1 Purpose

Generate a .bbl file from a list of citations, so that references can be included within a document. Simply copy the reference list from the .bbl file that Bib-TeX generates and paste it into the main .tex file (and delete the associated \bibliography and \bibliographystyle commands).

2 Example

Once bibtex is run, the .bbl file will contain the following:

```
\begin{thebibliography}{1}
\bibitem{snpsift}
P.~Cingolani, V.~M. Patel, M.~Coon, T.~Nguyen, S.~J. Land, D.~M. Ruden, and
  X.~Lu.
\newblock {{U}sing {D}rosophila melanogaster as a {M}odel for {G}enotoxic
  {C}hemical {M}utational {S}tudies with a {N}ew {P}rogram, {S}np{S}ift}.
\newblock {\emptysetem Front Genet}, 3:35, 2012.
\end{thebibliography}
```

Simply copy and paste this code into the main document.

3 Citing bioinformatic tools

- 1. SnpSift[6]
- 2. BWA-MEM[10]
- 3. FastQC[2]
- 4. SAMtools and BCFtools[11]
- 5. Trimmomatic[4]
- 6. bam.iobio[13]
- 7. Bpipe[19]
- 8. BEDTools[18]
- 9. PLINK2[5]

We sequenced 10 samples on 10 lanes on an Illumina HiSeq 2000, aligned the resulting reads to the hg19 reference genome with BWA-MEM[10], applied GATK[12] base quality score recalibration, indel realignment, duplicate removal, and performed SNP and INDEL discovery and genotyping across all 10 samples simultaneously using standard hard filtering parameters or variant quality score recalibration according to GATK Best Practices recommendations [8, 21].

4 Other citations

- 1. GWAS on Australian aboriginal population [1]
- 2. The Mardu aborigines: living the dream in Australia's desert[20]
- 3. Cleared out: first contact in the Western Desert[7]
- 4. Exome sequencing as a tool for Mendelian disease gene discovery[3]
- 5. Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome[15]
- 6. Exome sequencing identifies the cause of a Mendelian disorder[16]
- 7. Mimura et al.[14]
- 8. Inoue et al.[9]
- 9. Papantonis et al.[17]

References

- [1] D. Anderson, H. J. Cordell, M. Fakiola, R. W. Francis, G. Syn, E. S. Scaman, E. Davis, S. J. Miles, T. McLeay, S. E. Jamieson, and J. M. Blackwell. First genome-wide association study in an Australian aboriginal population provides insights into genetic risk factors for body mass index and type 2 diabetes. *PLoS ONE*, 10(3):e0119333, 2015.
- [2] S. Andrews. FastQC A Quality Control tool for High Throughput Sequence Data.
- [3] M. J. Bamshad, S. B. Ng, A. W. Bigham, H. K. Tabor, M. J. Emond, D. A. Nickerson, and J. Shendure. Exome sequencing as a tool for Mendelian disease gene discovery. *Nat. Rev. Genet.*, 12(11):745–755, Nov 2011.
- [4] A. M. Bolger, M. Lohse, and B. Usadel. Trimmomatic: a flexible trimmer for Illumina sequence data. *Bioinformatics*, 30(15):2114–2120, Aug 2014.
- [5] C. C. Chang, C. C. Chow, L. C. Tellier, S. Vattikuti, S. M. Purcell, and J. J. Lee. Second-generation PLINK: rising to the challenge of larger and richer datasets. *Gigascience*, 4:7, 2015.
- [6] P. Cingolani, V. M. Patel, M. Coon, T. Nguyen, S. J. Land, D. M. Ruden, and X. Lu. Using Drosophila melanogaster as a Model for Genotoxic Chemical Mutational Studies with a New Program, SnpSift. Front Genet, 3:35, 2012.
- [7] Sue Davenport. Cleared out: first contact in the Western Desert. Aboriginal Studies Press, Canberra, 2005.
- [8] M. A. DePristo, E. Banks, R. Poplin, K. V. Garimella, J. R. Maguire, C. Hartl, A. A. Philippakis, G. del Angel, M. A. Rivas, M. Hanna, A. McKenna, T. J. Fennell, A. M. Kernytsky, A. Y. Sivachenko, K. Cibulskis, S. B. Gabriel, D. Altshuler, and M. J. Daly. A framework for variation discovery and genotyping using next-generation DNA sequencing data. *Nat. Genet.*, 43(5):491–498, May 2011.

- [9] T. Inoue, T. Kohro, T. Tanaka, Y. Kanki, G. Li, H. M. Poh, I. Mimura, M. Kobayashi, A. Taguchi, T. Maejima, J. Suehiro, A. Sugiyama, K. Kaneki, H. Aruga, S. Dong, J. F. Stevens, S. Yamamoto, S. Tsutsumi, T. Fujita, X. Ruan, H. Aburatani, M. Nangaku, Y. Ruan, T. Kodama, and Y. Wada. Cross-enhancement of ANGPTL4 transcription by HIF1 alpha and PPAR beta/delta is the result of the conformational proximity of two response elements. Genome Biol., 15(4):R63, 2014.
- [10] H. Li. Aligning sequence reads, clone sequences and assembly contigs with BWA-MEM. ArXiv e-prints, March 2013.
- [11] H. Li, B. Handsaker, A. Wysoker, T. Fennell, J. Ruan, N. Homer, G. Marth, G. Abecasis, and R. Durbin. The Sequence Alignment/Map format and SAMtools. *Bioinformatics*, 25(16):2078–2079, Aug 2009.
- [12] A. McKenna, M. Hanna, E. Banks, A. Sivachenko, K. Cibulskis, A. Kernytsky, K. Garimella, D. Altshuler, S. Gabriel, M. Daly, and M. A. DePristo. The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. *Genome Res.*, 20(9):1297–1303, Sep 2010.
- [13] C. A. Miller, Y. Qiao, T. DiSera, B. D'Astous, and G. T. Marth. bam.iobio: a web-based, real-time, sequence alignment file inspector. *Nat. Methods*, 11(12):1189, Dec 2014.
- [14] I. Mimura, M. Nangaku, Y. Kanki, S. Tsutsumi, T. Inoue, T. Kohro, S. Yamamoto, T. Fujita, T. Shimamura, J. Suehiro, A. Taguchi, M. Kobayashi, K. Tanimura, T. Inagaki, T. Tanaka, T. Hamakubo, J. Sakai, H. Aburatani, T. Kodama, and Y. Wada. Dynamic change of chromatin conformation in response to hypoxia enhances the expression of GLUT3 (SLC2A3) by cooperative interaction of hypoxia-inducible factor 1 and KDM3A. *Mol. Cell. Biol.*, 32(15):3018–3032, Aug 2012.
- [15] S. B. Ng, A. W. Bigham, K. J. Buckingham, M. C. Hannibal, M. J. McMillin, H. I. Gildersleeve, A. E. Beck, H. K. Tabor, G. M. Cooper, H. C. Mefford, C. Lee, E. H. Turner, J. D. Smith, M. J. Rieder, K. Yoshiura, N. Matsumoto, T. Ohta, N. Niikawa, D. A. Nickerson, M. J. Bamshad, and J. Shendure. Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nat. Genet., 42(9):790–793, Sep 2010.
- [16] S. B. Ng, K. J. Buckingham, C. Lee, A. W. Bigham, H. K. Tabor, K. M. Dent, C. D. Huff, P. T. Shannon, E. W. Jabs, D. A. Nickerson, J. Shendure, and M. J. Bamshad. Exome sequencing identifies the cause of a mendelian disorder. *Nat. Genet.*, 42(1):30–35, Jan 2010.
- [17] A. Papantonis, T. Kohro, S. Baboo, J. D. Larkin, B. Deng, P. Short, S. Tsutsumi, S. Taylor, Y. Kanki, M. Kobayashi, G. Li, H. M. Poh, X. Ruan, H. Aburatani, Y. Ruan, T. Kodama, Y. Wada, and P. R. Cook. TNFα signals through specialized factories where responsive coding and miRNA genes are transcribed. *EMBO J.*, 31(23):4404–4414, Nov 2012.
- [18] A. R. Quinlan and I. M. Hall. BEDTools: a flexible suite of utilities for comparing genomic features. *Bioinformatics*, 26(6):841–842, Mar 2010.

- [19] S. P. Sadedin, B. Pope, and A. Oshlack. Bpipe: a tool for running and managing bioinformatics pipelines. *Bioinformatics*, 28(11):1525–1526, Jun 2012.
- [20] Robert Tonkinson. The Mardu aborigines: living the dream in Australia's desert. Wadsworth/Thomson Learning, Belmont, CA, 2002.
- [21] G. A. Van der Auwera, M. O. Carneiro, C. Hartl, R. Poplin, G. Del Angel, A. Levy-Moonshine, T. Jordan, K. Shakir, D. Roazen, J. Thibault, E. Banks, K. V. Garimella, D. Altshuler, S. Gabriel, and M. A. DePristo. From FastQ data to high confidence variant calls: the Genome Analysis Toolkit best practices pipeline. Curr Protoc Bioinformatics, 11(1110):1–11, Oct 2013.