Selected Publications Citing the Semison Software

1. The Gossypium stocksii genome as a novel resource for cotton improvement
Yuqing Hou, Lanjie Zhao, Wenqiang Han, Xiaoyang Kong, Fuguang Li, Hillel Fogelman, Yaniv Sapir, Jonathan F Wendel, Jinhong Yuan, Ji Liu
Iowa State University, Huazhong Agricultural University, Municipal State University, Chinese Academy of Agricultural Sciences, Tel Aviv University, University of Florida
Nature Genetics (2023) 55:1, 165–175 · doi.org/10.1038/s41588-022-01237-2 · PMID: 36539618 · PMCID: PMC9839892

2. A novel variant of CDH5 causes a stroke-neurodevelopmental disorder with inherited spastin
Shenglan Wang, Wenzhen Zhu, Jianhua Qiu, Yanling Wu, Zhifang Wu
Children's Hospital, Capital Institute of Pediatrics, Cipher Gene LLC
Annals of Clinical and Translational Neurology (2020) 7:11, 2241–2246 · doi.org/10.1002/acn3.51208 · PMID: 33058519 · PMCID: PMC7664268

3. Systematic characterization of multiomics altering protein degradation in human cancers
Yize Li, Shankara Anand, Kelly V. Ruggles, Bo Wen, Ana I. Robles, Henry Rodriguez, Zhidong Tu, Chen Zhao, Jun Qu, Jun Zhu, Xiaoling Zhang, Xiaohui Zhan, Shuangge Ma, Bing Zhang
Duke-Fudan Cancer Institute, Harvard T.H. Chan School of Public Health, Harvard Hopkins Medical Institute, Harvard Medical School, Yale School of Medicine, Fudan University, National Cancer Institute
Nature Communications (2022) 13:1, 5133 · doi.org/10.1038/s41467-022-32577-5 · PMID: 36050315 · PMCID: PMC9436941

4. Bulbilis loss of function in MMP1 is a cause of recessive skeletal cardiomyopathy
Yuan Yuan, Yuanyuan Zhang, Yuanyuan Zhang, Yuanyuan Zhang, Yuanyuan Zhang, Ejun Ji, Xiaobin Sun, Yuanyuan Zhang, Jianhua Qiu, Jianhua Sun, Yanling Wu, Zhifang Wu, Jianhua Qiu, Yanling Wu, Zhifang Wu, Jianhua Qiu, Yanling Wu, Zhifang Wu, Jianhua Qiu

deafness – a case report for dual diagnosis
Yan Zhang, Yi Zhang, Victor Wei Zhang, Chunyi Zhang, Hongke Ding and Aihua Yin
Guangdong Women and Children Hospital, Euler Genomics Co. Ltd, and Baylor College of Medicine.
MC Pediatrics volume 19, 3842(19). doi:10.1186/s12887-019-1733-y

88. Single nucleotide mapping of trait space reveals Pareto fronts that constrain adaptation
Yuping Li, Dmitri A. Petrov and Gavin Sherlock.
Stanford University.
Nature Ecology & Evolution 3, 1539–1551(2019). doi:10.1038/s41559-019-0993-0

89. Whole Genome Sequencing of 508 Patients Identified Key Molecular Features Associated with Poor Prognosis in Esophageal Squamous Cell Carcinoma
Cui, Yongqing and Chen, Hongyan and Xi, Ruibin and Cui, Heyang and Zhao, Yahui and Yan, Ting and Xu, Enwei and Liu, Xiaomei and Huang, Furong and Kong, Pengzhou and Li, Yang and Zhu, Xiaolin and Wang, Jiawei and Zhu, Wenjie and Wang, Jie and Ma, Yanchun and Zhou, Yong and Guo, Shiping and Zhang, Ling and Liu, Yiqian and Wang, Bin and Yu, Yanfeng and Sun, Ruifang and Yu, Xiao and Zhai, Yuanfang and Wang, Fang and Yang, Jian and Yang, Bin and Cheng, Caixia and Liu, Jing and Song, Bin and Liu Li, Hongyi and Wang, Yi and Zhang, Yingchun and Cheng, Xiaobing and Li, Yanhong and Liu, Zhihua and Zhao, Qinlong
Shanxi Medical University, Chinese Academy of Medical Sciences, Peking University, Shanxi Cancer Hospital, and WuXi NextCODE
Cancer Cell (2019). doi:10.2139/ssrn.3447322

90. CRISPR-Cas9 generated Pompe knock-in murine model exhibits early-onset cardiomyopathy and impaired autophagic clearance
Jeffrey Y. Huang, Shih-Hsin Kan, Anthony D. Rangel, Yunghang Chan, Jeremy Davis-Turak, Nancy D. Dalton, Jon Neumann, Raymond Y. Wang
CHOC Children's Research Institute, University of California San Diego, Orihamp Bioinformatics, Inc., and University of California Irvine
bioRxiv 785130 (2019). doi:10.1101/785130

91. Genetics of Unexplained Sudden Cardiac Death in Adult Caucasian and African American Individuals Living in the State of Maryland
Liang Guo, Sho Torii, Raquel Fernandez, Ryan E. Braunstein, Ka Hyun Paek, Daniela T. Fuller, Christina M Mayhew, Roya Zarpak, Hiroyuki Jimouchi, Atsushi Sakamoto, Yu Sato, Hiroyoshi Mori, Matthew D Kutyna, Parker J Lee, Leah M Weinstein, Carlos J Collado-Rivera, Neel V Gadhoke, Bake B Ali, Dheeraj R Attmakuri, Roma Dhingra, Emma LB Finn, Mack W Bell, Anne Cornelissen, Salome H Kuntz, Joohyung Park, Robert Kutyn, Libin Wang, Susie N Hong, Anuj Gupta, Frank D Kolodgie, Maria E Romero, Braxton D Mitchell, Dipti Surve, David R Fowler, Charles C Hong, Renu Virmani, Aloke V Finn
CVPath Institute, University of Maryland School of Medicine, and Office of the Chief Medical Examiner of Baltimore, Maryland
medRxiv 19007744 (2019). doi:10.1101/19007744

92. Comparative evolutionary analysis and prediction of deleterious mutation patterns between sorghum and maize
Roberto Lozano, Elodie Gazave, Jonathan PR dos Santos, Markus Stetter, Ravi Valluru, Nancy Bandillo, Samuel B. Fernandes, Patrick J. Brown, Nadia Shakoor, Todd C. Mockler, Jeffrey Ross-Ibarra, Edward S. Buckler, Michael A. Gore
Cornell University, University of Cologne, University of California Davis, University of Illinois at Urbana-Champaign, and Donald Danforth Plant Science Center
bioRxiv 777623 (2019). doi:10.1101/777623

93. Measuring Tumor Mutational Burden Using Whole-Exome Sequencing
Tomas Vilmas
National Institute of Health
Biomarkers for Immunotherapy of Cancer: "Methods in Molecular Biology", Vol 2055(2020). doi:10.1007/978-1-4939-9773-2_3

94. Development and analytical validation of a next-generation sequencing based microsatellite instability (MSI) assay
Sarabjot Pabla, Jonathan Andreas, Felicia L. Lenzo, Blake Burgher, Jacob Hagen, Vincent Giamo, Mary K. Nesline, Yirong Wang, Mark Gardner, Jeffrey M. Conroy, Antonios Papanicolau-Sengos, Carl Morrison, and Sean T. Glenn
Omnibsq Inc., and Roswell Park Comprehensive Cancer Center
Oncotarget 10503:1181 (2019). doi:10.18632/oncotarget.27142

95. Sentieon DNASeq variant calling workflow demonstrates strong computational performance and accuracy
Katherine I. Kendig, Saurabh Baheti, Matthew A. Bockol, Travis M. Drucker, Steven N. Hart, Jacob R. Heldenbrand, Mikel Hernaez, Matthew E. Hudson, Michael T. Kalmbach, Eric W. Klee, Nathan R. Mattson, Christian A. Ross, Morgan Taschuk, Eric D. Wieben, Mathieu Wiepert, Derek E. Wildman, Liudmila S. Mainzer
University of Illinois at Urbana-Champaign, Mayo Clinic, Ontario Institute for Cancer Research
Frontiers in Genetics (2019). doi:10.3389/fgene.2019.00736

96. Whole exome sequencing aids the diagnosis of Simpson–Golabi–Behmel syndrome in two male fetuses
Jingjing Xiang*, Qin Zhang, Xiaoyan Song, Yinghua Lu, Haibo Li, Hong Li, Ting Wang
Affiliated Suzhou Hospital of Nanjing Medical University, Suzhou Municipal Hospital
Journal of Internal Medicine Research (2019). doi:10.1177/0300060519859752

97. Genome-wide rare variant analysis for thousands of phenotypes in 54,000 exomes
Elizabeth T. Cirulli, Simon White, Robert W. Read, Gai Elhanan, William J Metcalf, Karen A. Schlauch, Joseph J. Grzymski, James Lu, Nicole L. Washington
Helix, Desert Research Institute, Renown Institute of Health Innovation
bioRxiv 697368 (2019). doi:10.1101/697368

98. A robust benchmark for germline structural variant detection
Justin M. Zook, Nancy F. Hansen, Nathan D. Olson, Lesley M. Chapman, James C. Mullikin, Chunlin Xiao, Stephen Sherry, Sergey Koren, Adam M. Phillippy, Paul C. Boutros, Sayed Mohammad E. Sahraeian, Vincent Huang, Alexandre Rouette, Noah Alexander, Christopher E. Mason, Imari Hajirasouliha, Camir Ricketts, Joyce Lee, Rick Tearle, Ian T. Fiddes, Alvaro Martinez Barrio, Jeremiah Wala, Andrew Carroll, Noushin Ghaffari, Oscar L. Rodriguez, Ali Bashir, Shaun Jackman, John J Farrell, Aaron M Wenger, Can Alkan, Arda Soylev, Michael C. Schatz, Shilpa Garg, George Church, Tobias Marschall, Ken Chen, Xian Fan, Adam C. English, Jeffrey A. Rosenfeld, Weichen Zhou, Ryan E. Mills, Jay M. Sage, Jennifer R. Davis, Michael D. Kaiser, John S. Oliver, Anthony P. Catalano, Mark JP Chaisson, Noah Spies, Fritz J. Sedlazeck, Marc Salit, the Genome in a Bottle Consortium
Genome in a Bottle Consortium
bioRxiv 064623 (2019). doi:10.1101/064623

99. Bioinformatic Methods and Bridging of Assay Results for Reliable Tumor Mutational Burden Assessment in Non-Small-Cell Lung Cancer
Han Chang, Ariella Sasson, Sujaya Srinivasan, Ryan Golhar, Danielle M. Greenawald, William J. Geese, George Green, Kim Zarba, Stefan Kirov, Joseph Stuckelberger
Bristol-Myers Squibb
Molecular Diagnosis and Therapy (2019). doi:10.1007/s40291-019-00408-7

100. Phylogenomics reveals an ancient hybrid origin of the Persian walnut
Bo-Wen Zhang, Lin-Lin Xu, Nan Li, Peng-Cheng Yan, Xin-Hua Jiang, Keith E. Woeste, Xui Lin, Susanne S. Renner, Da-Yong Zhang, Wei-Ning Bai
Beijing Normal University and University of Munich
Molecular Biology and Evolution (2019). doi:10.1093/molbev/msz152

101. Tumor Shedding into Cell-free DNA (cfDNA) is Associated with High-mortality Cancers
Oliver Venn, Earl Hubbell, Onur Sakarya, Christopher Chang, Meredith Halks-Miller, Kristan Steffen, Eric T. Fung, Anne-Renee Hartman, Tara Maddala, Alexander M. Aravanis
Grail, Inc.
2019 Cold Spring Harbor Laboratory Meeting: The Biology of Genomes. Link

102. Acquired Resistance to BET-PROTACs (Proteolysis Targeting Chimeras) Caused by Genomic Alterations in Core Components of E3 ligase Complexes
Lu Zhang, Bridget Riley-Gillis, Priyanka Vijay, Yu Shen
AbbVie Inc.
Molecular Cancer Therapeutics (2019). doi:10.1158/1535-7163.MCT-18-1129

103. Establishing reference samples for detection of somatic mutations and germline variants with NGS technologies
Li Tai Fang, Bin Zhu, Yongmei Zhao, Wanqiu Chen, Zhaowei Yang, Lu Kerrigan, Kurt Langenbach, Maryellen de Mars, Charles Lu, Kenneth Idler, Howard Jacob, Ying Yu, Luyao Ren, Yuanting Zheng, Erich Jaeger, Gary Schroth, Ogan D. Abaan, Justin Lack, Tsai-Wei Shen, Keyur Talsania, Zhong Chen, Sela Stanbouly, Jodi Shetty, Biao Tan, Daoud Meerzaman, Gu Nguyen, Virginie Petitjean, Marc Sultan, Margaret Cam, Tiffany Hung, Eric Peters, Rasika Kalamegham, Sayed Mohammad Ebrahim Sahraeian, Marghoob Mohiyuddin, Yunfei Guo, Liping Yao, Lei Song, Hugo YK Lam, Ali Bashir, Roberta Maestro, Daniela Gasparotto, Alex Kiku, Eva Hellen Hoppe, Jessica Nordlund, Ulrika Liljedahl, Roderick V Jensen, Mehdi Pirooznia, Zhipan Li, Chunlin Xiao, Stephen Sherry, Rebecca Kusko, Malcolm Moos, Eric Donaldson, Zivana Tezak, Baitang Ning, Jing Li, Penelope Duerksen-Hughes, Huixiao Hong, Leming Shi, Charles Wang, Wenming Xiao, The Somatic Working Group of SEQC-II Consortium
The Somatic Working Group of SEQC-II Consortium
bioRxiv 629504 (2019). doi:10.1101/629504

104. Towards best practice in cancer mutation detection with whole-genome and whole-exome sequencing
The Somatic Mutation Working Group of the SEQC-II Consortium
The Somatic Working Group of SEQC-II Consortium
bioRxiv 626440 (2019). doi:10.1101/626440

105. Immune gene expression profiling reveals heterogeneity in luminal breast tumors
Bin Zhu, Shelly Lap Ah Tse, Difei Wang, Hela Koka, Tongwu Zhang, Mustapha Abubaker, Priscilla Lee, Feng Wang, Cherry Wu, Koon Ho Tsang, Wing-cheong Chan, Sze Ling Lee, Mengjie Li, Wentao Li, Suyang Wu, Zhiguang Liu, Biwa Huang, Han Zhang, Eric Tang, Zhengyan Sun, Soohyoun Lee, Yeon-Hee Park, Seok Jin Nam, Mingyi Wang, Xuesheng Sun, Kristine Jones, Bin Zhu, Amy Hutchinson, Belynda Hicks, Ludmila Prokunina-Olsson, Jianxin Shi, Montserrat Garcia-Closas, Stephen Chanock, Xiaohong R. Yang
National Cancer Institute, Chinese Academy of Medical Sciences, Fredericks National.
Breast Cancer Research 21, 147 (2019). doi:10.1186/s13058-019-1218-9

106. Genetic Profile and Clinical Implications of Hepatoblastoma and Neuroblastoma Coexistence in a Child
Szymon Skoczen, Konrad Stępień, Marta Krzyżyńska, Teresa Łuszawska, Małgorzata Wachowiak-Kolasa, Michał Konstytucki, Marcin Piechota, Katarzyna Kolanek, Łukasz Wyrobek, Katarzyna Wysocka, Wojciech Gorecki, Walentyna Balwierz
University Children's Hospital, Jagiellonian University Medical College, and Institute of Pharmacology of Polish Academy of Sciences, Poland
Frontiers in Oncology (2019). doi:10.3389/fonc.2019.00230

107. Raptor genomes reveal evolutionary signatures of predatory and nocturnal lifestyles
Yun Sung Cho, Jamyoon Jun, Jung A Kim, Hak-Min Kim, Oksung Chung, Seung-Gu Kang, Jin-Young Park, Hwa-Jung Kim, Sunghyun Kim, Hee-Jong Kim, Jin-Ho Jang, Yi-Joong Na, Jeongho Kim, Seung Gu Park, Hwang-Yeol Lee, Andrea Manica, David P. Mindell, Jérôme Fuchs, Jeremy S. Edwards, Jessica A. Weber, Christopher C. Witt, Joo-Hong Yeo, Soonok Kim, Jong Bhak
Clinomics Inc., Korea National Institute of Biological Resources, Ulsan National Institute of Science and Technology, Kongju National University, Chunghua National University, Cheongju Zoo, University of California, Berkeley, Sorbonne Universite, University of New Mexico
Genome Biology 20, 181 (2019). doi:10.1186/s13059-019-1793-1

108. Efficient and unique co-barcoding of second-generation sequencing reads from long DNA molecules enabling cost effective and accurate sequencing, haplotyping, and de novo assembly
Ou Wang, Robert Chin, Xiaofang Cheng, Michelle Ka Yan Wu, Qing Mao, Jingbo Tang, Yuhui Sun, Eric Anderson, Hank K. Lam, Dan Chen, Yujun Zhou, Linying Wang, Fei Fan, Yan Zou, Yinlong Xie, Rebecca Yu Zhang, Snezana Drmanac, Darlene Kingsley, Chongjun Xu, Christian Villarosa, Scott Gabriel, Nina Barua, Staci Nguyen, Wenlan Tian, Jia Sophie Liu, Jingwen Shen, Mari Sall
National Institute of Standards and Technology (NIST), 10x Genomics, Real Time Genomics, Invitae Corporation, Verily Life Sciences, Stanford University School of Medicine, National Institute of Health
Nature Biotechnology (2019). doi:10.1038/s41587-019-0074-6

111. Genomic and Transcriptomic Landscape of Triple-Negative Breast Cancers: Subtypes and Treatment Strategies
Yi-Zhou Jiang, Ding Ma, Chen Suo, Jinxu Shi, Mengchu Xue, Xin Hu, Yi Xiao, Ke-Da Yu, Yi-Rong Liu, Ying Yu, Yuanting Zheng, Xiangnan Li, Chenhui Zhang, Pengchen Hu, Jing Zhang, Qi Hua, Jiyang Zhang, Wanwan Hou, Luyao Ren, Ding Bao, Binyang Li, Jingcheng Yang, Ling Yao, Wen-Jia Zuo, Shen Zhao, Yue Gong, Yi-Xing Wen, Ya-Xin Zhao, Yun-Song Yang, Zhenmin Niu, Zhi-Ming Cao, Daniel G. Stover, Claire Verschraegen, Virginia Kaklamani, Anneleen Daemen, John W. Benson, Kazuaki Takabe, Fan Bai, Da-Qiang Li, Peng Wang, Leming Shi, Wei Huang, Zhi-Ming Shao
Fudan University, Chinese National Human Genome Center at Shanghai and Shanghai Industrial Technology Institute, Chinese Academy of Sciences, et al.
Cancer Cell (2019). doi:10.1016/j.ccell.2019.02.001

112. Rapid Genome Sequencing in the Critically Ill
Jillian G. Buchan, Shana White, Ruchi Joshi, Euan A. Ashley
Stanford Health Care
Clinical Chemistry 65 (2019). doi:10.1373/clinchem.2018.293506

113. TAMM41 is required for heart valve differentiation via regulation of PINK-PARK2 dependent mitophagy
Ru-Meng Yang, Jiong Tao, Ming-Zhan, Hao Yuan, Hai-Hong Wang, Sai-Juan Chen, Zhu Chen, Hugues de Thé, Jun Zhou, Ying Guo, Jun Zhu
Shanghai Jiao Tong University School of Medicine and Affiliated First Hospital
Cell Death & Differentiation (2019). doi:10.1038/s41418-019-0311-7

114. Discovery of Novel Titin (TTN) Gene Truncating Mutations among a Consecutive Cohort of Patients Presenting with Idiopathic Dilated Cardiomyopathy (IDC)
Jeffrey L Anderson, VictoriaJacobs, G. BryceChristensen, Helaman Escobar, Benjamin Horne, Stacey Knight, Kia Afshar, Virginia B Hebl, Kirk Knowlton, J Muhlestein, John Carlquist, Lincoln D Nadauld
Intermountain Medical Center, University of Utah
Journal of the American College of Cardiology 73, 699 (2019). doi:10.1016/S0735-1097(19)31767-7

115. The Galleria mellonella Hologenome Supports Microbiota-Independent Metabolism of Long-Chain Hydrocarbon Beeswax
Hyun Gi Kong, Hyun Ho Kim, Joon-hui Chung, JeHoon Jun, Soohyun Lee, Hak-Min Kim, Sungwon Jeon, Seung Gu Park, Jong Bhak, Choong-Min Ryu
Korea Research Institute of Bioscience & Biotechnology, Ulsan National Institute of Science and Technology, et al.
Cell Reports 26, 2451(2019). doi:10.1016/j.celrep.2019.02.018

116. Comparison of Biomarker Assays for Cytotoxic T Cell Infiltration for Precision Medicine in Patients with Glioblastoma
Andrew B. Lassman, Lisa Roberts-Rapp, Irina Sokolova, Minghao Song, Ekaterina Pestova, Rajender Kular, Carolyn Mullan, Zheng Zha, Xin Lu, Erica Gomez, Anahita Bhathena, David Maag, Priya Kumthekar, Hui K. Gan, Andrew M. Scott, Maria Guseva, Kyle D. Holen, Peter J. Ansell, Martin J. van den Bent
Columbia University Irving Medical Center, AbbVie, Abbott Molecular, Northwestern University Feinberg School of Medicine, Olivia Newton-John Cancer Research Institute, Erasmus MC Cancer Center.
Clinical Cancer Research (2019). doi:10.1158/1078-0432.CCR-18-3934

117. Analysis of mucosal metagenome-whole-genome sequencing reveals clinically relevant genomic aberrations
Rong Zhou, Chaoji Shi, Wenjie Tao, Jiang Li, Jing Wu, Yong Han, Guofu Wang, Ziyue Gu, Shengrong Xu, Yujue Wang, Lizhen Wang, Yanan Wang, Guoyu Zhou, Chenping Zhang, Zhiyuan Zhang, Shuyang Sun
Shanghai Jiao Tong University, National Clinical Research Center for Oral Diseases, Shanghai Key Laboratory of Stomatology and Shanghai Research Institute of Stomatology
Clinical Cancer Research (2019). doi:10.1158/1078-0432.CCR-18-3442

118. BMAL1 knockout macaque monkeys display reduced sleep and psychiatric disorders
Peiyuan Qiu, Jian Jiang, Zhen Liu, Yiyun Cai, Tao Huang, Yan Wang, Qiming Liu Yanhong Nie, Fang Liu, Jiumu Cheng, Qing Li, Yun-Chi Tang, Mu-ming Poo, Gang Sun, Hung-Chun Chang
Chinese Academy of Sciences, Shanghai Research Center for Brain Science and Brain-Inspired Technology
National Science Review (2019). doi:10.1093/nsr/nwz002

119. Deleterious Mutation Burden and its Association with Complex Traits in Sorghum (Sorghum bicolor)
Ravi Valluru, Elodie E. Gazave, Samuel B. Fernandes, John N. Ferguson, Roberto Lozano, Pradeep Hiramsash, Tao Zuo, Patrick J. Brown, Andrew D. B. Leakey, Michael A. Gore, Edward S. Buckler, Nancy Bandillo
Cornell University, University of Illinois at Urbana-Champaign, University of California, Davis
Genetics (2019). doi:10.1534/genetics.118.301742

120. Genetic Profiles Associated with Chemoresistance in Patient-Derived Xenograft Models of Ovarian Cancer
Lan Ying Li, Hee Jung Kim, Sun Ae Park, So Hyun Lee, Lee Kyung Kim, Jung Yun Lee, Sunghoon Kim, Young Tae Kim, Sang Wun Kim, Eun Ji Nam
Yonsei University College of Medicine
Cancer Research and Treatment (2018). doi:10.4143/crt.2018.495

121. PreMedKB: an integrated precision medicine knowledgebase for interpreting relationships between diseases, genes, variants and drugs
Ying Yu, Yunjin Wang, Zhaojia Xia, Xiangyu Zhang, Kailiang Jin, Jingcheng Yang, Luyao Ren, Zheng Zhou, Dong Yu, Tao Ding, Chengdong Zhang, Li Jin, Yuanting Zheng, Li Guo, Leming Shi
Fudan University and Chinese Academy of Sciences
Nucleic Acids Research, gky1034 (2018). doi:10.1093/nar/gky1034

122. How well can we create phased, diploid, human genomes?- An assessment of FALCON-Unzip phasing using a human trio
Arkarachai Fungtammasan, Brett Hannigan
DNANexus
bioRxiv 261296 (2018). doi:10.1101/261296

123. Variations in maternal adenylate cyclase genes are associated with congenital Zika syndrome in a cohort from Northeast, Brazil
A. D. Rossi, F. B. Faucz, A. Melo, P. Pezzuto, G. S. de Azevedo, B. L. F. Schaefer-Reis, J. S. Tavares, J. J. Mattapallil, A. Tanuri, R. S. Aguiar, C. C. Cardoso, C. A. Sirtaldo
Universidade Federal do Rio de Janeiro, National Cancer Institute, Instituto de Pesquisa Professor Joaquim Amorim Neto, Centro Universitario UniFacisa, Uniformed Services University
Journal of Internal Medicine (2018). doi:10.1111/joim.12829

124. Clinical, pedigree and genetic analysis of Alcardi-Gouterizes syndrome type 6 in a patient
Min Xu, Hu Guo, Xiaojiang Lu
Children's Hospital of Nanjing Medical University
Journal of Clinical Pediatrics 36, 686 (2018). doi:10.3969/j.issn.1000-3606.2018.09.010

125. High-resolution crossover mapping reveals similarities and differences of male and female recombination in maize
Penny M. A. Kianian, Minghui Wang, Kristin Simons, Farhad Ghavami, Yan He, Stefanie Dukowic-Schulze, Aintha Sundaresan, Qi Sun, Jaroslaw Pillardy, Joann Mudge, Changbin Chen, Shahryar F. Kianian, Wojciech P. Pawlowski
University of Minnesota, Cornell University, North Dakota State University, National Center for Genome Resources, USDA-ARS
Nature Communications 9, 2370 (2018). doi:10.1038/s41467-018-04562-5

126. Early Identification of Unusually Clustered Mutations and a Cue Protein in Therapeutic Antibody Development
Yuerning Qian, Zhiqiang Chen, Xin Huang, Xuning Wang, Xuankuo Xu, Stefan Kirov, Richard Ludwig, Nan-Xin Qian, Kandasamy Ravi, Li Tao, Michael C. Borys, Zheng Jian Li
Bristol-Myers Squibb Company
Biotechnology and Bioengineering 115, 2377 (2018). doi:10.1002/bit.26718

127. Tumor Mutational Burden and Efficacy of Nivolumab Monotherapy and in Combination with Ipilimumab in Small-Cell Lung Cancer
Matthew D. Hellmann, Margaret K. Callahan, Mark M. Awad, Emiliano Calvo, Paolo A. Ascierto, Ani Atmaca, Naiyer A. Rizvi, Fred R. Hirsch, Giovanni Selvaggi, Joseph D. Szustakowski, Arella Sasson, Ryan Golhar, Patrik Vitazka, Han Chang, William J. Geese, Scott J. Antonia
Weill Cornell Medical College, Dana-Farber Cancer Institute, et al.
Cancer Cell 33, 1-9 (2018). doi:10.1016/j.ccell.2018.04.001

128. Pathways impacted by genomic alterations in pulmonary carcinoid tumors
Michael R. Rossiello, Charles F. Thomas Jr., Jie Dong, Sandra C. Schulte, Prasiddha Khadka, Zhifu Sun, Farhad Kosari, Jin Jen, Julian Molina, George Vasmatzis, Ray Kuang, Maria Christine Aubry, Ping Yang, Dennis A. Wigle
Mayo Clinic College of Medicine, University of Minnesota
Clinical Cancer Research 24, 1651 (2018). doi:10.1158/1078-0432.CCR-17-0252

129. First-Line Nivolumab in Stage IV or Recurrent Non–Small-Cell Lung Cancer
David P. Carbone, Martin Reck, Luis Paz-Ares, Benjamin Creelan, Leora Horn, Martin Steins, Enriqueta Felip, Michel M. van den Heuvel, Tudor-Eliade Ciuleanu, Firas Badin, Neal Ready, T. Jeroen N. Hiltermann, Suresh Nair, Rosalyn Juergens, Solange Peters, Elisa Minenza, John M. Wrangle, Delvys Rodriguez-Abreu, Hossein Borghaei, D.O., George R. Blumenschein, Jr, Lyu G. Villaruz, Libor Havel, Jana Krejci, Jesus Corral Jaime, Han Chang, William J. Geese, Prabhu Bhagavatheeswaran, Allen C. Chen, Mark A. Socinski for CheckMate 026 investigators
Ohio State University Comprehensive Cancer Center, Bristol-Myers Squibb, et al.
New England Journal of Medicine 2017 376:2415-2426. doi:10.1056/NEJMoa1613493

130. Splicing modulators act at the spliceosome catalytic step in pre-mRNA processing
Teng Teng, Jennifer HC Tsai, Xiaoling Puyang, Michael Seiler, Shouyong Peng, Sudeep Prajapati, Daniel Aird, Silva Buonamici, Benjamin Caleb, Betty Chan, Laura Corson, Jacob Feala, Peter Fekkes, Baudouin Gerard, Craig Kan, Manav Korpal, Xiang Liu, Jason T. Lowe, Yoshiharu Mizui, James Palacino, Eunice Park, Peter G. Smith, Vanitha Subramanian, Zhenhua Jeremy Wu, Jian Zou, Lihua Yu, Agustin Chicas, Markus Warmuth, Nicholas Larsen, Ping Zhu
H3 Biomedicine Inc
Nature Communications 8, 1525((2017). doi:10.1038/ncomms15522

131. Using High Performance Computing to Create and Freely Distribute the South Asian Genomic Database, Necessary for Precision Medicine in this Population
Asma H. Shah, Jonathan D. Picker, Saumya S. Jamuar
Global Gene Corporation Pte Ltd
Supercomputing Frontiers and Innovations: 4, 2016(17). doi:10.14529/jsfi170201

132. Identification of a novel mutation in the APTX gene associated with ataxia-oculomotor apraxia
Jingga Inaya M, Reza Sailani, Hamidreza Khodadadi, Ahmad Teymourirad, Shinichi Takahashi, Jonathan A. Bernstein, Masoud Garshasbi, Michael P. Snyder
Stanford University, Shahid Beheshti University of Medical Sciences, Ilam University of Medical Sciences, Tarbiat Modares University, Tehran DeNA Laboratory
Cold Spring Harb Molecular Case Studies 3, mcs.a002014 (2017). doi:10.1101/mcs.a002014

133. Genetic architecture drives seasonal onset of hibernation in the 13-lined ground squirrel
Katharine R. Grabek, Thomas F. Cooke, L. Elaine Epperson, Kaitlyn K. Speas, Gleyce F. Cabral, Shirley C. Sutton, Dana K. Merriman, Sandy L. Martin, Carlos D. Bustamante
Stanford University School of Medicine, National Jewish Health, University of Wisconsin Oshkosh, University of Colorado School of Medicine
bioRxiv 222307 (2017). doi:10.1101/222307

To quote Sentieon tools, please use:

The Sentieon Genomics Tools - A fast and accurate solution to variant calling from next-generation sequence data
Donald H. Freed, Rafael Aldana, Jessica A Weber, Jeremy S Edwards
bioRxiv 115717; doi:10.1101/115717

For Sentieon TNscope, please use:

TNscope: Accurate Detection of Somatic Mutations with Haplotype-based Variant Candidate Detection and Machine Learning Filtering
Donald Freed, Renke Pan, Rafael Aldana
bioRxiv 250647; doi:10.1101/250647