GLOSSARY

Actionable genomic alteration

A genomic alteration can be considered “actionable” if it: i) predicts therapy response, ii) Affects the function of a cancer-related gene and can be targeted directly or indirectly with approved or investigational therapies, iii) is a specific eligibility criterion for enrollment onto genotype-selected trials, iv) has demonstrated the ability to establish diagnosis or influence prognosis, v) is a germline alteration that predicts drug metabolism and/or adverse effects, vi) is a germline alteration that predicts future risk of cancer or other diseases.

API

API is the abbreviation of Application Programming Interface. It is a system by which different applications can exchange information. If an application has its own API, the information can be accessed in a programmatic way following a set of instructions included in its documentation.

Assembly

http://www.ensembl.org/Help/Faq?id=216

CNV

Copy Number Variation. Regions in the sequence that are repeated or depleted. They can encompass genes causing dosage imbalances.

Deletion

Loss of one or more base pairs in the nucleotidic sequence.

Driver

A mutation that gives a selective advantage increasing the survival or the reproduction of a cell population subclone.

Germline

Relative to the inherited material that comes from the germ cells of an individual and is passed on to offspring.

Homopolymer

In DNA, portion of molecule where a nucleotide is repeated in a contiguous manner in the sequence.

Insertion

Addition of one or more base pairs in the nucleotidic sequence.

Machine learning

Field of the computer science that constructs algorithms that learn from data. The constructed algorithms build complex models that are used to make predictions.

Mutational recurrence

Instances in which the same mutation has occurred -multiple times independently throughout the history of the sequenced populations.

Oncogene

https://www.broadinstitute.org/education/glossary/oncogene

Passenger

A mutation that has no effect in the fitness of a subclone of cells.

Pipeline

Set of programming elements executed in a sequential order where the output of one element is the input of the next one.

Reference Genome

Sequence of nucleic acids assembled by specialists representative of a specie. It is constructed from the sequencing of multiple individuals and can be publicly accessed in browsers as Ensembl or UCSC genome browsers.

Script

Piece of informatics code codified in a particular programming language containing a set of orders that when executed performs a particular task.

SNV

Single Nucleotide Variant. Variation in a single base.

Somatic

Relative to the biological material forming the body of an organism, with tissue-specific characteristics and that is not passes on to offspring.

Structural variant

Region of DNA greater than 1 kb with a variation in the chromosome. It encompasses deletions, duplications, CNVs, insertions, inversions and translocations

Transcriptional alteration types

http://www.ensembl.org/info/genome/variation/predicted_data.html

Tumor suppressor gene

https://www.broadinstitute.org/education/glossary/tumor-suppressor-gene