SAMPLE_ATTRIBUTE>
    <SAMPLE_ATTRIBUTE>
      <TAG>Donor ID</TAG>
      <VALUE>CLLD0015</VALUE>
    </SAMPLE_ATTRIBUTE>
  <SAMPLE_ATTRIBUTES>
</SAMPLE>
```

- **Fragment of the EXPERIMENT xml file**

```
<EXPERIMENT alias="EXP12345" ….. >
<STUDY_REF refname="Pancreatic Cancer Genome Sequencing"/>
<SAMPLE_DESCRIPTOR refname="CLLS0123"/>
```

- **Fragment of the RUN xml file**

```
<EXPERIMENT_REF refname="EXP12345"/>
<DATA_BLOCK member_name="CLLS0123">
<!-- member_name should be the name (usually sample alias) given in the experiment xml of a
pooled experiment. -->
          <FILES>
             <FILE filename="CLLS0001.bam" filetype="bam"/>
        </FILES>
</DATA_BLOCK>
```