

# Overview of Illumina's portfolio in life sciences

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# BaseSpace™ Sequence Hub

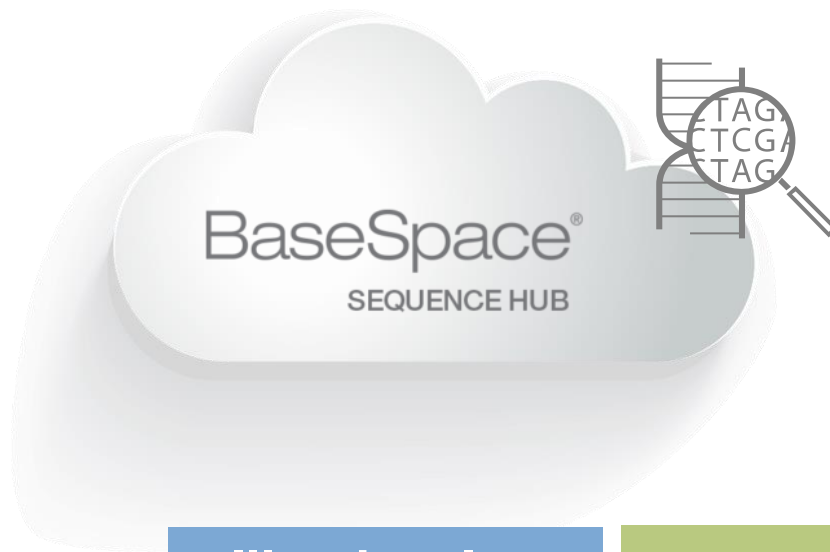


# BaseSpace<sup>®</sup>

## SEQUENCE HUB



## Over 70 published applications in BaseSpace™ Sequence Hub



<b>16S Metagenomics</b> Illumina, Inc. Analyze DNA from amplicon sequencing of prokaryotic 16S small subunit rRNA genes. Provides results of taxonomic... Categories: Targeted Sequencing, Metagenomics	<b>Ampliseq DS</b> Illumina, Inc. Enables streamlined analysis of the Illumina TruSight Tumor 25 and dual post... Categories: Resequencing, Targeted Sequencing	<b>BWA Enrichment</b> Illumina, Inc. The core algorithms in the BWA Enrichment workflow are the BWA-SW and BWA-MEM... Categories: Resequencing, Targeted Sequencing, Variant Analysis	<b>Cuffdiff Assembly &amp; DE</b> Illumina, Inc. Quickly assess novel transcript isoforms and gene expression levels from RNA-Seq alignment results. Categories: Targeted Sequencing, RNA-Seq, Differential Expression, HPA
<b>Enrichment</b> Illumina, Inc. The Enrichment app rapidly aligns samples using base and performs read, 5'... Categories: Resequencing, Targeted Sequencing, Variant Analysis, HPA	<b>MethylKit</b> Illumina, Inc. DNA methylation analysis for targeted and whole genome bisulfite sequence data. Categories: Targeted Sequencing, MethylSeq, HPA	<b>mtDNA Variant Analyzer</b> Illumina, Inc. The mtDNA Variant Analyzer app enables simplified visualization of a loop and... Categories: Targeted Sequencing, Variant Analysis	<b>mtDNA Variant Processor</b> Illumina, Inc. The mtDNA Processor app enables simplified visualization of a loop and... Categories: Targeted Sequencing, Variant Analysis
<b>RNA-Seq Express</b> Illumina, Inc. Align RNA-seq reads with the STAR aligner, followed by differential expression... Categories: RNA-Seq, Differential Expression, HPA	<b>RNA-Seq Alignment</b> Illumina, Inc. Aligns RNA-seq reads and detects gene fusions using standard methods. Integrates... Categories: Targeted Sequencing, RNA-Seq, Gene Fusion Detection, HPA	<b>Small RNA</b> Illumina, Inc. Aligns reads and outputs hits. Performs optional novel precursor discovery... Categories: Small RNA, RNA-Seq, Differential Expression	<b>SingleCell RNA-Seq</b> Illumina, Inc. Single-cell RNA-Seq analysis. Categories: RNA-Seq
<b>TruSight Amplification</b> Illumina, Inc. The TruSight Amplification workflow enables variant detection and detection of... Categories: Resequencing, Targeted Sequencing, Variant Analysis	<b>TruSight Long Read Assembly</b> Illumina, Inc. Combines synthetic long reads from shorter sequencing reads, for genome... Categories: Synthetic Long Reads	<b>TruSight Phasing Analysis</b> Illumina, Inc. Performs haplotype phasing for analysis of alleles originating from... Categories: Variant Analysis, Synthetic Long Reads	<b>TruSight Targeted RNA</b> Illumina, Inc. Analyzes RNA samples prepared using TruSight Targeted RNA Expression Kits... Categories: Targeted Sequencing, RNA-Seq, Gene Fusion Detection, Differ...
<b>TruSight Tumor 15</b> Illumina, Inc. The TruSight Tumor 15 is a 150K amplicon-based NGS assay for 15 genes... Categories: Targeted Sequencing, Variant Analysis	<b>TruSight Tumor 170</b> Illumina, Inc. The TruSight Tumor 170 BaseSpace app analyzes reads from DNA and RNA... Categories: Targeted Sequencing, Variant Analysis, HPA	<b>TruSight Tumor 170 + Watson for Genomics Converter</b> Illumina, Inc. The app converts TruSight 170 raw results to a format that is compatible with... Categories: Targeted Sequencing, Variant Analysis, HPA	<b>Tumor Normal</b> Illumina, Inc. The TumorNormal Sequencing App is designed to detect somatic variants... Categories: Tumor Normal, Variant Analysis, HPA

### Illumina, Inc



**16S Metagenomics**  
Illumina, Inc.  
Targeted Sequencing, Metagenomics



**RNA-Seq Alignment**  
Illumina, Inc.  
RNA-Seq, Gene Fusion Detection, Var...

Fully supported  
by Illumina

### DRAGEN™



**DRAGEN Enrichment**  
Illumina, Inc.  
Resequencing, Targeted Sequencing

Fully supported  
by Illumina

### BaseSpace Labs



**Kraken Metagenomics**  
BaseSpace Labs  
Resequencing, Metagenomics



**Pindel**  
BaseSpace Labs  
Resequencing, Targeted Sequencing,...

basespacelabs@ill  
umina.com for  
additional support

### 3rd Party



**DeepChek@-HBV**  
ABL S.A.  
Targeted Sequencing, Variant Analysis...



**MiXCR Immune Repertoire ...**  
MiLaboratory  
Targeted Sequencing, Variant Analysis

Supported by  
3rd party



**Extensive and secure global framework** built on Amazon Web Services (AWS)



**ISO 27001** Information Security and Quality Management Systems



**Support for HIPAA-compliance** in a regulated environment



**ISO 13485 certified** for the design, development, manufacture, distribution, installation, and servicing



**General Data Protection Regulation** to manage authorization, authentication, and data control

# 16S Metagenomics app

# 2

# Overview: 16S Metagenomics app



16S Metagenomics  
Illumina, Inc.  
Targeted Sequencing, Metagenomics, HIPAA

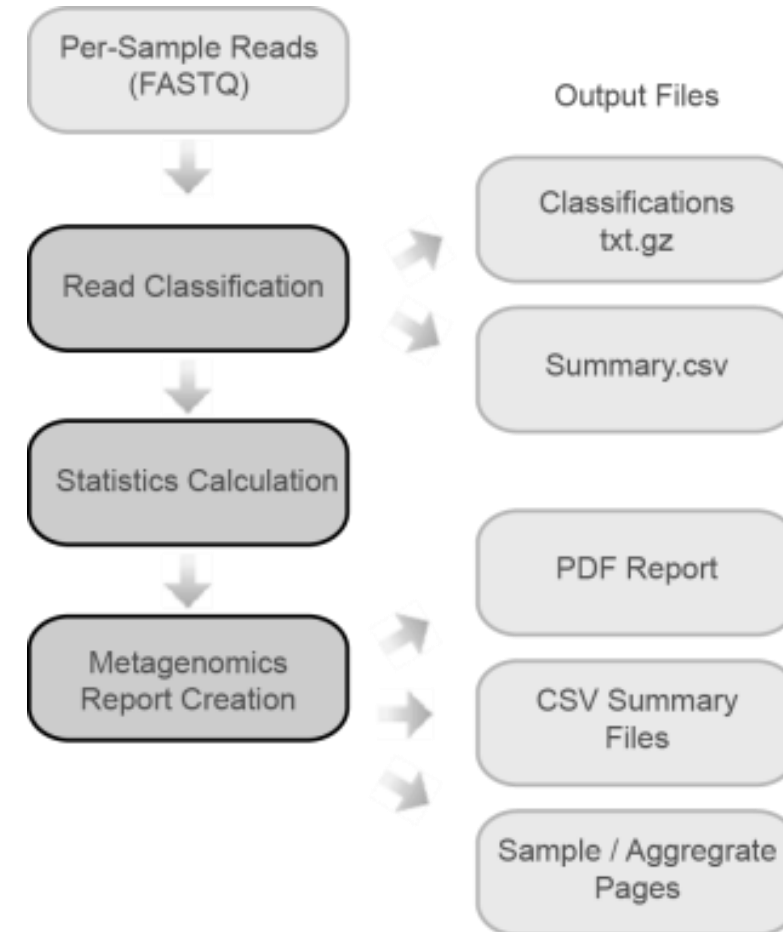


Taxonomic classification of Prokaryotes

Relative abundances

Species Richness and diversity

New update contains choice of database

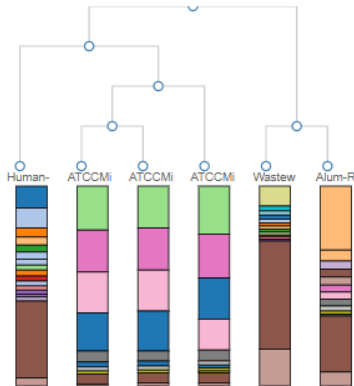


# Overview: 16S Metagenomics app

## Visualizations

### HIERARCHICAL CLUSTERING DENDROGRAM

This dendrogram shows a hierarchical clustering of samples based on genus-level classifications. The barchart beneath each sample show the relative abundance of its genus-level classifications. Mouseover the barcharts for taxonomic labels.



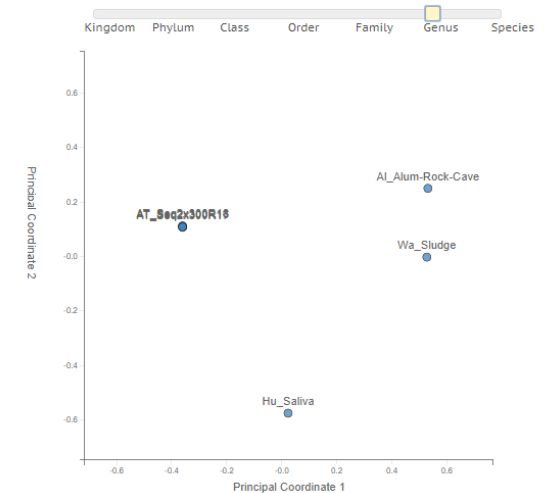
### SPECIES DIVERSITY RESULTS

Shannon species diversity measures the entropy of Species-level classifications in the sample. See [http://en.wikipedia.org/wiki/Shannon-Wiener\\_index](http://en.wikipedia.org/wiki/Shannon-Wiener_index) for more information.

Sample Number	Sample ID	Shannon Species Diversity	Number of Species Identified
1	Wastewater_Sludge	3.635	4,886
2	Alum_Rock_Cave	2.879	4,030
3	Human_Saliva	3.379	2,989
4	ATCCMISeq2x300R18	1.308	370
5	ATCCMISeq2x300R17	1.322	426
6	ATCCMISeq2x300R16	1.338	535

### PRINCIPAL COORDINATE ANALYSIS (PCOA)

This scatterplot shows a Principal Coordinate Analysis (PCoA) of the normalized relative abundance of all samples. The PCoA measures differences in the distribution of taxonomic classifications between samples, up to a fixed taxonomic level. Select the maximum taxonomic level to compare sample classification results:



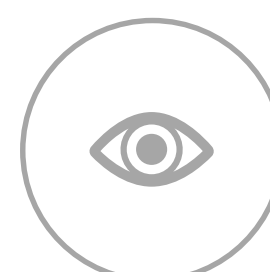
Easy to launch



Basic statistical  
analysis



Reports



Interpretation



# Overview: 16S Metagenomics app

## 16S database creator



### 16S Metagenomics Database Creator

BaseSpace Labs

 Bookmarked  Help

#### Analysis Name

16S Metagenomics Database Creator 04/09/2020 9:06:51

#### Save Results To

Select Project

#### Input File

Select Dataset File(s)

#### Seed Size

32

#### BaseSpace Labs Disclaimer

##### BaseSpace Labs

☐ I acknowledge and agree that (i) this is a BaseSpace Labs App, (ii) I am using it AS-IS without any warranty of any kind, (iii) Illumina has no obligation to provide any technical support for this App, and (iv) Illumina has no liability for my use of this App, including without limitation, any loss of data, incorrect results, or any costs, liabilities, or damages that result from use of this App.

Launch Application



<https://basespace.illumina.com/projects/140501372>

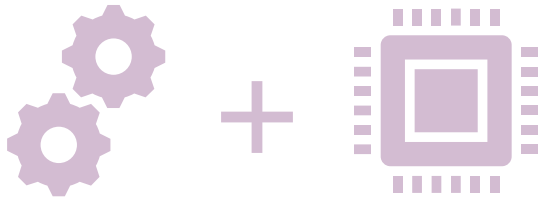
# DRAGEN™ Metagenomics app

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# Hardware accelerated secondary analysis



## What is DRAGEN?



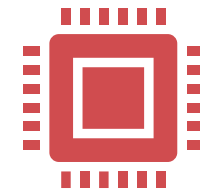
Dynamic Read Analysis for  
**GEN**omics

## What does it do?



Processing massive amounts of  
data.

## How does it work?



DRAGEN's Field Programmable  
Gate Array (FPGA) cards



Library Prep

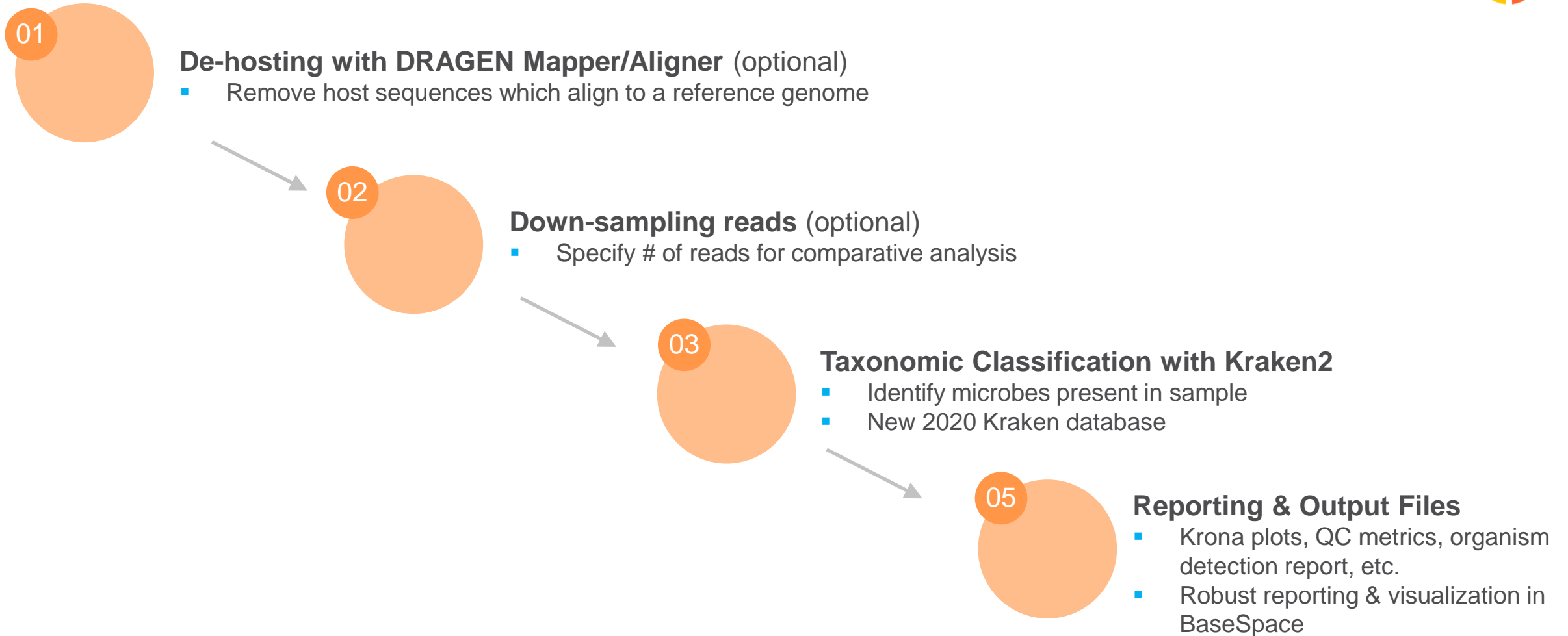
Sequencing

**Secondary Analysis**

Tertiary Analysis

# Shotgun metagenomics

## DRAGEN™ Metagenomics app



# Shotgun metagenomics

## DRAGEN™ Metagenomics app – Kraken2



**K-mer-based approach**



**Provides a fast taxonomic classification of metagenomic sequence data**



**A translated search mode, providing increased sensitivity in viral metagenomics analysis**

### Expanded Kraken2 database (March 2020) :

- archaea: RefSeq complete archaeal genomes/proteins
- bacteria: RefSeq complete bacterial genomes/proteins
- viral: RefSeq complete viral genomes/proteins
- human: GRCh38 human genome/proteins
- fungi: RefSeq complete fungal genomes/proteins
- plant: RefSeq complete plant genomes/proteins
- protozoa: RefSeq complete protozoan genomes/proteins

# Shotgun metagenomics

## DRAGEN™ Metagenomics app

Analysis: DRAGEN Metagenomics Pipeline Feature - de-hosting on

SUMMARY
REPORTS
INPUTS
FILES

Launch App
File
Status
Share
Move to Trash

Summary
CoV229ECtrlPat Report
CoVOC43CtrlPat Report

### Summary Report

#### AGGREGATE SUMMARY

Aggregate Organism Detection Report [Export \(CSV\)](#)

Detection Threshold (minimum 1):  Color Palette:

Copy CSV Excel

Search:

Sample ID	Severe acute respiratory syndrome coronavirus 2	Human coronavirus 229E	Human coronavirus HKU1	Human coronavirus NL63	Human coronavirus OC43	coronavirus	Influenza A	Influenza B	Listeria
CoV229ECtrlPat	ND (0)	Detected (283652)	ND (0)	ND (0)	ND (0)	Detected (286171)	ND (0)	ND (0)	Detected (1)
CoVOC43CtrlPat	ND (0)	Detected (1)	ND (0)	ND (0)	Detected (187546)	Detected (281216)	ND (0)	ND (0)	ND (0)

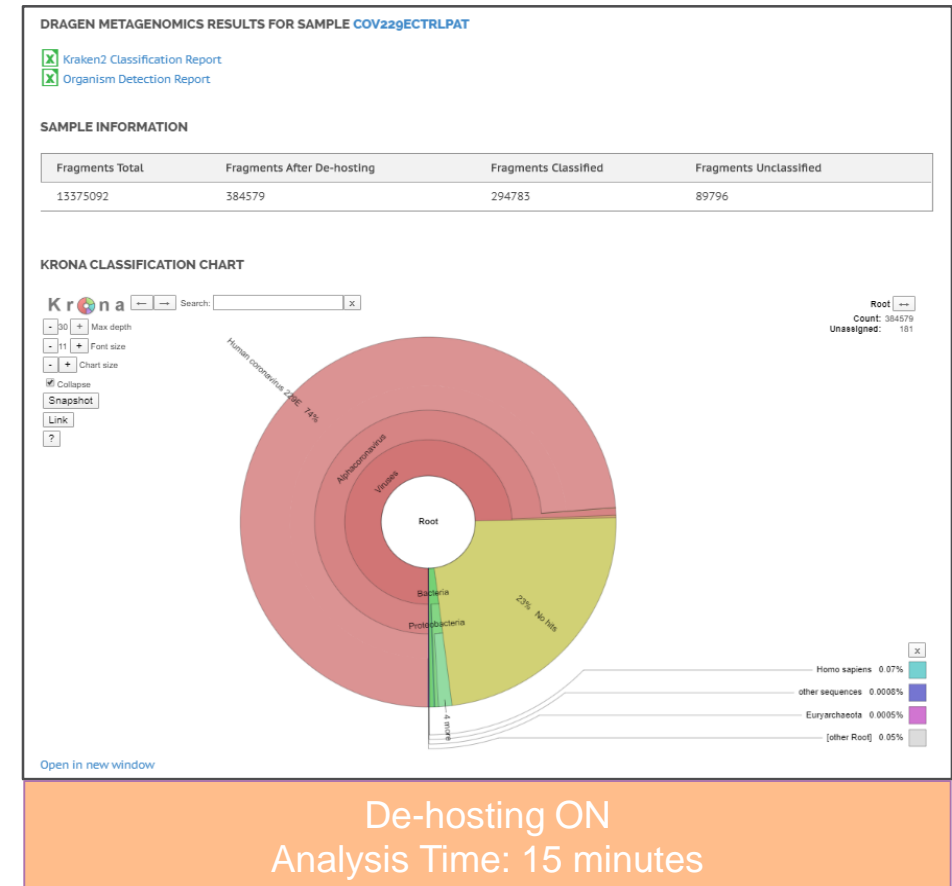
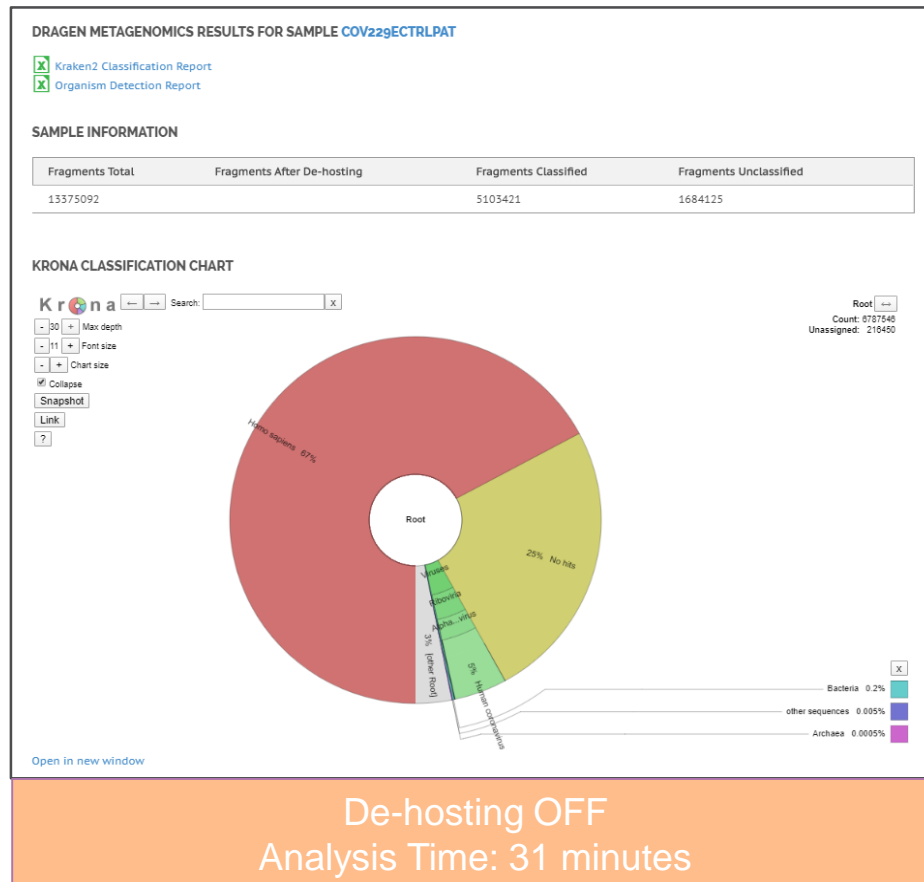
Showing 1 to 2 of 2 entries

Multi-sample reporting available in BaseSpace Sequence Hub only

# Shotgun metagenomics

## DRAGEN™ Metagenomics app - Built-in de-hosting

Accelerate analysis using the DRAGEN Aligner to de-host



# Thank you

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