



Netflow y automatización de pipeline de análisis, uso de pipeline y reproducibilidad.

Evelin González F.
evelyn.gonzalez@uoh.cl

Organización de las clases: Parte 1

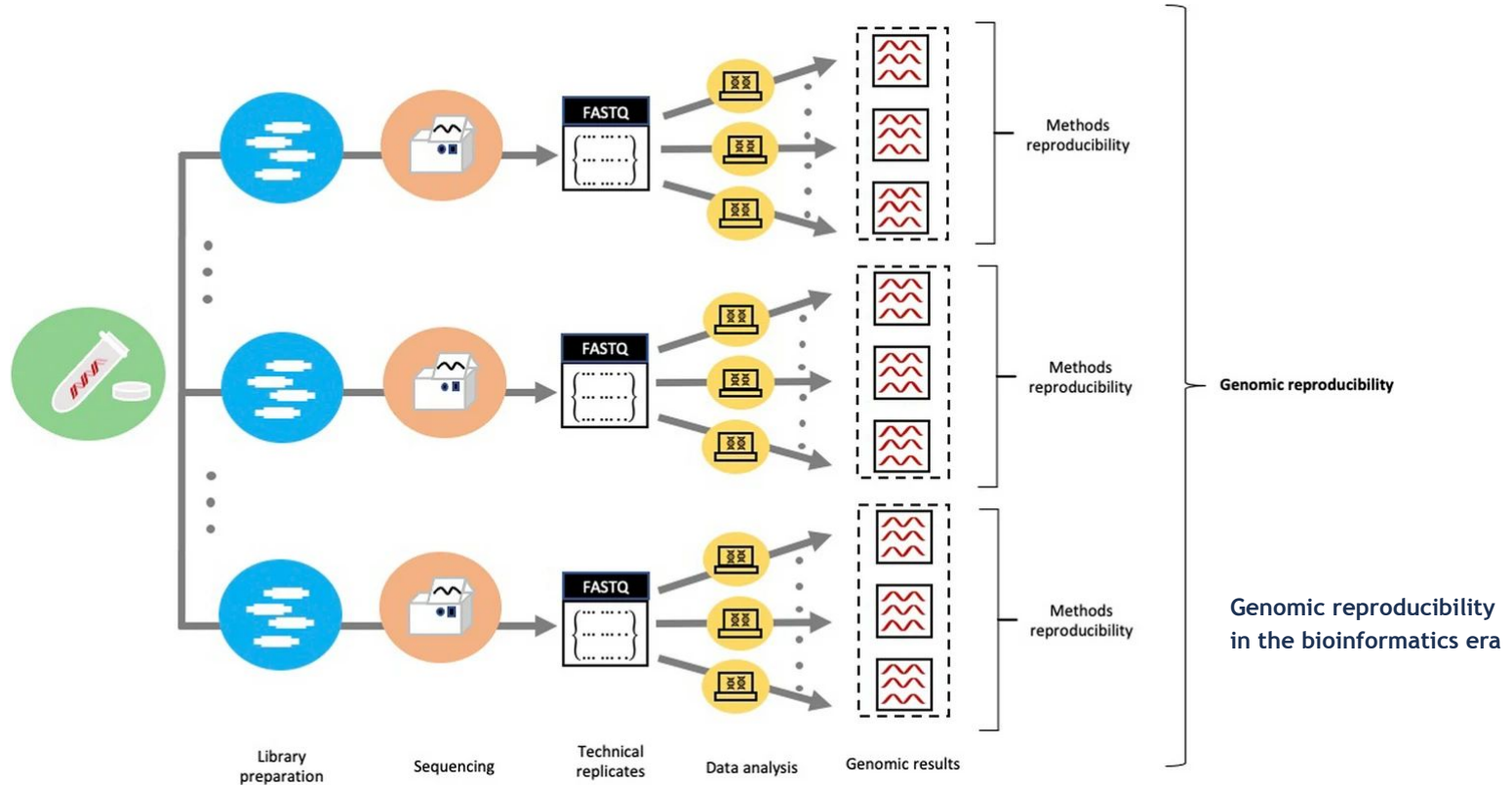
Clase 1: Preprocesamiento de los datos y reportes de calidad.

Clase 2: Llamado de variantes y visualización con IGV

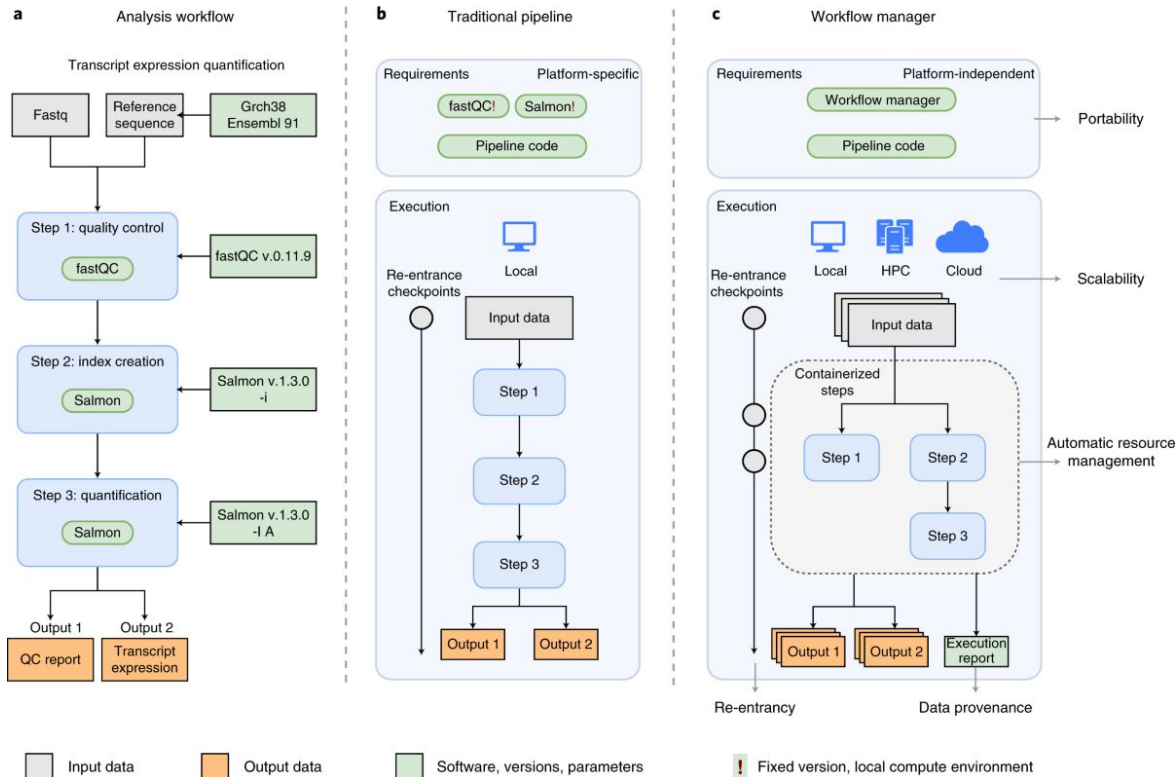
Clase 3: Anotación de variantes (uso de bases de datos y clasificación de variantes patogénicas).

Clase 4: Netflow y automatización de pipeline de análisis, uso de pipeline y reproducibilidad. Taller Práctico.

Reproducibilidad genómica en bioinformática



Flujos de trabajos



NEXTFLOW

Un motor de flujos de trabajo para la automatización y escalabilidad de análisis bioinformáticos.

Principales características:

Integración con **contenedores** (Docker, Singularity).

Escalabilidad en **HPC** y la nube (AWS, Azure, Google Cloud).

Reproducibilidad y portabilidad de pipelines.

Beneficios en bioinformática:

- Optimización de análisis complejos.
- Trazabilidad y manejo de datos masivos.
- Reducción del tiempo de ejecución mediante paralelización.

Gestores de flujos de trabajo

Table 1 | Overview of workflow managers for bioinformatics (top, editable version; bottom, image version)

| Tool | Class | Ease of use ^a | Expressiveness ^b | Portability ^c | Scalability ^d | Learning resources ^e | Pipeline initiatives ^f |
|---------------------------|------------------------------------|--------------------------|-----------------------------|--------------------------|--------------------------|---------------------------------|-----------------------------------|
| Galaxy | Graphical | ●●● | ●○○ | ●●● | ●●● | ●●● | ●●○ |
| KNIME | Graphical | ●●● | ●○○ | ○○○ | ●●○ | ●●● | ●●○ |
| Nextflow | DSL | ●●○ | ●●● | ●●● | ●●● | ●●● | ●●● |
| Snakemake | DSL | ●●○ | ●●● | ●●○ | ●●● | ●●○ | ●●● |
| GenPipes | DSL | ●●○ | ●●● | ●●○ | ●●○ | ●●○ | ●●○ |
| bPipe | DSL | ●●○ | ●●● | ●●○ | ●●○ | ●●○ | ●○○ |
| Pachyderm | DSL | ●●○ | ●●● | ●○○ | ●●○ | ●●● | ○○○ |
| SciPipe | Library | ●●○ | ●●● | ○○○ | ○○○ | ●●○ | ○○○ |
| Luigi | Library | ●●○ | ●●● | ●○○ | ●●○ | ●●○ | ○○○ |
| Cromwell + WDL | Execution + workflow specification | ●○○ | ●●○ | ●●● | ●●○ | ●●○ | ●●○ |
| cwltool + CWL | Execution + workflow specification | ●○○ | ●●○ | ●●○ | ○○○ | ●●● | ●●○ |
| Toil + CWL/ WDL/Python | Execution + workflow specification | ●○○ | ●●● | ●○○ | ●●● | ●●○ | ●●○ |

Please refer to Supplementary Table 1 for details. This information is based on online documentation and manuscripts and may not be reflective of the current state of the projects. Scores for Galaxy are based on the graphical user interface. ^aEase of use: graphical interface with execution environment (score of 3), programming interface with in-built execution environment (score of 2), separated development and execution environment (score of 1). ^bExpressiveness: based on an existing programming language (3) or a new language or restricted vocabulary (2), primary interaction with graphical user interface (1). ^cPortability: integration with three or more container and package manager platforms (3), two platforms are supported (2), one platform is supported (1). ^dScalability: considers cloud support, scheduler and orchestration tool integration, and executor support. Please refer to Supplementary Table 1. ^eLearning resources: official tutorials, forums, and events (3), tutorials and forums (2), tutorials or forums (1). ^fPipelines initiatives: community and curated (3), community or curated (2), not community or curated (1).

Seqera Seqera AI (Beta) Pipelines Containers Products Forum Docs Login Sign up

Pipelines Showing all 80

Search pipelines...

nf-core/rnaseq
RNA sequencing analysis pipeline using STAR, RSEM, HISAT2 or Salmon with gene/isoform counts and extensive quality control.
nextflow pipeline workflow nf-core + 2 more
☆ 786 · 198 KB · Updated 6 months ago Launch

nf-core/sarek
Analysis pipeline to detect germline or somatic variants (pre-processing, variant calling and annotation) from WGS / targeted sequencing
nextflow pipeline workflow nf-core + 16 more
☆ 340 · 470 KB · Updated 6 months ago Launch

nf-core/mag
Assembly and binning of metagenomes
nextflow pipeline workflow nf-core + 9 more
☆ 182 · 260 KB · Updated 6 months ago Launch

nf-core/chipseq
ChIP-seq peak-calling, QC and differential analysis pipeline.
nextflow pipeline workflow nf-core + 5 more
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nextflow pipeline workflow nf-core + 9 more
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nf-core/atacseq
ATAC-seq peak-calling and QC analysis pipeline
nextflow pipeline workflow nf-core + 2 more
☆ 161 · 198 KB · Updated 6 months ago Launch

Filters

Organizations

nf-core epi2me-labs
FredHutch qbic-pipelines
sanger-tol Arcadia-Science
annotation-cache PGScatalog
genepi NBISweden

Keywords

nextflow pipeline workflow
nf-core genomics ma
metagenomics illumina
rna-seq variant-calling
nanopore single-cell
bioinformatics
mass-spectrometry annotation
next-generation-sequencing
assembly long-read-sequencing
chip-seq atac-seq

Help

What is Seqera Pipelines?
Where are these pipelines hosted?
How is this list collected?
How are pipelines tested?
How can I run these pipelines?
Can I submit my own pipeline?
How do I suggest a feature?

Pipelines de análisis

Seqera es una compañía que desarrolla soluciones para la gestión y escalabilidad de flujos de trabajo bioinformáticos, siendo la creadora de **Nextflow**.

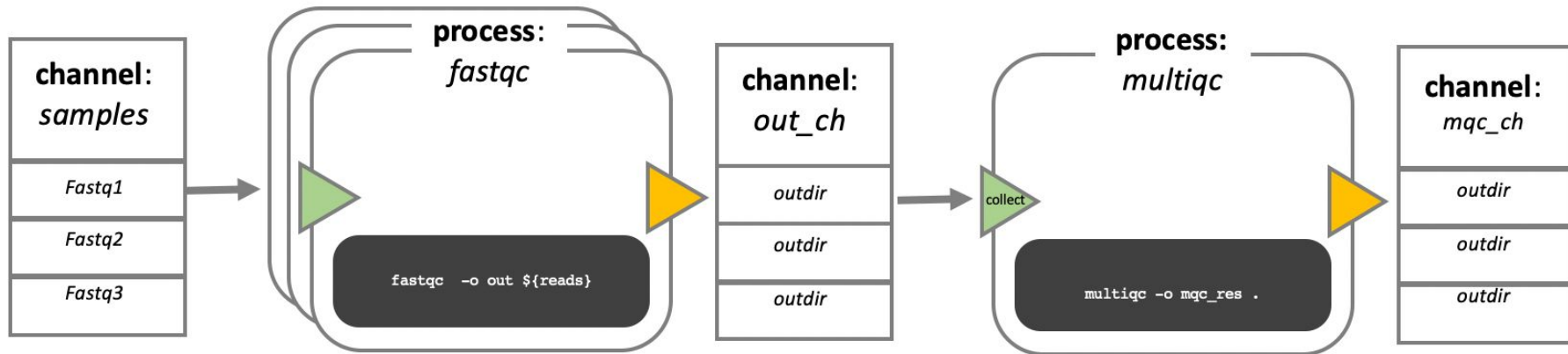
<https://seqera.io/pipelines/>

¿Qué es Nextflow?

- Herramienta para automatizar flujos de trabajo en bioinformática.
- Diseñada para procesar grandes volúmenes de datos.
- Compatible con múltiples entornos: local, clústeres HPC, nube.
- Lenguaje declarativo sencillo, basado en scripts.
- Facilita la reproducibilidad y el control de versiones.
- Ejecuta tareas en paralelo de manera eficiente.

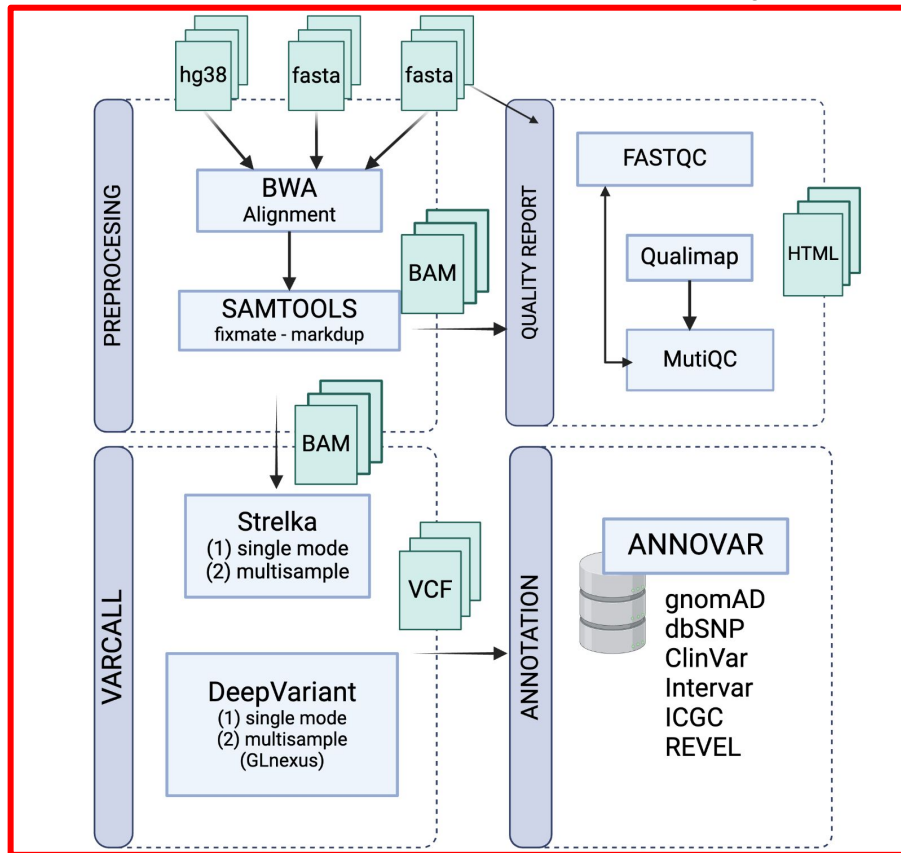


Procesos en Nextflow



Un **proceso** en Nextflow es la **unidad básica de ejecución** dentro de un pipeline. Representa un paso específico de análisis que se ejecuta de manera independiente, con entradas definidas, un script que realiza la tarea y salidas generadas.

Flujo de trabajo bionformático para la detección de variantes en *BRCA1* y *BRCA2*

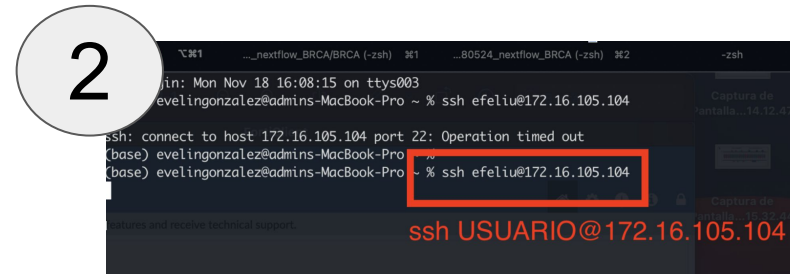
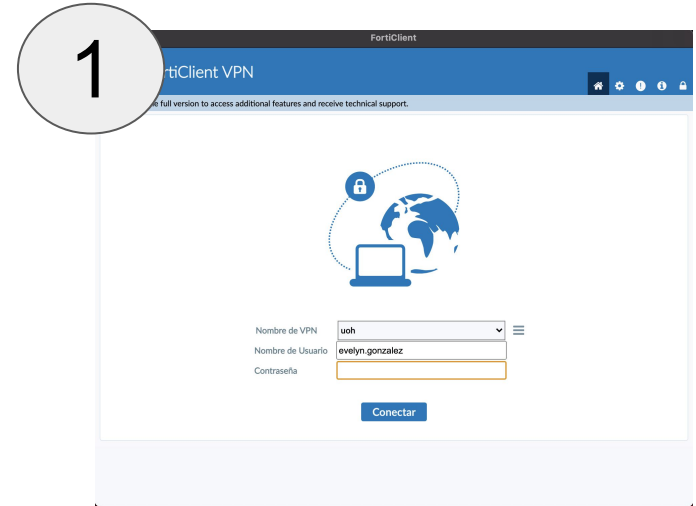
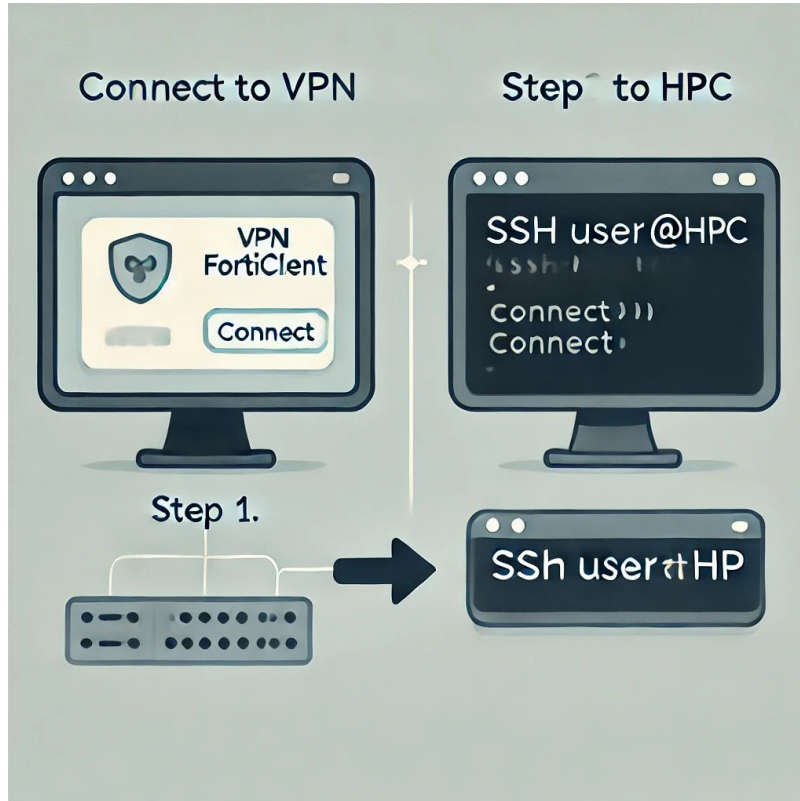


Ejecución de todo el flujo de trabajo de manera automática en un solo paso.

Interpretación de resultados y procesamiento de datos

The workflow, including the configurations and tools, is publicly available on the GitHub repository:
<https://github.com/digenoma-lab/BRCA>.

Primer Paso :Conexión a cluster HPC



Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

Ejecutar pipeline en Nextflow de BRCA1

```
(brca12) [efeliu@host04 BRCA]$ nextflow run main.nf -c nextflow.config -profile kutral -params-file ../params-brca.yml --csv ../readsHRR_1-4.csv -resume
e
curl: (6) Could not resolve host: www.nextflow.io
N E X T F L O W ~ version 23.10.1
Launching `main.nf` [cranky_euclid] DSL2 - revision: 4b9c1c4390
[a4/2c84e0] process > PRINT_VERSIONS [100%] 1 of 1, cached: 1 0
[-          ] process > FASTQC          [ 0%] 0 of 3
[-          ] process > BWAMEM          [ 0%] 0 of 3
[-          ] process > MERGE          -
[-          ] process > SAMTOOLS          -
[-          ] process > QUALIMAP          -
[-          ] process > B2C          -
[-          ] process > STRELKA_ONESAMPLE -
[-          ] process > STRELKA_POOL          -
[-          ] process > BCFTOOLS_FILTER -
[-          ] process > BF          -
[-          ] process > ANNOVAR_SP          -
[-          ] process > ANNOVAR_SS          -
```

Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

```

workflow {
  // merge bam files
  MERGEB(greads)

  //ONT alignment
  MINIMAP2_ALIGN(MERGEB.out.mbam,
                 ch_ref_fasta,
                 true,
                 'bai',
                 false,
                 false)

  //nanoplot - nanostat
  NANOPLOT(MINIMAP2_ALIGN.out.bam)

  //germline varcall to log reads, ONT
  CLAIR3(MINIMAP2_ALIGN.out.bam,
         MINIMAP2_ALIGN.out.index,
         ch_ref_fasta,
         ch_ref_index)

  //
  LONGPHASE(CLAIR3.out.vcf,
            MINIMAP2_ALIGN.out.bam,
            MINIMAP2_ALIGN.out.index,
            ch_ref_fasta,
            ch_ref_index)

  //
  METHYLARTIST_SEG(LONGPHASE.out.phasebam,
                  LONGPHASE.out.phaseindex,
                  params.bed,
                  ch_ref_fasta,
                  ch_ref_index)

  //DSS
  METHYLARTIST_WGMETH(LONGPHASE.out.phasebam,
                     LONGPHASE.out.phaseindex,
                     ch_ref_fasta,
                     ch_ref_index)
}

```

BODY:
Inputs
Workflow

nextflow run main.nf

```

#!/usr/bin/env nextflow
nextflow.enable.dsl = 2

//loading scripts from modules
include {MERGEB} from './modules/mergeb'
include {MINIMAP2_ALIGN} from './modules/minimap2'
include {CLAIR3} from './modules/clair3'
include {NANOPLOT} from './modules/nanoplot'
include {LONGPHASE} from './modules/longphase'
include {METHYLARTIST_SEG} from './modules/methylartist'
include {METHYLARTIST_WGMETH} from './modules/methylartist_wgmeth'

process PRINT_VERSIONS {
  publishDir "$params.outdir/software", mode: "copy"

  output:
  path("versions.txt")
  """
  echo "samtools: 1.19" > versions.txt
  echo "minimap2: 2.28-r1209" >> versions.txt
  echo "clair3: 1.0.10" >> versions.txt
  echo "nanoplot: 1.41.6" > versions.txt
  echo "longphase: 1.5.1" >> versions.txt
  echo "methylartist: 1.3.0" >> versions.txt
  """
}

```

HEAD:
MÓDULOS

Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

-c nextflow.config -profile kutral

```
process {  
  withName: 'PRINT_VERSIONS()' {  
    cpus = 1  
    memory = '1 GB'  
  }  
  withName: 'MERGEB' {  
    cpus = 10  
    memory = '20 GB'  
  }  
  withName: 'MINIMAP2_ALIGN' {  
    cpus = 20  
    memory = '40 GB'  
  }  
}
```

```
profiles {  
  kutral {  
    singularity.enabled = true  
    singularity.autoMounts = true  
    docker.enabled = false  
    podman.enabled = false  
    shifter.enabled = false  
    charliecloud.enabled = false  
    executor.queueSize = 8  
    process.executor = 'slurm'  
    process.queue = 'uohhm'  
  }  
}
```

```
params {  
  debug = false  
}
```

-c nextflow.config:

Especifica un archivo de configuración adicional, en este caso `nextflow.config`. Este archivo puede contener configuraciones como:

- Recursos (CPU, memoria).
- Parámetros globales del pipeline.
- Configuraciones específicas para perfiles y entornos.

Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

-params-file [ARCHIVO_PARAMETROS.YML]

```
120924_Nextflow_meth > ! methont-param-ont.yml
1  ref: /mnt/beegfs/labs/DiGenomaLab/Prunus_dulcis/references/rosaceae/GCA_021292205.2_OSU_Pdul_2.5_genomic.fa
2  minimap2: "-ax map-ont"
3  outdir: ont_result
4  aln_only: true
5  bed: /mnt/beegfs/home/efeliu/work2024/120924_Nextflow_meth/protein_coding_prunusDulcis.bed
6
```

-params-file:

Permite cargar un archivo de parámetros en formato YAML para configurar el pipeline. Este archivo contiene valores específicos para las variables que se utilizan en el pipeline, facilitando la personalización sin modificar el código.

Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

—csv [ARCHIVO_ENTRADA]

```
GNU nano 2.3.1 File: ../readsHRR_1-4.csv
Una linea por muestra (n=3)
sampleId,part,read1,read2
AL,0,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/AL_S9.R1.fastq.gz,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/AL_S9.R2.fastq.gz
DC,0,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/DC_S14.R1.fastq.gz,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/DC_S14.R2.fastq.gz
JC,0,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/JC_S13.R1.fastq.gz,/mnt/beegfs/labs/DiGenomaLab/HRR/reads/JC_S13.R2.fastq.gz
```

Archivo de entrada

Ejecutar pipeline en Nextflow

```
nextflow run main.nf \  
-c nextflow.config \  
-profile kutral \  
-params-file ../params-brca.yml \  
--csv ../readsHRR_1-4.csv \  
-resume
```

-resume

```
NEXTFLOW ~ version 24.04.4
```

```
Launching 'main.nf' [condescending_heisenberg] DSL2 - revision: 5080d20c78
```

```
[07/871c81] process > PRINT_VERSIONS [100%] 1 of 1, cached: 1 ✓
[07/871c81] process > PRINT_VERSIONS [100%] 1 of 1, cached: 1 ✓
[07/871c81] process > PRINT_VERSIONS [100%] 1 of 1, cached: 1 ✓
[1a/2f1261] process > MERGEB (CHI01-merge) [100%] 2 of 2, cached: 2 ✓
[65/942cb9] process > MINIMAP2_ALIGN (CHI01) [100%] 2 of 2, cached: 2 ✓
[fb/977a7a] process > NANOPLOT (B0872) [100%] 2 of 2, cached: 2 ✓
[0d/92f393] process > CLAIR3 (B0872) [100%] 2 of 2, cached: 2 ✓
[ac/39c3d7] process > LONGPHASE (CHI01) [100%] 2 of 2, cached: 2 ✓
[83/aae843] process > METHYLARTIST_WGMETH (B0872) [100%] 2 of 2, cached: 2 ✓
[B0872, [1, 2], [/home/adigenova/methylation-Chilean/bams/B0872.PAQ20471.fail.mod.bam, /home/adigenova/methylation-Chilean/bams/B0872.PAQ20471.pass.mod.bam]]
[CHI01, [1, 2, 3], [/home/adigenova/methylation-Chilean/bams/CHI01.PAQ20767.fail.mod.bam, /home/adigenova/methylation-Chilean/bams/CHI01.PAQ20767.pass.mod.bam, /home/adigenova/methylation-Chilean/bams/CHI01.PAQ20767.rep.pass_fail.mod.bam]]
```

-resume:

Indica que Nextflow debe **reutilizar los resultados previos** de ejecuciones anteriores. Esto es útil si la ejecución se interrumpe o si se desean realizar pequeños ajustes sin reanalizar todos los pasos. Nextflow verifica los pasos ya completados y reanuda desde donde se quedó, ahorrando tiempo y recursos.

Archivos de salida: BRCA pipeline

- Reporte de calidad: archivo HTML con parámetros de calidad. MultiQC.
- Archivos BAM (uno para cada muestra)
- Archivos VCF (uno para cada muestra)
- Anotación de variantes - ANNOVAR (uno para cada muestra)

QC, datos de calidad



A modular tool to aggregate results from bioinformatics analyses across many samples into a single report.

Report generated on 2024-11-11, 11:30 -03 based on data in: /mnt/beegfs/home/efeliu/work2024/080524_nextflow_BRCA/Runs_nextflow_BRCA/HHR_parte2

[Welcome! Not sure where to start?](#) [Watch a tutorial video](#) (6:06)

General Statistics

[Copy table](#) [Configure columns](#) [Scatter plot](#) [Violin plot](#) Showing 215/215 rows and 22/22 columns.

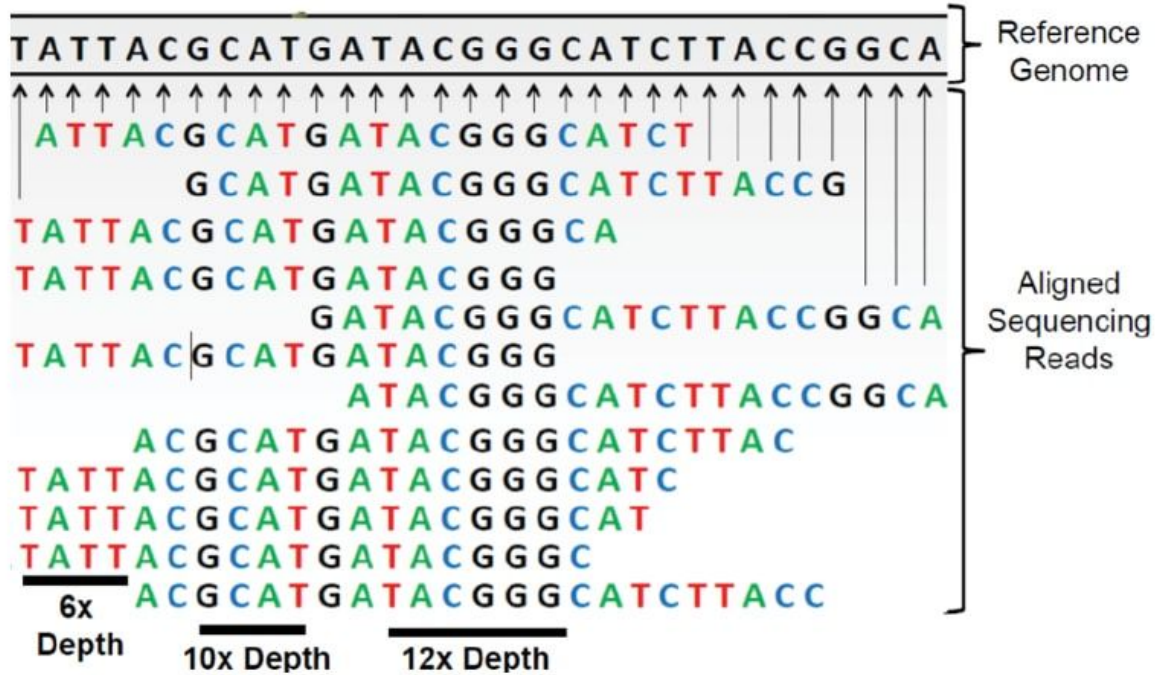
| Sample Name | % GC | Ins. size | ≥ 1X | ≥ 5X | ≥ 10X | ≥ 30X | ≥ 50X | Median cov | Mean cov | Error rate | % Aligned | M Aligned | M Total reads | Reads | Reads mapped |
|-------------|------|-----------|---------|---------|---------|---------|---------|------------|----------|------------|-----------|-----------|---------------|-------|--------------|
| 58.qualimap | 38 % | 105 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 554 X | | | | | | | |
| 46.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 549 X | | | | | | | |
| 81.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 524 X | | | | | | | |
| 60.qualimap | 38 % | 105 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 502 X | | | | | | | |
| 87.qualimap | 38 % | 102 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 483 X | | | | | | | |
| 54.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 482 X | | | | | | | |
| 66.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 477 X | | | | | | | |
| 50.qualimap | 38 % | 102 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 475 X | | | | | | | |
| 67.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 458 X | | | | | | | |
| 51.qualimap | 38 % | 103 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 456 X | | | | | | | |
| 47.qualimap | 38 % | 103 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 452 X | | | | | | | |
| 76.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 447 X | | | | | | | |
| 63.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 434 X | | | | | | | |
| 59.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 430 X | | | | | | | |
| 53.qualimap | 38 % | 104 | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 100.0 % | 426 X | | | | | | | |

FastQC
Qualimap
MultiQC

QualiMap

QualiMap is a platform-independent application to facilitate the quality control of alignment sequencing data and its derivatives like feature counts. DOI: 10.1093/bioinformatics/btv566; 10.1093/bioinformatics/bts503.

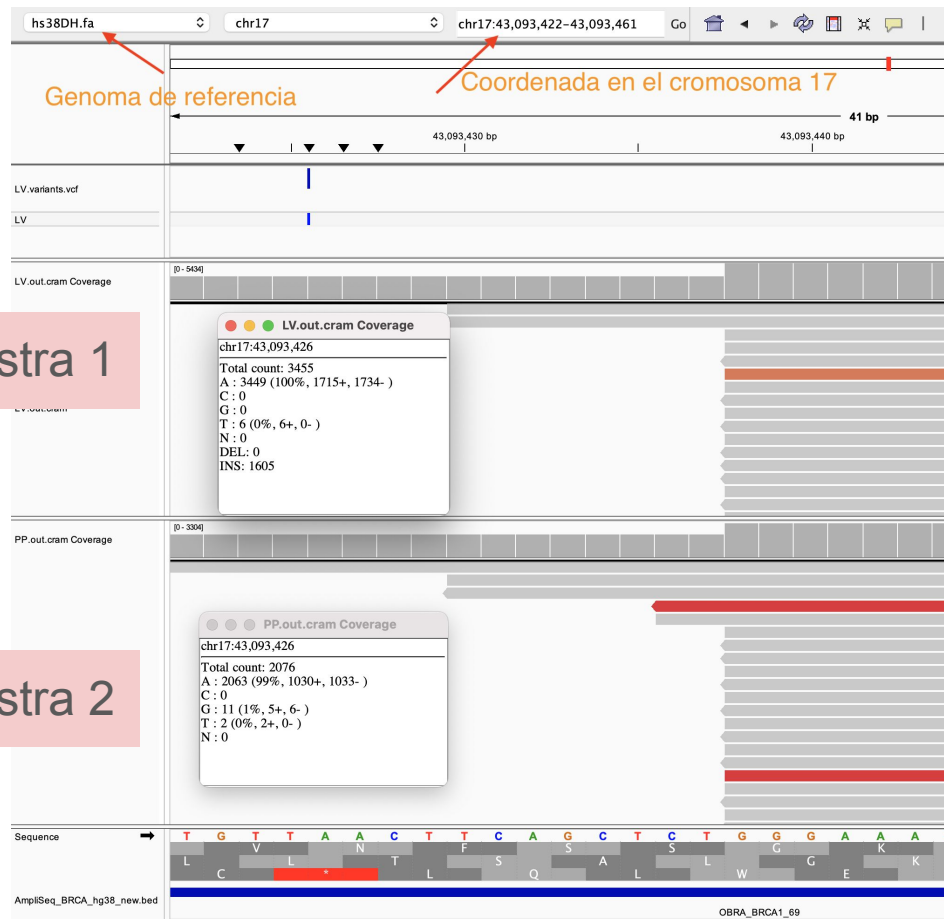
Archivos BAM: Lecturas alineadas al genoma



BAM: Formato binario para el almacenamiento de datos de secuenciación.

La extensión de archivo (.bam) contiene información sobre lecturas de secuencias después de haber sido estas **alineadas contra un genoma de referencia**.

Utilizado para almacenar datos de secuenciación masiva.



Ejemplo: Variante frameshift insertion p.L655Ffs*10 *BRCA1*

Inspección visual de las
lecturas alineadas al genoma
de referencia de dos
pacientes.

Variant Call Format: VCF

A VCF example

Header

```
##fileformat=VCFv4.1
##fileDate=20110413
##source=VCFtools
##reference=file:///refs/human_NCBI36.fasta
##contig=<ID=1,length=249250621,md5=1b22b98cdeb4a9304cb5d48026a85128,species="Homo Sapiens">
##contig=<ID=X,length=155270560,md5=7e0e2e580297b7764e31dbc80c2540dd,species="Homo Sapiens">
##INFO=<ID=AA,Number=1,Type=String,Description="Ancestral Allele">
##INFO=<ID=H2,Number=0,Type=Flag,Description="HapMap2 membership">
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
##FORMAT=<ID=DP,Number=1,Type=Integer,Description="Read Depth">
##ALT=<ID=DEL,Description="Deletion">
##INFO=<ID=SVTYPE,Number=1,Type=String,Description="Type of structural variant">
##INFO=<ID=END,Number=1,Type=Integer,Description="End position of the variant">
```

Body

| #CHROM | POS | ID | REF | ALT | QUAL | FILTER | INFO | FORMAT | SAMPLE1 | SAMPLE2 |
|--------|-----|------|-----|-------|------|--------|--------------------|----------|---------|-----------|
| 1 | 1 | . | ACG | A,AT | 40 | PASS | . | GT:DP | 1/1:13 | 2/2:29 |
| 1 | 2 | . | C | T,CT | . | PASS | H2;AA=T | GT | 0 1 | 2/2 |
| 1 | 5 | rs12 | A | G | 67 | PASS | . | GT:DP | 1 0:16 | 2/2:20 |
| X | 100 | . | T | | . | PASS | SVTYPE=DEL;END=299 | GT:GQ:DP | 1:12:. | 0/0:20:36 |

B SNP

| Alignment | VCF representation |
|-----------|--------------------|
| 1234 | POS REF ALT |
| ACGT | 2 C T |
| ATGT | |
| ^ | |

C Insertion

| | |
|-------|-------------|
| 12345 | POS REF ALT |
| AC-GT | 2 C CT |
| ACTGT | |
| ^ | |

D Deletion

| | |
|------|-------------|
| 1234 | POS REF ALT |
| ACGT | 1 ACG A |
| A--T | |
| ^^ | |

E Replacement

| | |
|------|-------------|
| 1234 | POS REF ALT |
| ACGT | 1 ACG AT |
| A-TT | |
| ^^ | |

| Chr | Start | End | Ref | Alt | Func | Gene | GeneDetail | ExonicFunc | AAChange |
|-------|----------|----------|-----|-----|--------|-------|------------|----------------------|-----------------------------------------------|
| chr13 | 32332343 | 32332343 | A | C | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon10:c.A865C:p.N289H |
| chr13 | 32332592 | 32332592 | A | C | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon10:c.A1114C:p.N372H |
| chr13 | 32332843 | 32332843 | A | G | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon10:c.A1365G:p.S455S |
| chr13 | 32336584 | 32336584 | T | C | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon11:c.T2229C:p.H743H |
| chr13 | 32336744 | 32336744 | A | C | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon11:c.A2389C:p.K797Q |
| chr13 | 32337326 | 32337326 | A | G | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon11:c.A2971G:p.N991D |
| chr13 | 32337751 | 32337751 | A | G | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon11:c.A3396G:p.K1132K |
| chr13 | 32338162 | 32338162 | T | C | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon11:c.T3807C:p.V1269V |
| chr13 | 32338918 | 32338918 | A | G | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon11:c.A4563G:p.L1521L |
| chr13 | 32340099 | 32340099 | C | T | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon11:c.C5744T:p.T1915M |
| chr13 | 32340678 | 32340678 | G | A | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon11:c.G6323A:p.R2108H |
| chr13 | 32340868 | 32340868 | G | C | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon11:c.G6513C:p.V2171V |
| chr13 | 32355095 | 32355095 | A | G | exonic | BRCA2 | | synonymous SNV | BRCA2:NM_000059.4:exon14:c.A7242G:p.S2414S |
| chr13 | 32355250 | 32355250 | T | C | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon14:c.T7397C:p.V2466A |
| chr13 | 32356461 | 32356461 | T | C | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon15:c.T7469C:p.I2490T |
| chr13 | 32379413 | 32379413 | G | A | exonic | BRCA2 | | nonsynonymous SNV | BRCA2:NM_000059.4:exon22:c.G8851A:p.A2951T |
| chr17 | 43071077 | 43071077 | T | C | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon14:c.A4696G:p.S1566G, E |
| chr17 | 43082453 | 43082453 | A | G | exonic | BRCA1 | | synonymous SNV | BRCA1:NM_007297.4:exon11:c.T4167C:p.S1389S, E |
| chr17 | 43091983 | 43091983 | T | C | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon9:c.A3407G:p.K1136R, BF |
| chr17 | 43092412 | 43092412 | C | T | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon9:c.G2978A:p.S993N, BR |
| chr17 | 43092418 | 43092418 | T | C | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon9:c.A2972G:p.E991G, BR |
| chr17 | 43092919 | 43092919 | G | A | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon9:c.C2471T:p.P824L, BR |
| chr17 | 43093220 | 43093220 | A | G | exonic | BRCA1 | | synonymous SNV | BRCA1:NM_007297.4:exon9:c.T2170C:p.L724L, BR |
| chr17 | 43093425 | 43093425 | - | A | exonic | BRCA1 | | frameshift insertion | BRCA1:NM_007297.4:exon9:c.1964dupT:p.L655Ffs |
| chr17 | 43093449 | 43093449 | G | A | exonic | BRCA1 | | synonymous SNV | BRCA1:NM_007297.4:exon9:c.C1941T:p.S647S, BR |
| chr17 | 43093454 | 43093454 | C | T | exonic | BRCA1 | | nonsynonymous SNV | BRCA1:NM_007297.4:exon9:c.G1936A:p.D646N, BR |

Sort by:

Buscar variantes "Pathogenic" en CLINVAR

Filter by:

| | | | | | | |
|---------------|----------------------|----------------------|----------------------|-----------------------|----------------------|----------------------|
| CLNALLEID: | <input type="text"/> | CLNDN: | <input type="text"/> | CLNSIG: | <input type="text"/> | <input type="text"/> |
| SIFT_pred: | <input type="text"/> | SIFT4G_pred: | <input type="text"/> | Polyphen2_HVAR_score: | <input type="text"/> | <input type="text"/> |
| LRT_pred: | <input type="text"/> | MutationTaster_pred: | <input type="text"/> | FATHMM_score: | <input type="text"/> | <input type="text"/> |
| PROVEAN_pred: | <input type="text"/> | MetaSVM_score: | <input type="text"/> | MetaLR_score: | <input type="text"/> | <input type="text"/> |
| MetaLR_pred: | <input type="text"/> | Reliability_index: | <input type="text"/> | | | |

Chr:

Start:

End:

Gene:

CLNALLEID:

VCF anotados con ANNOVAR

Filtrado de Variantes para la Identificación de:

- Variantes Patogénicas
- VUS (Variantes de Significado Incierto)
- Variantes Novel
- Variantes Benignas

Repositorio del pipeline BRCA en GitHub



The screenshot shows the GitHub interface for the 'BRCA' repository by 'digenoma-lab'. The repository is public and has 1 branch, 1 tag, 0 forks, and 0 stars. The main branch is 'main'. The repository contains a file tree with the following files and their last commit dates:

| File | Last Commit |
|-----------------|------------------------------------------------------|
| modules | order modules (4 months ago) |
| reads | working in strelka pool (last year) |
| scripts | adding commands to build nextflow script (last year) |
| Dockerfile | adding containers (last year) |
| LICENSE | Initial commit (2 years ago) |
| README.md | Update README.md (last month) |
| main.nf | first update (4 months ago) |
| micromamba.yml | first update (4 months ago) |
| nextflow.config | first update (4 months ago) |

The repository also has a README file and a MIT license. The README is titled 'BRCA' and describes the pipeline as 'A Nextflow pipeline for processing target NGS BRCA data' and 'Nextflow Pipeline for BRCA Variant Calling Using Amplicon Illumina Data'. The repository is also listed in the 'Releases' section with a 'BRCA pipeline' release on Sep 30, marked as 'Latest'. The 'Packages' section shows no published packages. The 'Contributors' section lists three contributors: adigenova, ManuelM11, and another user.