

Haematologica
HAEMATOL/2019/230821
Version 3

RNAmut: robust identification of somatic mutations in acute myeloid
leukemia using RNA-seq

Muxin Gu, Maximilian Zwiebel, Swee Hoe Ong, Nick Boughton, Josep Nomdedeu, Faisal Basheer, Yasuhito Nannya, Pedro M. Quiros, Seishi Ogawa, Mario Cazzola, Roland Rad, Adam P. Butler, MS Vijayabaskar, and George Vassiliou

Disclosures: This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking under grant agreement No 116026. This Joint Undertaking receives support from the European Union's Horizon 2020 research and innovation and EFPIA. MG is funded by Horizon 2020 (No. 116026) and Cancer Research UK (C22324/A23015). GSV is funded by a Cancer Research UK Senior Cancer Research Fellowship (C22324/A23015) and work in his laboratory is also funded by the Wellcome Trust, European Research Council, Kay Kendall Leukaemia Fund, Bloodwise, The Leukemia Lymphoma Society and Rising Tide Foundation for Clinical Cancer Research. MSV is funded by the Wellcome Trust (WT098051). G.S.V. is a consultant for Kymab and Oxstem, and receives an educational grant from Celgene. The remaining authors declare no competing financial interests.

Contributions: G.S.V. and MS.V. conceived and designed the study. M.G. wrote and implemented the software, conducted the bulk of bioinformatics analyses and wrote the manuscript with input from all authors. M.Z. performed bioinformatics analyses on variant calling. J.N., R.R. and F.B. provided clinical and diagnostic advice to aid the software development. S.O., M.C. and Y.N. provided the RNA-seq data and genotype information of the MDS cohort. S.H.O., N.B. and A.P.B. developed the web server for customizing gene panels. P.M.Q. and MS.V helped with bioinformatics work and generate the figures.