GLOSSARY

Systematic screens Studies covering substantial fraction of the genome, reporting

somatic variants mapped to genomic coordinates in a tabular

format.

Targeted screens Studies providing sequencing data for a subset of genes as

opposed to indiscriminate whole exome or whole genome screening. Often, information about particular variants is spread across the different sections of the publication, its

figures, and supplementary data.

VEP Variant Effect Predictor is Ensembl tool used by COSMIC to

annotate genetic variants, i.e. map them to the reference genome assemblies for further classification into different

variant types.

Data In the context of COSMIC curation, a unit of curated

information referring to a genetic variant and a cancer type.

Metadata Data providing information about one or more aspects of the

COSMIC data. For the core COSMIC curation, it includes ethnicity, age, drug response, therapy, cytogenetics, tumour

grade and stage, and others.

HGVS Human Genome Variation Society nomenclature for

standardised description of sequence variants in DNA, RNA,

and protein sequences.

Somatic mutations mutations (variants) that arise in somatic cells. Somatic

mutations are not inherited by the offspring.