Course overview

Bioinformatic Analysis for Cancer Genomics

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Content

- The Teams help US!
- What we will learn from the course?
- What you can do after the course?
- Github and youtube

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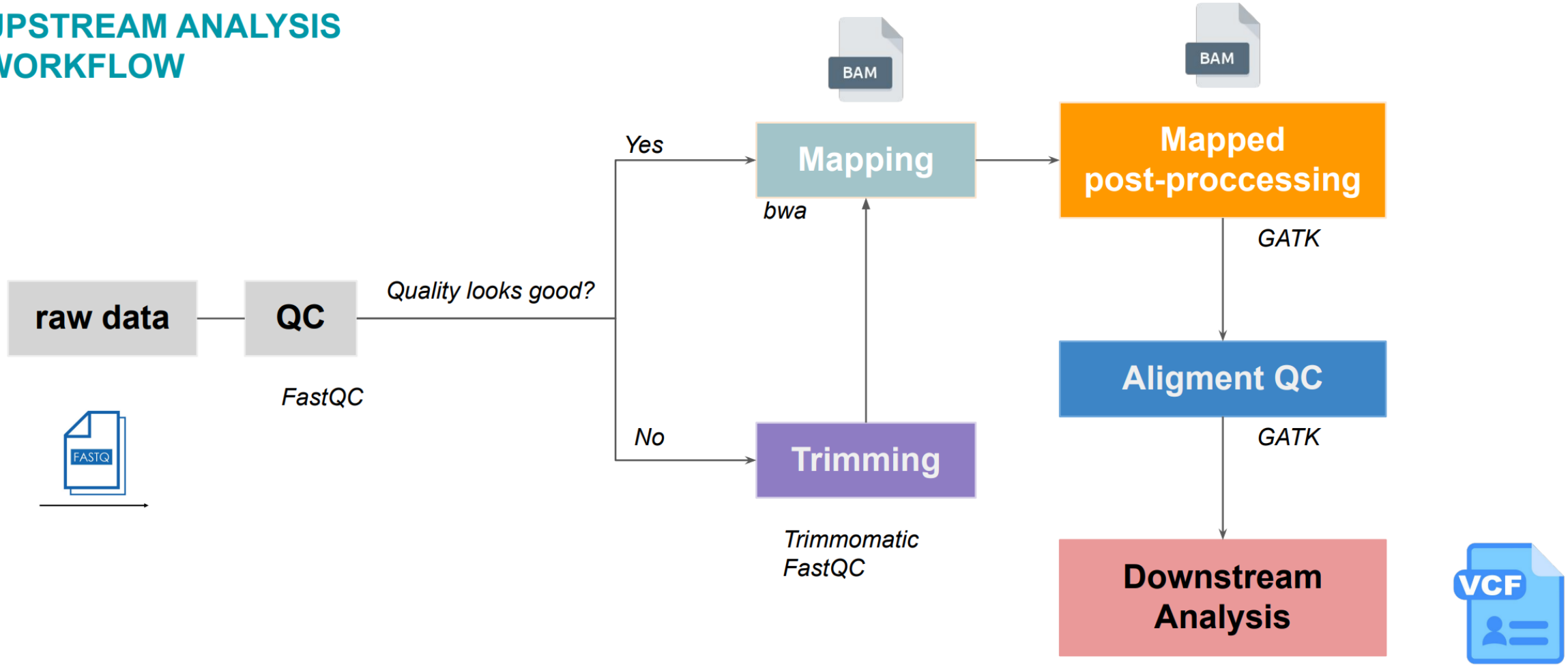
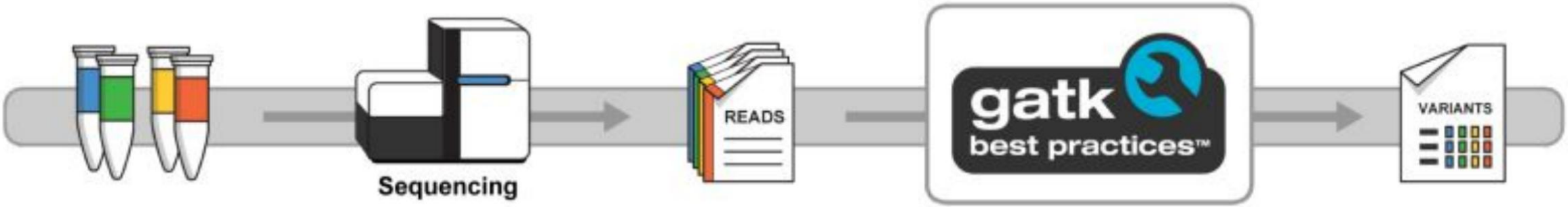
UPSTREAM ANALYSIS WORKFLOW



Adapted from Duy's slide

Workflows for all major variant classes



	GERMLINE	SOMATIC
SNPs & INDELs	**HaplotypeCaller GVCF**	**Mutect2**
Copy Number	**GATK gCNV**	**GATK CNV + aCNV**
Structure Variation	**GATK SVDiscovery** (beta)	(planned)

Adapted from Minh's slide

Methods

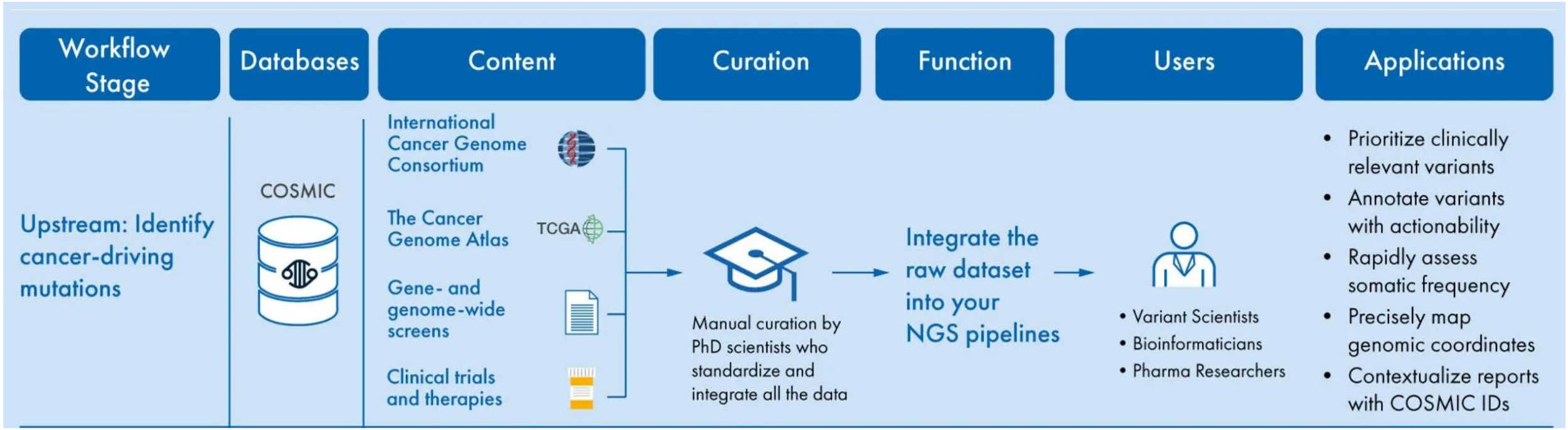
No	Sequencing Platforms	Read mapping	Single Nucleotide Variant (SNV) Calling	Structural Variant (SV) and Copy Number Variant (CNV) Calling	Variant Annotation
1	Illumina	BWA-mem, Bowtie2, SOAP2, Minimap2, BLASR, NGMLR …	SAMtools, Deepvariant, GATK, SpeedSeq …	DELLY, BreakDancer, LUMPY, GRIDSS, TARDIS, SURVIVOR, PBHoney, NanoSV, Manta, CNVnator …	HPO, ClinVar, Varsome, Gnomad, Cosmic, ANOVAR …
2	Onso (Short read, PacBio) Revio (Long read, PacBio)				
3	Ion Torrent PGM				
4	MGI				
5	GeneMind				
6	Aviti (Element Biosciences)				
7	ONT				

10.1186/s12864-024-10239-9 and 10.1186/s13059-019-1828-7

What we will learn from the course?

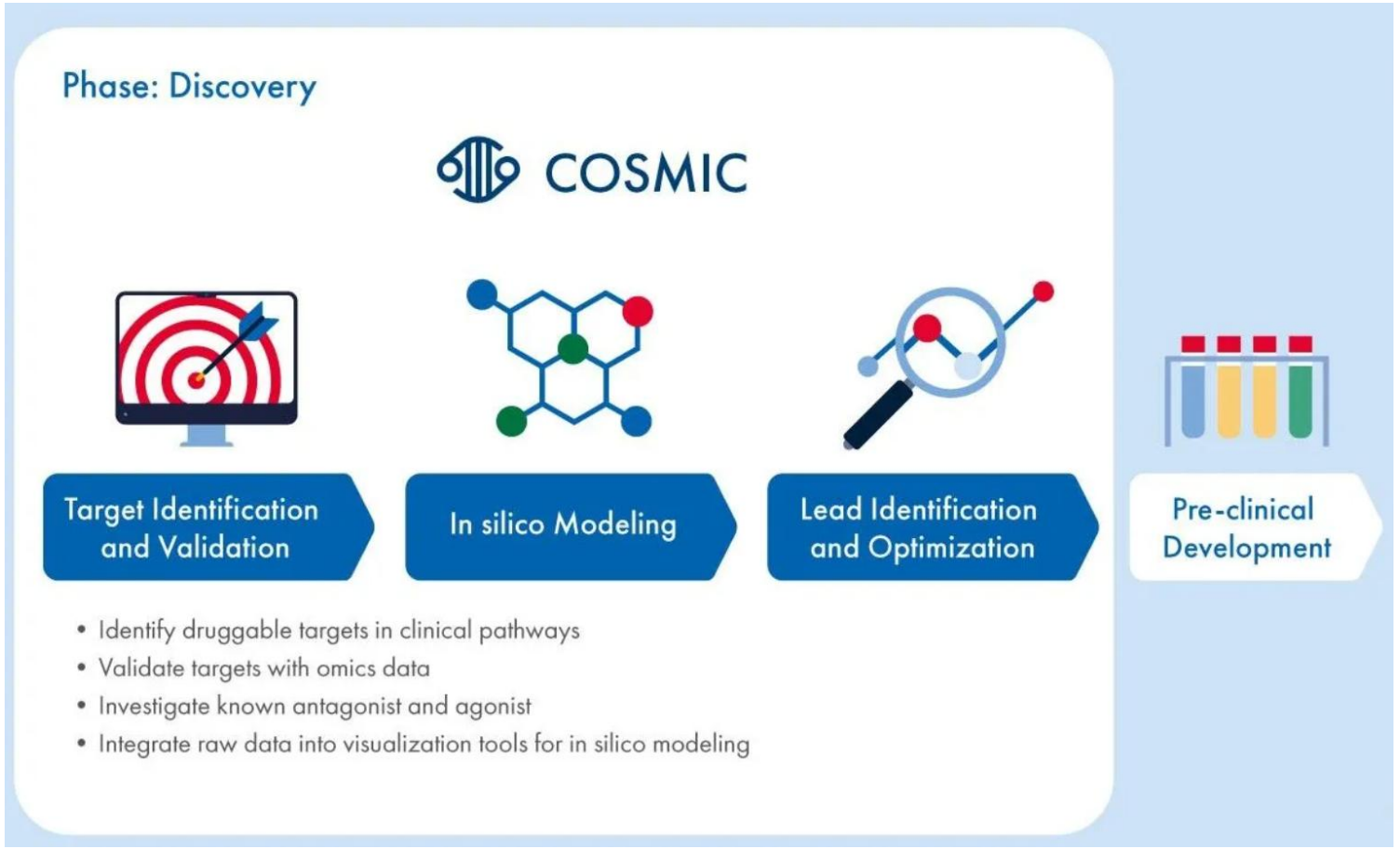
1. Install and use Ubuntu or Google Colab
2. Master programming language (Bash and R), Linux command line, and some bioinformatic tools
3. Manipulate Next Generation Sequencing data of DNA for both research and clinical application
4. Plot ready for publication figures and statistically analyzing for a manuscript
5. Downstream analysis of DNA-seq
6. Understanding publications of DNA-seq

COSMIC for clinical NGS testing



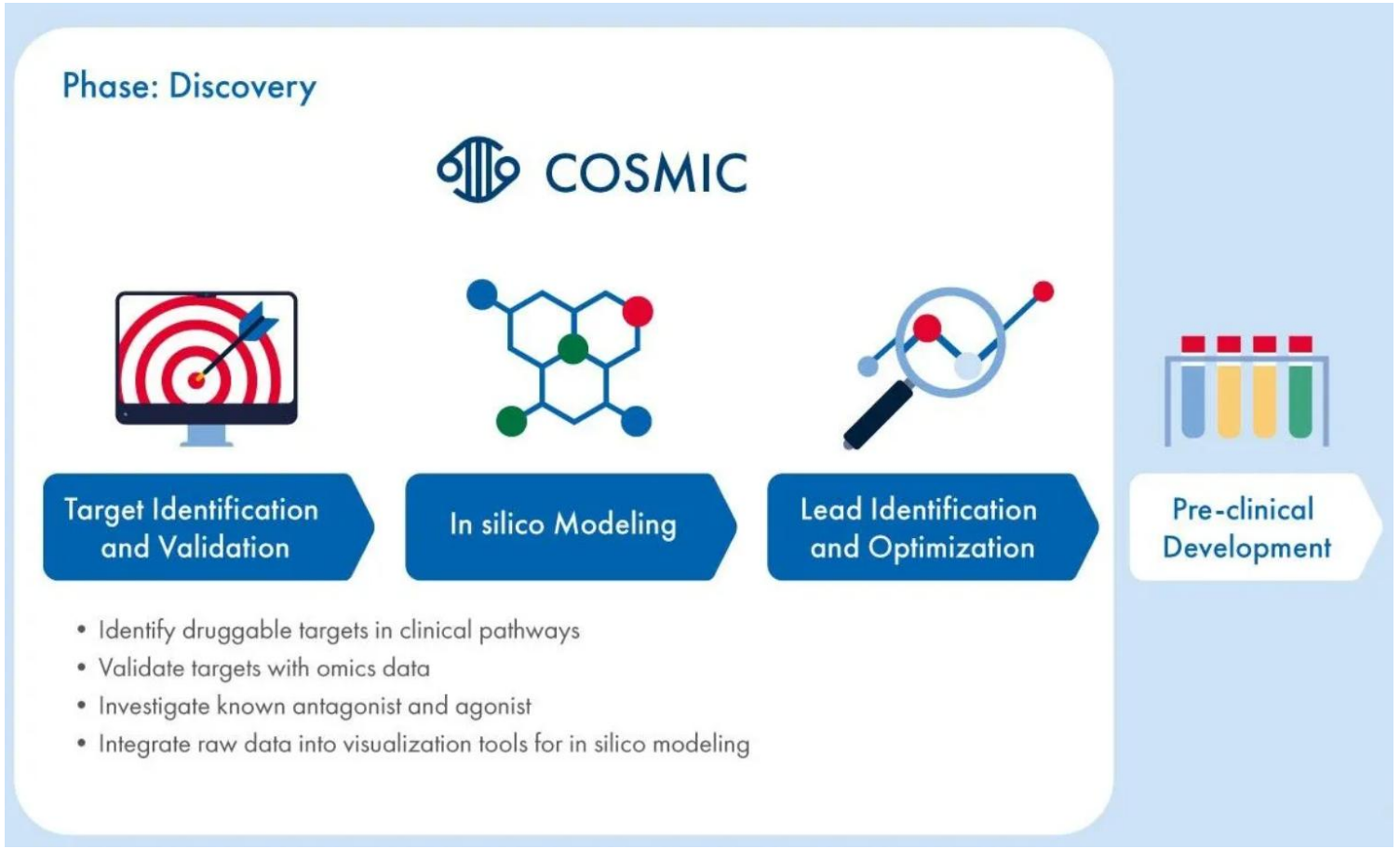
https://digitalinsights.qiagen.com/somatic-mutation-databases-for-clinical/

COSMIC in the cancer drug discovery



https://digitalinsights.qiagen.com/cosmic-hsmd-biopharma/

COSMIC in the cancer drug discovery



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COSMIC for biopharma research

View applications of COSMIC for biopharma research

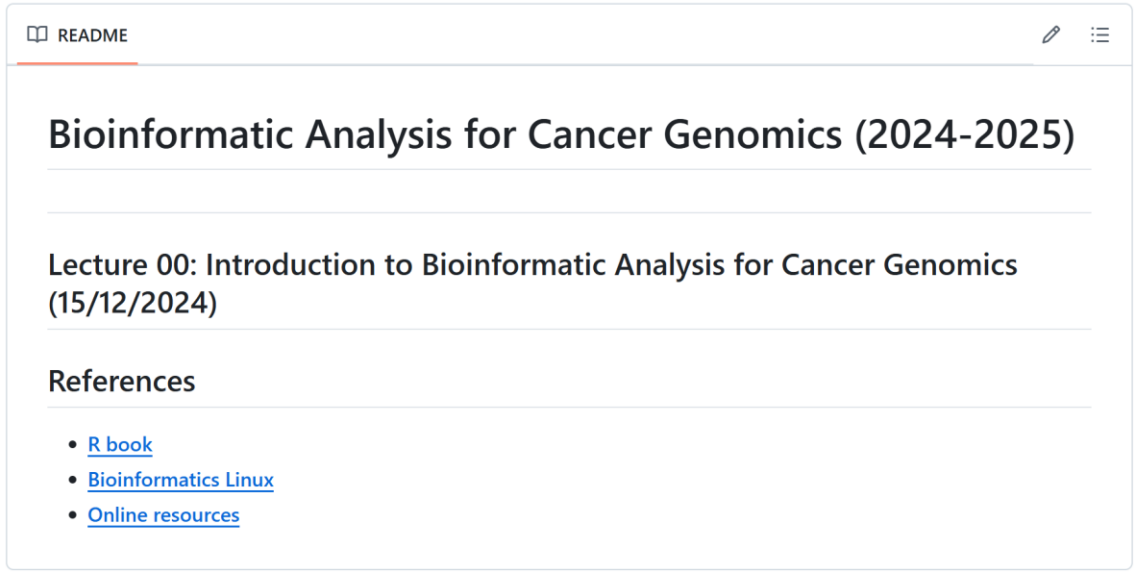
Developed and maintained by Wellcome Sanger Institute, the latest release, COSMIC v101 (November 2024), provides comprehensive coverage of all human genes, featuring over 24.9 million genetic variants derived from 1.5 million tumor samples spanning more than 1,400 cancer types. Notably, the release focused on curating somatic mutation profiles in rare cancers and cancers with significant medical need, as well as the prioritization of studies focusing on whole genome or whole exome data.

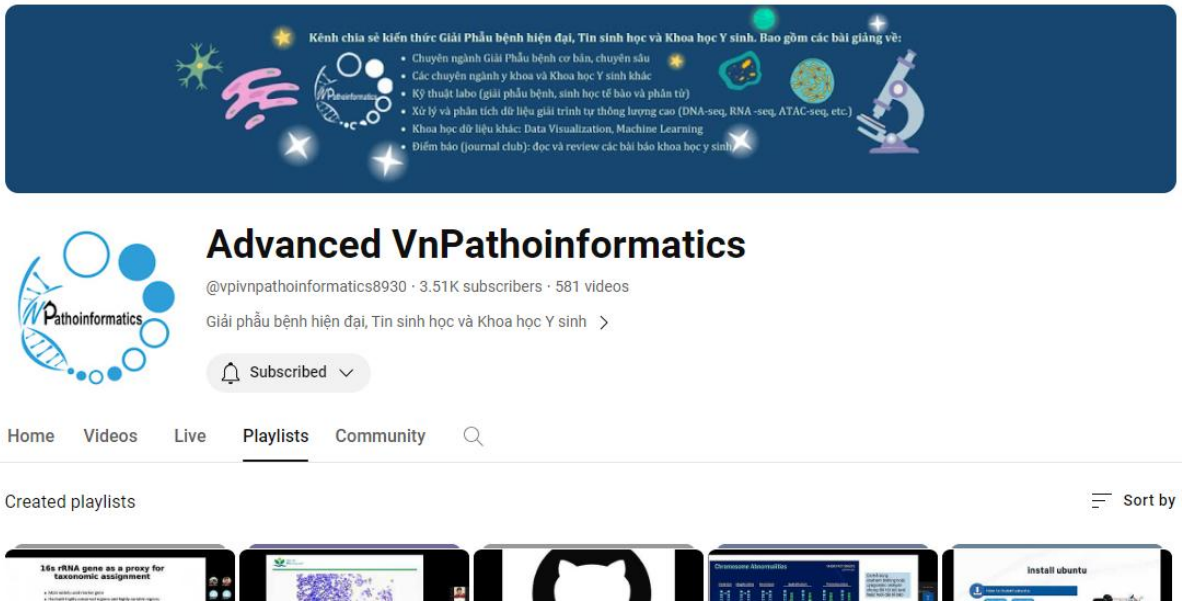
- Integrates somatic data from multiple sources, including the International Cancer Genome Consortium, The Cancer Genome Atlas, gene- and genome-wide screens, clinical trials and therapies.
- Data is translated into a standardized format and available through downloadable datasets and user-friendly data exploration tools.
- Every six months, COSMIC content and features are updated to ensure you remain informed on the latest findings.

Github and youtube

1. Github: Lecture Slides, Exercises, Books and Q&A

   https://github.com/luuloi/Bioinformatic_Analysis_for_Cancer_Genomics





2. Youtube:

https://www.youtube.com/@vpivnpathoinformatics8930/playlists

Thank you for your listening!