hmnfusion

Release 1.2.12

guillaume-gricourt

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1.1 Install

1.1.1 Conda

```
$ conda install -c bioconda hmnfusion
```

1.1.2 Pip

```
$ wget https://github.com/guillaume-gricourt/HmnFusion/releases/download/1.2.12/pip.zip
$ unzip pip.zip
$ pip install hmnfusion-1.2.12-py3-none-any.whl
$ rm pip.zip hmnfusion-1.2.12-py3-none-any.whl hmnfusion-1.2.12.tar.gz
```

1.1.3 Docker

Pull `hmnfusion`. It contains Genefuse, Lumpy and HmnFusion.

```
$ docker pull ghcr.io/guillaume-gricourt/hmnfusion:1.2.12
```

Pull `hmmnfusion-align` to create BAM files

```
$ docker pull ggricourt/hmnfusion-align:1.2.12
```

The reference files used to build BAM files refer to the DOI 10.5281/zenodo.6619597

> **Warning:** The size of the image is almost 15Go

1.1.4 Dependencies

Genefuse and Lumpy are available in the `docker` image. But they are not installed with `conda` and `pip`. You can install them in the current environment with this:

```
$ hmnfusion install-software
```

1.2 Usage

HmnFusion is designed to analyze some panel of genes sequenced with the aCAP-Seq library and performs with a high sensitivity and specificity 10.1016/j.jmoldx.2022.07.004

1.2.1 Steps

1. BAM files are analyzed by Lumpy and Genefuse

2. The results are aggregated by HmnFusion

3. Now you can calculate the fusion frequency and you can define the MMEJ sequence

These steps involved one or more commands. A workflow is available to grouped these commands into one command.

1.2.2 How to start with HmnFusion

It depends what you have:

- If you have FASTQ files, you need first to create BAM files, please go to *Create BAM files*

- If you have FASTQ, BAM files, please go to *Fusion callers*

- If you have FASTQ, BAM files, HTML of JSON from Genefuse and VCF from Lumpy, please go to *Fusion frequency* or *MMEJ sequences*

Questions about extra files: hg19 reference, BED files ? please read the *FAQ*.

1.2.3 Create BAM files

It's only available through `docker`

```
$ docker run -it \
    --rm \
    hmnfusion-align:latest \
    workflow-align \
    --input-forward-fastq <FASTQ forward, file> \
    --input-reverse-fastq <FASTQ reverse, file> \
    --output-directory <Output directory> \
    --threads 4
```

1.2.4 Fusion callers

It will run Genefuse, Lumpy and HmnFusion, to detect and quantify fusions.

```
$ hmnfusion workflow-fusion \
    # Sample
    --name <Name of sample> \
    --input-forward-fastq <Fastq file forward> \
    --input-reverse-fastq <Fastq file reverse> \
    --input-sample-bam <Bam file> \
    # Bed
    --input-genefuse-bed <Genefuse bed file> \
    --input-lumpy-bed <Lumpy bed file> \
    --input-hmnfusion-bed <HmnFusion bed file> \
    # Reference
    --input-reference-fasta <Reference fasta file (hg19)> \
    # Output
    --output-hmnfusion-vcf <Vcf file output> \
    --output-genefuse-html <Genefuse html file output> \
    --output-lumpy-vcf <Lumpy vcf file output> \
    --threads [1-6]
```

1.2.5 Fusion frequency

It will extract fusions from Genefuse and Lumpy to quantify them.

```
$ hmnfusion workflow-fusion \
    # Sample
    --input-lumpy-vcf <Lumpy Vcf file> \
    --input-genefuse-json <Genefuse, json file> OR --input-genefuse-html <Genefuse, html␣
→file> \
    --input-sample-bam <Bam file> \
    --name <Name of sample> \
    # Bed
    --input-hmnfusion-bed <HmnFusion bed file> \
    # Output
    --output-hmnfusion-vcf <Vcf file output>
```

1.2.6 MMEJ sequences

Define fusions of interest.

```
$ hmnfusion extractfusion \
    # Sample
    --input-genefuse-json <Genefuse, json file> \
    --input-genefuse-html <Genefuse, html file> \
    --input-lumpy-vcf <Lumpy vcf file> \
    # Bed
    --input-hmnfusion-bed <Bed file> \
    # Output
    --output-hmnfusion-json <Json file output>
```

Extract MMEJ sequences.

```
$ hmnfusion mmej-fusion \
    # Sample
    --input-hmnfusion-json <HmnFusion, json file> \
    --input-sample-bam <Bam file> \
    --name <Name of sample> \
    # References
    --input-reference-fasta <Reference, fasta file> \
    # Output
    --output-hmnfusion-xlsx <Excel file output> \
    --output-hmnfusion-json <Json file output>
```

1.3 FAQ

1.3.1 References

HmnFusion uses hg19 human genome.

1.3.2 Bed files

3 BED files are used by default or you can provide custom BED files:

- Lumpy. The original BED has been slightly modified (`download`)
- Genefuse. The original BED has been slightly modified (`download`)
- HmnFusion. Contains the location of the primers used in 10.1016/j.jmoldx.2022.07.004 (`download`)

If you want to use your own design, and you have some questions, feel free to open an issue

1.4 Code of Conduct

- Be friendly and patient
- Be respectful
- Be welcoming
- Be considerate
- Focus on what's best for the community
- Gracefully accept constructive criticism
- Show courtesy and respect towards other community members
- Be respectful of different viewpoints and experiences
- Use welcoming and inclusive language

1.4.1 Reporting an issue

If you see a violation of the code of conduct, please report it to [@guillaume-gricourt](https://github.com/guillaume-gricourt) To protect volunteers from abuse and burnout, I reserve the right to reject any report I believe to have been made with the deliberate intention of deceiving or misleading myself.

1.4.2 Enforcement

Upon receiving a report, the Maintainer core team will meet privately to discuss the matter and determine the best course of action.

Possible responses may include:

- taking no further action (if we determine no violation occurred)

- a private reprimand from the core members to the individual(s) involved. In this case, a core member will deliver that reprimand via email, copying the rest of the core.

- a public reprimand. In this case a core member will deliver the reprimand via the same channel as the violation occurred (e.g., via comment on PR or issue)

- an imposed vacation (e.g., asking the individual to refrain from participating for a week). A core member will deliver a request for the individual to take such a "vacation" voluntarily, but if they don't disagree then a temporary ban may be imposed.

- a permanent or temporary ban from the repository

Once a resolution is agreed upon, but before it is enacted, I will contact the original reporter and discuss the resolution and will note any feedback for the record, but is not obligated to act on such feedback.

I will never publicly discuss the issue; any public statements will be made by myself.

This policy was modified from [django project](#)

1.5 Contributing

Contributions are welcome, and they are greatly appreciated! Every little bit helps, and credit will always be given.

1.5.1 Ways you can help

Report Bugs

Report bugs at https://github.com/guillaume-gricourt/HmnFusion/issues.

If you are reporting a bug, please include:

- Your operating system name and version, versions of HmnFusion

- Any details about your local setup that might be helpful in troubleshooting.

- Detailed steps to reproduce the bug.

Fix Bugs

Look through the GitHub issues for bugs. Anything tagged with "bug" is open to whoever wants to implement it.

Implement Features

Look through the GitHub issues for features. Anything tagged with "enhancement" is open to whoever wants to implement it.

Write Documentation

Same as Implement Features

Submit Feedback

The best way to send feedback is to file an issue at https://github.com/guillaume-gricourt/HmnFusion/issues.

If you are proposing a feature:

- Explain in detail how it would work.
- Keep the scope as narrow as possible, to make it easier to implement.
- This will hopefully become a community-driven project and contributions are welcome :)

1.5.2 Pull Request Guidelines

Before you submit a pull request, check that it meets these guidelines:

1. If the pull request adds functionality, the docs should be updated. Put your new functionality into a function with a docstring.
2. Passing `pytest`

   ```
   $ python -m pytest
   ```

3. Passing super-linter

This policy was modified from planemo project

1.6 Changelog

1.6.1 1.2.11 (2023-04-19)

Full Changelog

1.6.2 1.2.10 (2023-04-19)

Full Changelog

1.6.3 1.2.9 (2023-04-19)

Full Changelog

1.6.4 1.2.8 (2023-04-19)

Full Changelog

Merged pull requests:

- Read the docs #9 (guillaume-gricourt)

1.6.5 1.2.7 (2023-04-17)

Full Changelog

Merged pull requests:

- style(github): super-linter #8 (guillaume-gricourt)
- chore(deps): bump github/super-linter from 4 to 5 #7 (dependabot[bot])
- chore(deps): bump schneegans/dynamic-badges-action from 1.1.0 to 1.6.0 #6 (dependabot[bot])
- chore(deps): bump actions/checkout from 2 to 3 #5 (dependabot[bot])
- chore(deps): bump actions/download-artifact from 2 to 3 #4 (dependabot[bot])
- chore(deps): bump actions/upload-artifact from 2 to 3 #3 (dependabot[bot])
- chore(deps): bump mathieudutour/github-tag-action from 6.0 to 6.1 #2 (dependabot[bot])
- chore(deps): bump actions/setup-python from 2 to 4 #1 (dependabot[bot])

1.6.6 1.2.6 (2022-09-16)

Full Changelog

1.6.7 1.2.4 (2022-09-16)

Full Changelog

1.6.8 1.2.5 (2022-09-16)

Full Changelog

1.6.9 1.2.3 (2022-06-15)

Full Changelog

1.6.10 1.2.2 (2022-06-14)

Full Changelog

1.6.11 1.2.1 (2022-06-14)

Full Changelog

1.6.12 1.2.0 (2022-06-09)

Full Changelog

1.6.13 1.1.0 (2022-05-06)

Full Changelog

1.6.14 1.0.3 (2022-04-13)

Full Changelog

1.6.15 1.0.2 (2022-04-13)

Full Changelog

1.6.16 1.0.1 (2022-03-21)

Full Changelog

1.6.17 1.0.0 (2022-03-16)

Full Changelog

1.6.18 0.8.0 (2022-02-14)

Full Changelog

1.6.19 0.7.3 (2021-08-08)

Full Changelog

1.6.20 0.7.2 (2021-08-08)

Full Changelog

1.6.21 0.7.1 (2021-06-16)

Full Changelog

1.6.22 0.7.0 (2021-06-14)

Full Changelog

1.6.23 0.6.0 (2021-03-08)

Full Changelog

1.6.24 0.5.0 (2020-12-10)

Full Changelog

** This Changelog was automatically generated by*github_changelog_generator