1. Abrudan J, Ramalho-Ortigao M, O'Neil S, Stayback G, Wadsworth M, Bernard M, Shoue D, Emrich S, Lawyer P, Kamhawi S et al. 2013. The characterization of the Phlebotomus papatasi transcriptome. *Insect Molecular Biology* **22**(2): 211-232.
2. Abubucker S, Martin J, Taylor CM, Mitreva M. 2011. HelmCoP: An Online Resource for Helminth Functional Genomics and Drug and Vaccine Targets Prioritization. *Plos One* **6**(7).
3. Abubucker S, McNulty SN, Rosa BA, Mitreva M. 2014. Identification and characterization of alternative splicing in parasitic nematode transcriptomes. *Parasites & Vectors* **7**.
4. Abubucker S, Segata N, Goll J, Schubert AM, Izard J, Cantarel BL, Rodriguez-Mueller B, Zucker J, Thiagarajan M, Henrissat B et al. 2012. Metabolic Reconstruction for Metagenomic Data and Its Application to the Human Microbiome. *Plos Computational Biology* **8**(6).
5. Adiconis X, Borges-Rivera D, Satija R, DeLuca DS, Busby MA, Berlin AM, Sivachenko A, Thompson DA, Wysoker A, Fennell T et al. 2013. Comparative analysis of RNA sequencing methods for degraded or low-input samples. *Nat Methods* **10**(7): 623-629.
6. Agrawal N, Frederick MJ, Pickering CR, Bettegowda C, Chang K, Li RJ, Fakhry C, Xie TX, Zhang J, Wang J et al. 2011. Exome sequencing of head and neck squamous cell carcinoma reveals inactivating mutations in NOTCH1. *Science* **333**(6046): 1154-1157.
7. Aird D, Ross MG, Chen WS, Danielsson M, Fennell T, Russ C, Jaffe DB, Nusbaum C, Gnirke A. 2011. Analyzing and minimizing PCR amplification bias in Illumina sequencing libraries. *Genome Biol* **12**(2): R18.
8. Akizu N, Shembesh NM, Ben-Omran T, Bastaki L, Al-Tawari A, Zaki MS, Koul R, Spencer E, Rosti RO, Scott E et al. 2013. Whole-exome sequencing identifies mutated c12orf57 in recessive corpus callosum hypoplasia. *Am J Hum Genet* **92**(3): 392-400.
9. Akizu N, Silhavy JL, Rosti RO, Scott E, Fenstermaker AG, Schroth J, Zaki MS, Sanchez H, Gupta N, Kabra M et al. 2014. Mutations in CSPP1 lead to classical Joubert syndrome. *Am J Hum Genet* **94**(1): 80-86.
10. Alegado RA, Ferriera S, Nusbaum C, Young SK, Zeng Q, Imamovic A, Fairclough SR, King N. 2011. Complete genome sequence of Algoriphagus sp. PR1, bacterial prey of a colony-forming choanoflagellate. *Journal of bacteriology* **193**(6): 1485-1486.
11. Alfoldi J, Di Palma F, Grabherr M, Williams C, Kong L, Mauceli E, Russell P, Lowe CB, Glor RE, Jaffe JD et al. 2011. The genome of the green anole lizard and a comparative analysis with birds and mammals. *Nature* **477**(7366): 587-591.
12. Alfoldi J, Lindblad-Toh K. 2013. Comparative genomics as a tool to understand evolution and disease. *Genome Res* **23**(7): 1063-1068.
13. Ali BR, Silhavy JL, Akawi NA, Gleeson JG, Al-Gazali L. 2012. A mutation in KIF7 is responsible for the autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia and distinctive facial appearance. *Orphanet journal of rare diseases* **7**: 27.
14. Altshuler D Durbin RM Abecasis GR Bentley DR Chakravarti A Clark AG Collins FS De la Vega FM Donnelly P Egholm M et al. 2010. A map of human genome variation from population-scale sequencing. *Nature* **467**(7319): 1061-1073.
15. Altshuler DM, Gibbs RA, Peltonen L, Altshuler DM, Peltonen L, Dermitzakis E, Schaffner SF, Yu F, Peltonen L, Dermitzakis E et al. 2010. Integrating common and rare genetic variation in diverse human populations. *Nature* **467**(7311): 52-58.
16. Amemiya CT, Alfoldi J, Lee AP, Fan S, Philippe H, Maccallum I, Braasch I, Manousaki T, Schneider I, Rohner N et al. 2013. The African coelacanth genome provides insights into tetrapod evolution. *Nature* **496**(7445): 311-316.
17. Amirian ES, Bondy ML, Mo Q, Bainbridge MN, Scheurer ME. 2014. Presence of viral DNA in whole-genome sequencing of brain tumor tissues from the cancer genome atlas *JVirol* **88**(1): 774.
18. Andersson AK, Ma J, Wang J, Chen X, Rusch M, Wu G, Easton J, Parker M, Raimondi SC, Holmfeldt L et al. 2011. Whole Genome Sequence Analysis of 22 MLL Rearranged Infant Acute Lymphoblastic Leukemias Reveals Remarkably Few Somatic Mutations: A Report From the St Jude Children's Research Hospital - Washington University Pediatric Cancer Genome Project. *Blood* **118**(21): 33-34.
19. Antonacci F, Kidd JM, Marques-Bonet T, Teague B, Ventura M, Girirajan S, Alkan C, Campbell CD, Vives L, Malig M et al. 2010. A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. *Nature Genetics* **42**(9): 745-U729.
20. Arnaout R, Lee W, Cahill P, Honan T, Sparrow T, Weiand M, Nusbaum C, Rajewsky K, Koralov SB. 2011. High-resolution description of antibody heavy-chain repertoires in humans. *PLoS One* **6**(8): e22365.
21. Axelsson E, Ratnakumar A, Arendt ML, Maqbool K, Webster MT, Perloski M, Liberg O, Arnemo JM, Hedhammar A, Lindblad-Toh K. 2013. The genomic signature of dog domestication reveals adaptation to a starch-rich diet. *Nature* **495**(7441): 360-364.
22. Axelsson E, Webster MT, Ratnakumar A, Consortium L, Ponting CP, Lindblad-Toh K. 2012. Death of PRDM9 coincides with stabilization of the recombination landscape in the dog genome. *Genome Res* **22**(1): 51-63.
23. Baca SC, Prandi D, Lawrence MS, Mosquera JM, Romanel A, Drier Y, Park K, Kitabayashi N, MacDonald TY, Ghandi M et al. 2013. Punctuated evolution of prostate cancer genomes. *Cell* **153**(3): 666-677.
24. Bacino CA, Arriola LA, Wiszniewska J, Bonnen PE. 2012. WDR62 missense mutation in a consanguineous family with primary microcephaly *AmJMedGenetA* **158A**(3): 622-625.
25. Bacino CA, Dhar SU, Brunetti-Pierri N, Lee B, Bonnen PE. 2012. WDR35 mutation in siblings with Sensenbrenner syndrome: a ciliopathy with variable phenotype. *AmJMedGenetA* **158A**(11): 2917-2924.
26. Bai X, Adams BJ, Ciche TA, Clifton S, Gaugler R, Kim K-s, Spieth J, Sternberg PW, Wilson RK, Grewal PS. 2013. A Lover and a Fighter: The Genome Sequence of an Entomopathogenic Nematode Heterorhabditis bacteriophora. *Plos One* **8**(7).
27. Bainbridge MN, Hu H, Muzny DM, Musante L, Lupski JR, Graham BH, Chen W, Gripp KW, Jenny K, Wienker TF et al. 2013. De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. *Genome Med* **5**(2): 11.
28. Bainbridge MN, Wang M, Burgess DL, Kovar C, Rodesch MJ, D'Ascenzo M, Kitzman J, Wu YQ, Newsham I, Richmond TA et al. 2010. Whole exome capture in solution with 3 Gbp of data. *Genome Biol* **11**(6): R62.
29. Bainbridge MN, Wang M, Wu Y, Newsham I, Muzny DM, Jefferies JL, Albert TJ, Burgess DL, Gibbs RA. 2011. Targeted enrichment beyond the consensus coding DNA sequence exome reveals exons with higher variant densities *Genome Biol* **12**(7): R68.
30. Balakrishnan CN, Ekblom R, Voelker M, Westerdahl H, Godinez R, Kotkiewicz H, Burt DW, Graves T, Griffin DK, Warren WC et al. 2010. Gene duplication and fragmentation in the zebra finch major histocompatibility complex. *Bmc Biology* **8**.
31. Ball WS, Byars AW, Schapiro M, Bommer W, Carr A, German A, Dunn S, Rivkin MJ, Waber D, Mulkern R et al. 2012. Total and Regional Brain Volumes in a Population-Based Normative Sample from 4 to 18 Years: The NIH MRI Study of Normal Brain Development. *Cerebral Cortex* **22**(1): 1-12.
32. Bamshad MJ, Shendure JA, Valle D, Hamosh A, Lupski JR, Gibbs RA, Boerwinkle E, Lifton RP, Gerstein M, Gunel M et al. 2012. The Centers for Mendelian Genomics: a new large-scale initiative to identify the genes underlying rare Mendelian conditions. *AmJMedGenetA* **158A**(7): 1523-1525.
33. Banerji S, Cibulskis K, Rangel-Escareno C, Brown KK, Carter SL, Frederick AM, Lawrence MS, Sivachenko AY, Sougnez C, Zou L et al. 2012. Sequence analysis of mutations and translocations across breast cancer subtypes. *Nature* **486**(7403): 405-409.
34. Barbieri CE, Baca SC, Lawrence MS, Demichelis F, Blattner M, Theurillat JP, White TA, Stojanov P, Van Allen E, Stransky N et al. 2012. Exome sequencing identifies recurrent SPOP, FOXA1 and MED12 mutations in prostate cancer. *Nat Genet* **44**(6): 685-689.
35. Barretina J, Caponigro G, Stransky N, Venkatesan K, Margolin AA, Kim S, Wilson CJ, Lehar J, Kryukov GV, Sonkin D et al. 2012. The Cancer Cell Line Encyclopedia enables predictive modelling of anticancer drug sensitivity. *Nature* **483**(7391): 603-607.
36. Barretina J, Taylor BS, Banerji S, Ramos AH, Lagos-Quintana M, Decarolis PL, Shah K, Socci ND, Weir BA, Ho A et al. 2010. Subtype-specific genomic alterations define new targets for soft-tissue sarcoma therapy. *Nat Genet* **42**(8): 715-721.
37. Bass AJ, Lawrence MS, Brace LE, Ramos AH, Drier Y, Cibulskis K, Sougnez C, Voet D, Saksena G, Sivachenko A et al. 2011. Genomic sequencing of colorectal adenocarcinomas identifies a recurrent VTI1A-TCF7L2 fusion. *Nat Genet* **43**(10): 964-968.
38. Baud A, Hermsen R, Guryev V, Stridh P, Graham D, McBride MW, Foroud T, Calderari S, Diez M, Ockinger J et al. 2013. Combined sequence-based and genetic mapping analysis of complex traits in outbred rats. *NatGenet* **45**(7): 767-775.
39. Bell D Berchuck A Birrer M Chien J Cramer DW Dao F Dhir R DiSaia P Gabra H Glenn P et al. 2011. Integrated genomic analyses of ovarian carcinoma. *Nature* **474**(7353): 609-615.
40. Bellone RR, Forsyth G, Leeb T, Archer S, Sigurdsson S, Imsland F, Mauceli E, Engensteiner M, Bailey E, Sandmeyer L et al. 2010. Fine-mapping and mutation analysis of TRPM1: a candidate gene for leopard complex (LP) spotting and congenital stationary night blindness in horses. *Briefings in functional genomics* **9**(3): 193-207.
41. Bellott DW, Hughes JF, Skaletsky H, Brown LG, Pyntikova T, Cho TJ, Koutseva N, Zaghlul S, Graves T, Rock S et al. 2014. Mammalian Y chromosomes retain widely expressed dosage-sensitive regulators. *Nature* **508**(7497): 494-499.
42. Bellott DW, Hughes JF, Skaletsky H, Brown LG, Pyntikova T, Cho T-J, Koutseva N, Zaghlul S, Graves T, Rock S et al. 2014. Mammalian Y chromosomes retain widely expressed dosage-sensitive regulators. *Nature* **508**(7497): 494-+.
43. Bellott DW, Skaletsky H, Pyntikova T, Mardis ER, Graves T, Kremitzki C, Brown LG, Rozen S, Warren WC, Wilson RK et al. 2010. Convergent evolution of chicken Z and human X chromosomes by expansion and gene acquisition. *Nature* **466**(7306): 612-U613.
44. Berger MF, Hodis E, Heffernan TP, Deribe YL, Lawrence MS, Protopopov A, Ivanova E, Watson IR, Nickerson E, Ghosh P et al. 2012. Melanoma genome sequencing reveals frequent PREX2 mutations. *Nature* **485**(7399): 502-506.
45. Berger MF, Lawrence MS, Demichelis F, Drier Y, Cibulskis K, Sivachenko AY, Sboner A, Esgueva R, Pflueger D, Sougnez C et al. 2011. The genomic complexity of primary human prostate cancer. *Nature* **470**(7333): 214-220.
46. Berger MF, Levin JZ, Vijayendran K, Sivachenko A, Adiconis X, Maguire J, Johnson LA, Robinson J, Verhaak RG, Sougnez C et al. 2010. Integrative analysis of the melanoma transcriptome. *Genome Res* **20**(4): 413-427.
47. Beroukhim R, Mermel CH, Porter D, Wei G, Raychaudhuri S, Donovan J, Barretina J, Boehm JS, Dobson J, Urashima M et al. 2010. The landscape of somatic copy-number alteration across human cancers. *Nature* **463**(7283): 899-905.
48. Biankin AV Waddell N Kassahn KS Gingras MC Muthuswamy LB Johns AL Miller DK Wilson PJ Patch AM Wu J et al. 2012. Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes *Nature*.
49. Bick AG, Flannick J, Ito K, Cheng S, Vasan RS, Parfenov MG, Herman DS, DePalma SR, Gupta N, Gabriel SB et al. 2012. Burden of rare sarcomere gene variants in the Framingham and Jackson Heart Study cohorts. *Am J Hum Genet* **91**(3): 513-519.
50. Bis JC, DeStefano A, Liu X, Brody JA, Choi SH, Verhaaren BF, Debette S, Ikram MA, Shahar E, Butler KR, Jr. et al. 2014. Associations of NINJ2 Sequence Variants with Incident Ischemic Stroke in the Cohorts for Heart and Aging in Genomic Epidemiology (CHARGE) Consortium. *PLoS One* **9**(6): e99798.
51. Bis JC, White CC, Franceschini N, Brody J, Zhang X, Muzny D, Santibanez J, Gibbs R, Liu X, Lin H et al. 2014. Sequencing of 2 Subclinical Atherosclerosis Candidate Regions in 3669 Individuals: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Targeted Sequencing Study. *Circulation Cardiovascular genetics* **7**(3): 359-364.
52. Blumenstiel B, Cibulskis K, Fisher S, DeFelice M, Barry A, Fennell T, Abreu J, Minie B, Costello M, Young G et al. 2010. Targeted exon sequencing by in-solution hybrid selection. *Current protocols in human genetics / editorial board, Jonathan L Haines [et al]* **Chapter 18**: Unit 18 14.
53. Boone PM, Campbell IM, Baggett BC, Soens ZT, Rao MM, Hixson PM, Patel A, Bi W, Cheung SW, Lalani SR et al. 2013. Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. *Genome Res* **23**(9): 1383-1394.
54. Boone PM, Soens ZT, Campbell IM, Stankiewicz P, Cheung SW, Patel A, Beaudet AL, Plon SE, Shaw CA, McGuire AL et al. 2013. Incidental copy-number variants identified by routine genome testing in a clinical population. *GenetMed* **15**(1): 45-54.
55. Borloo J, De Graef J, Peelaers I, Nguyen DL, Mitreva M, Devreese B, Hokke CH, Vercruysse J, Claerebout E, Geldhof P. 2013. In-Depth Proteomic and Glycomic Analysis of the Adult-Stage Cooperia oncophora Excretome/Secretome. *Journal of Proteome Research* **12**(9): 3900-3911.
56. Bosch DG, Boonstra FN, Gonzaga-Jauregui C, Xu M, de LJ, Jhangiani S, Wiszniewski W, Muzny DM, Yntema HG, Pfundt R et al. 2014. NR2F1 mutations cause optic atrophy with intellectual disability *AmJHumGenet* **94**(2): 303-309.
57. Bose R, Kavuri SM, Searleman AC, Shen W, Shen D, Koboldt DC, Monsey J, Goel N, Aronson AB, Li S et al. 2013. Activating HER2 Mutations in HER2 Gene Amplification Negative Breast Cancer. *Cancer Discovery* **3**(2): 224-237.
58. Bowne SJ, Humphries MM, Sullivan LS, Kenna PF, Tam LCS, Kiang AS, Campbell M, Weinstock GM, Koboldt DC, Ding L et al. 2011. A dominant mutation in RPE65 identified by whole-exome sequencing causes retinitis pigmentosa with choroidal involvement. *European Journal of Human Genetics* **19**(10): 1074-1081.
59. Bowne SJ, Sullivan LS, Koboldt DC, Ding L, Fulton R, Abbott RM, Sodergren EJ, Birch DG, Wheaton DH, Heckenlively JR et al. 2011. Identification of Disease-Causing Mutations in Autosomal Dominant Retinitis Pigmentosa (adRP) Using Next-Generation DNA Sequencing. *Investigative Ophthalmology & Visual Science* **52**(1): 494-503.
60. Bradnam KR, Fass JN, Alexandrov A, Baranay P, Bechner M, Birol I, Boisvert S, Chapman JA, Chapuis G, Chikhi R et al. 2013. Assemblathon 2: evaluating de novo methods of genome assembly in three vertebrate species. *Gigascience* **2**(1): 10.
61. Brastianos PK, Taylor-Weiner A, Manley PE, Jones RT, Dias-Santagata D, Thorner AR, Lawrence MS, Rodriguez FJ, Bernardo LA, Schubert L et al. 2014. Exome sequencing identifies BRAF mutations in papillary craniopharyngiomas. *Nat Genet* **46**(2): 161-165.
62. Brennan CW, Verhaak RG, McKenna A, Campos B, Noushmehr H, Salama SR, Zheng S, Chakravarty D, Sanborn JZ, Berman SH et al. 2013. The somatic genomic landscape of glioblastoma *Cell* **155**(2): 462-477.
63. Browne SK, Rosen LB, Freeman AF, Yang L, Jutivorakool K, Olivier KN, Angkasekwinai N, Suputtamongkol Y, Bennett J, Pyrgos V et al. 2013. ANTI-GRANULOCYTE-MACROPHAGE COLONY STIMULATING FACTOR AUTOANTIBODIES IN PATIENTS WITH CRYPTOCOCCAL MENINGITIS. *Journal of Clinical Immunology* **33**(3): 677-677.
64. Browne SK, Zaman R, Sampaio EP, Jutivorakool K, Rosen LB, Ding L, Pancholi MJ, Yang LM, Priel DL, Uzel G et al. 2012. Anti-CD20 (rituximab) therapy for anti-IFN-gamma autoantibody-associated nontuberculous mycobacterial infection. *Blood* **119**(17): 3933-3939.
65. Brunicardi FC, Gibbs RA, Wheeler DA, Nemunaitis J, Fisher W, Goss J, Chen C. 2011. Overview of the development of personalized genomic medicine and surgery. *World JSurg* **35**(8): 1693-1699.
66. Burrage LC, Lu JT, Liu DS, Moss TJ, Gibbs R, Schlesinger AE, Bacino CA, Campeau PM, Lee BH. 2013. Early childhood presentation of Czech dysplasia. *ClinDysmorphol* **22**(2): 76-80.
67. Butte NF, Voruganti VS, Cole SA, Haack K, Comuzzie AG, Muzny DM, Wheeler DA, Chang K, Hawes A, Gibbs RA. 2011. Resequencing of IRS2 reveals rare variants for obesity but not fasting glucose homeostasis in Hispanic children *Physiol Genomics* **43**(18): 1029-1037.
68. Buxbaum JD, Daly MJ, Devlin B, Lehner T, Roeder K, State MW, Autism Sequencing C. 2012. The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron* **76**(6): 1052-1056.
69. Cabanski CR, Magrini V, Griffith M, Griffith OL, McGrath S, Zhang J, Walker J, Ly A, Demeter R, Fulton RS et al. 2014. cDNA hybrid capture improves transcriptome analysis on low-input and archived samples. *The Journal of molecular diagnostics : JMD* **16**(4): 440-451.
70. Caliskan M, Chong JX, Uricchio L, Anderson R, Chen P, Sougnez C, Garimella K, Gabriel SB, dePristo MA, Shakir K et al. 2011. Exome sequencing reveals a novel mutation for autosomal recessive non-syndromic mental retardation in the TECR gene on chromosome 19p13. *Hum Mol Genet* **20**(7): 1285-1289.
71. Calvo SE, Compton AG, Hershman SG, Lim SC, Lieber DS, Tucker EJ, Laskowski A, Garone C, Liu S, Jaffe DB et al. 2012. Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing. *Science translational medicine* **4**(118): 118ra110.
72. Campbell NG, Zhu CB, Lindler KM, Yaspan BL, Kistner-Griffin E, Hewlett WA, Tate CG, Blakely RD, Sutcliffe JS. 2013. Rare coding variants of the adenosine A3 receptor are increased in autism: on the trail of the serotonin transporter regulome *MolAutism* **4**(1): 28.
73. Campeau PM, Kasperaviciute D, Lu JT, Burrage LC, Kim C, Hori M, Powell BR, Stewart F, Felix TM, van den Ende J et al. 2014. The genetic basis of DOORS syndrome: an exome-sequencing study *Lancet Neurol* **13**(1): 44-58.
74. Campeau PM, Kim JC, Lu JT, Schwartzentruber JA, Abdul-Rahman OA, Schlaubitz S, Murdock DM, Jiang MM, Lammer EJ, Enns GM et al. 2012. Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome *AmJHumGenet* **90**(2): 282-289.
75. Campeau PM, Lenk GM, Lu JT, Bae Y, Burrage L, Turnpenny P, Roman Corona-Rivera J, Morandi L, Mora M, Reutter H et al. 2013. Yunis-Varon syndrome is caused by mutations in FIG4, encoding a phosphoinositide phosphatase *AmJHumGenet* **92**(5): 781-791.
76. Campeau PM, Lu JT, Dawson BC, Fokkema IF, Robertson SP, Gibbs RA, Lee BH. 2012. The KAT6B-related disorders genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. *HumMutat*.
77. Campeau PM, Lu JT, Sule G, Jiang MM, Bae Y, Madan S, Hogler W, Shaw NJ, Mumm S, Gibbs RA et al. 2012. Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. *HumMolGenet* **21**(22): 4904-4909.
78. Cancer Genome Atlas N. 2012. Comprehensive molecular characterization of human colon and rectal cancer. *Nature* **487**(7407): 330-337.
79. -. 2012. Comprehensive molecular portraits of human breast tumours. *Nature* **490**(7418): 61-70.
80. Cancer Genome Atlas Research N. 2011. Integrated genomic analyses of ovarian carcinoma. *Nature* **474**(7353): 609-615.
81. -. 2012. Comprehensive genomic characterization of squamous cell lung cancers. *Nature* **489**(7417): 519-525.
82. -. 2013. Comprehensive molecular characterization of clear cell renal cell carcinoma. *Nature* **499**(7456): 43-49.
83. -. 2013. Genomic and epigenomic landscapes of adult de novo acute myeloid leukemia. *The New England journal of medicine* **368**(22): 2059-2074.
84. Cancer Genome Atlas Research N, Kandoth C, Schultz N, Cherniack AD, Akbani R, Liu Y, Shen H, Robertson AG, Pashtan I, Shen R et al. 2013. Integrated genomic characterization of endometrial carcinoma. *Nature* **497**(7447): 67-73.
85. Cantacessi C, Gasser RB, Strube C, Schnieder T, Jex AR, Hall RS, Campbell BE, Young ND, Ranganathan S, Sternberg PW et al. 2011. Deep insights into Dictyocaulus viviparus transcriptomes provides unique prospects for new drug targets and disease intervention. *Biotechnology Advances* **29**(3): 261-271.
86. Cantacessi C, Hofmann A, Pickering D, Navarro S, Mitreva M, Loukas A. 2013. TIMPs of parasitic helminths - a large-scale analysis of high-throughput sequence datasets. *Parasites & Vectors* **6**.
87. Cantacessi C, Jex AR, Hall RS, Young ND, Campbell BE, Joachim A, Nolan MJ, Abubucker S, Sternberg PW, Ranganathan S et al. 2010. A practical, bioinformatic workflow system for large data sets generated by next-generation sequencing. *Nucleic Acids Research* **38**(17).
88. Cantacessi C, Mitreva M, Campbell BE, Hall RS, Young ND, Jex AR, Ranganathan S, Gasser RB. 2010. First transcriptomic analysis of the economically important parasitic nematode, Trichostrongylus colubriformis, using a next-generation sequencing approach. *Infection Genetics and Evolution* **10**(8): 1199-1207.
89. Cantacessi C, Mitreva M, Jex AR, Young ND, Campbell BE, Hall RS, Doyle MA, Ralph SA, Rabelo EM, Ranganathan S et al. 2010. Massively Parallel Sequencing and Analysis of the Necator americanus Transcriptome. *Plos Neglected Tropical Diseases* **4**(5).
90. Caramins M, Colebatch JG, Bainbridge MN, Scherer SS, Abrams CK, Hackett EL, Freidin MM, Jhangiani SN, Wang M, Wu Y et al. 2013. Exome sequencing identification of a GJB1 missense mutation in a kindred with X-linked spinocerebellar ataxia (SCA-X1) *HumMolGenet* **22**(21): 4329-4338.
91. Carl MA, Ndao IM, Springman AC, Manning SD, Johnson JR, Johnston BD, Burnham C-AD, Weinstock ES, Weinstock GM, Wylie TN et al. 2014. Sepsis From the Gut: The Enteric Habitat of Bacteria That Cause Late-Onset Neonatal Bloodstream Infections. *Clinical Infectious Diseases* **58**(9): 1211-1218.
92. Carneiro MO, Russ C, Ross MG, Gabriel SB, Nusbaum C, DePristo MA. 2012. Pacific biosciences sequencing technology for genotyping and variation discovery in human data. *BMC Genomics* **13**: 375.
93. Carter SL, Cibulskis K, Helman E, McKenna A, Shen H, Zack T, Laird PW, Onofrio RC, Winckler W, Weir BA et al. 2012. Absolute quantification of somatic DNA alterations in human cancer. *Nat Biotechnol* **30**(5): 413-421.
94. Carvalho CM, Pehlivan D, Ramocki MB, Fang P, Alleva B, Franco LM, Belmont JW, Hastings PJ, Lupski JR. 2013. Replicative mechanisms for CNV formation are error prone. *NatGenet* **45**(11): 1319-1326.
95. Carvalho CM, Zuccherato LW, Williams CL, Neill NJ, Murdock DR, Bainbridge M, Jhangiani SN, Muzny DM, Gibbs RA, Ip W et al. 2014. Structural variation and missense mutation in SBDS associated with Shwachman-Diamond syndrome. *BMC medical genetics* **15**(1): 64.
96. Castoe TA, Bronikowski AM, Brodie ED, III, Edwards SV, Pfrender ME, Shapiro MD, Pollock DD, Warren WC. 2011. A proposal to sequence the genome of a garter snake (Thamnophis sirtalis). *Standards in Genomic Sciences* **4**(2): 257-270.
97. Castoe TA, de Koning APJ, Hall KT, Card DC, Schield DR, Fujita MK, Ruggiero RP, Degner JF, Daza JM, Gu W et al. 2013. The Burmese python genome reveals the molecular basis for extreme adaptation in snakes. *Proceedings of the National Academy of Sciences of the United States of America* **110**(51): 20645-20650.
98. Castoe TA, de Koning APJ, Hall KT, Yokoyama KD, Gu W, Smith EN, Feschotte C, Uetz P, Ray DA, Dobry J et al. 2011. Sequencing the genome of the Burmese python (Python molurus bivittatus) as a model for studying extreme adaptations in snakes. *Genome Biology* **12**(7).
99. Cejkova D, Zobanikova M, Chen L, Pospisilova P, Strouhal M, Qin X, Mikalova L, Norris SJ, Muzny DM, Gibbs RA et al. 2012. Whole Genome Sequences of Three Treponema pallidum ssp. pertenue Strains: Yaws and Syphilis Treponemes Differ in Less than 0.2% of the Genome Sequence *PLoSNeglTropDis* **6**(1): e1471.
100. Chaki M, Airik R, Ghosh AK, Giles RH, Chen R, Slaats GG, Wang H, Hurd TW, Zhou W, Cluckey A et al. 2012. Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling. *Cell* **150**(3): 533-548.
101. Challis D, Yu J, Evani US, Jackson AR, Paithankar S, Coarfa C, Milosavljevic A, Gibbs RA, Yu F. 2012. An integrative variant analysis suite for whole exome next-generation sequencing data *BMCBioinformatics* **13**(1): 8.
102. Chapman MA, Lawrence MS, Keats JJ, Cibulskis K, Sougnez C, Schinzel AC, Harview CL, Brunet JP, Ahmann GJ, Adli M et al. 2011. Initial genome sequencing and analysis of multiple myeloma. *Nature* **471**(7339): 467-472.
103. Chen F, Ding L. 2012. Co-survival of the fittest few: mosaic amplification of receptor tyrosine kinases in glioblastoma. *Genome Biology* **13**(1).
104. Chen K, Chen L, Fan X, Wallis J, Ding L, Weinstock G. 2014. TIGRA: A targeted iterative graph routing assembler for breakpoint assembly. *Genome Research* **24**(2): 310-317.
105. Chen K, Navin NE, Wang Y, Schmidt HK, Wallis JW, Niu B, Fan X, Zhao H, McLellan MD, Hoadley KA et al. 2013. BreakTrans: uncovering the genomic architecture of gene fusions. *Genome Biology* **14**(8).
106. Chen K, Wallis JW, Kandoth C, Kalicki-Veizer JM, Mungall KL, Mungall AJ, Jones SJ, Marra MA, Ley TJ, Mardis ER et al. 2012. BreakFusion: targeted assembly-based identification of gene fusions in whole transcriptome paired-end sequencing data. *Bioinformatics* **28**(14): 1923-1924.
107. Chen X, Bahrami A, Pappo A, Easton J, Dalton J, Hedlund E, Ellison D, Shurtleff S, Wu G, Wei L et al. 2014. Recurrent Somatic Structural Variations Contribute to Tumorigenesis in Pediatric Osteosarcoma. *Cell Reports* **7**(1): 104-112.
108. Chen X, Stewart E, Shelat AA, Qu C, Bahrami A, Hatley M, Wu G, Bradley C, McEvoy J, Pappo A et al. 2013. Targeting Oxidative Stress in Embryonal Rhabdomyosarcoma. *Cancer Cell* **24**(6): 710-724.
109. Chen Y, Hong J, Cui W, Zaneveld J, Wang W, Gibbs R, Xiao Y, Chen R. 2013. CGAP-align: a high performance DNA short read alignment tool *PLoSOne* **8**(4): e61033.
110. Cheng CS, Rai K, Garber M, Hollinger A, Robbins D, Anderson S, Macbeth A, Tzou A, Carneiro MO, Raychowdhury R et al. 2013. Semiconductor-based DNA sequencing of histone modification states. *Nat Commun* **4**: 2672.
111. Cheng SWG, Kuzyk MA, Moradian A, Ichu T-A, Chang VCD, Tien JF, Vollett SE, Griffith M, Marra MA, Morin GB. 2012. Interaction of Cyclin-Dependent Kinase 12/CrkRS with Cyclin K1 Is Required for the Phosphorylation of the C-Terminal Domain of RNA Polymerase II. *Molecular and Cellular Biology* **32**(22): 4691-4704.
112. Cheung N-KV, Zhang J, Lu C, Parker M, Bahrami A, Tickoo SK, Heguy A, Pappo AS, Federico S, Dalton J et al. 2012. Association of Age at Diagnosis and Genetic Mutations in Patients With Neuroblastoma. *Jama-Journal of the American Medical Association* **307**(10): 1062-1071.
113. Cheung YH, Gayden T, Campeau PM, LeDuc CA, Russo D, Nguyen VH, Guo J, Qi M, Guan Y, Albrecht S et al. 2013. A recurrent PDGFRB mutation causes familial infantile myofibromatosis *AmJHumGenet* **92**(6): 996-1000.
114. Chiang HR, Schoenfeld LW, Ruby JG, Auyeung VC, Spies N, Baek D, Johnston WK, Russ C, Luo S, Babiarz JE et al. 2010. Mammalian microRNAs: experimental evaluation of novel and previously annotated genes. *Genes & development* **24**(10): 992-1009.
115. Chmielecki J, Crago AM, Rosenberg M, O'Connor R, Walker SR, Ambrogio L, Auclair D, McKenna A, Heinrich MC, Frank DA et al. 2013. Whole-exome sequencing identifies a recurrent NAB2-STAT6 fusion in solitary fibrous tumors. *Nat Genet* **45**(2): 131-132.
116. Cho J, Bass AJ, Lawrence MS, Cibulskis K, Cho A, Lee SN, Yamauchi M, Wagle N, Pochanard P, Kim N et al. 2014. Colon cancer-derived oncogenic EGFR G724S mutant identified by whole genome sequence analysis is dependent on asymmetric dimerization and sensitive to cetuximab. *Molecular cancer* **13**(1): 141.
117. Church DM, Schneider VA, Graves T, Auger K, Cunningham F, Bouk N, Chen H-C, Agarwala R, McLaren WM, Ritchie GRS et al. 2011. Modernizing Reference Genome Assemblies. *Plos Biology* **9**(7).
118. Cibulskis K, Lawrence MS, Carter SL, Sivachenko A, Jaffe D, Sougnez C, Gabriel S, Meyerson M, Lander ES, Getz G. 2013. Sensitive detection of somatic point mutations in impure and heterogeneous cancer samples. *Nat Biotechnol* **31**(3): 213-219.
119. Clarke L, Zheng-Bradley X, Smith R, Kulesha E, Xiao C, Toneva I, Vaughan B, Preuss D, Leinonen R, Shumway M et al. 2012. The 1000 Genomes Project: data management and community access. *Nat Methods* **9**(5): 459-462.
120. Comuzzie AG, Cole SA, Laston SL, Voruganti VS, Haack K, Gibbs RA, Butte NF. 2012. Novel genetic Loci identified for the pathophysiology of childhood obesity in the Hispanic population. *PLoSOne* **7**(12): e51954.
121. Conrad DF, Keebler JE, DePristo MA, Lindsay SJ, Zhang Y, Casals F, Idaghdour Y, Hartl CL, Torroja C, Garimella KV et al. 2011. Variation in genome-wide mutation rates within and between human families. *Nat Genet* **43**(7): 712-714.
122. Consortium AG. 2010. Genome sequence of the pea aphid Acyrthosiphon pisum *PLoSBiol* **8**(2): e1000313.
123. Consortium BG. 2012. Butterfly genome reveals promiscuous exchange of mimicry adaptations among species *Nature* **487**(7405): 94-98.
124. Consortium HM. 2012. Evaluation of 16S rDNA-based community profiling for human microbiome research. *PLoSOne* **7**(6): e39315.
125. -. 2012. Structure, function and diversity of the healthy human microbiome *Nature* **486**(7402): 207-214.
126. Consortium iK. 2013. The i5K Initiative: advancing arthropod genomics for knowledge, human health, agriculture, and the environment. *JHered* **104**(5): 595-600.
127. Consortium STD, Estrada K, Aukrust I, Bjorkhaug L, Burtt NP, Mercader JM, Garcia-Ortiz H, Huerta-Chagoya A, Moreno-Macias H, Walford G et al. 2014. Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. *JAMA* **311**(22): 2305-2314.
128. Cornes BK, Brody JA, Nikpoor N, Morrison AC, Dang HC, Ahn BS, Wang S, Dauriz M, Barzilay JI, Dupuis J et al. 2014. Association of Levels of Fasting Glucose and Insulin With Rare Variants at the Chromosome 11p11.2-MADD Locus: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Targeted Sequencing Study. *Circulation Cardiovascular genetics* **7**(3): 374-382.
129. Costello M, Pugh TJ, Fennell TJ, Stewart C, Lichtenstein L, Meldrim JC, Fostel JL, Friedrich DC, Perrin D, Dionne D et al. 2013. Discovery and characterization of artifactual mutations in deep coverage targeted capture sequencing data due to oxidative DNA damage during sample preparation. *Nucleic Acids Res* **41**(6): e67.
130. Coventry A, Bull-Otterson LM, Liu X, Clark AG, Maxwell TJ, Crosby J, Hixson JE, Rea TJ, Muzny DM, Lewis LR et al. 2010. Deep resequencing reveals excess rare recent variants consistent with explosive population growth *NatCommun* **1**(8): 131.
131. Creighton CJ, Hernandez-Herrera A, Jacobsen A, Levine DA, Mankoo P, Schultz N, Du Y, Zhang Y, Larsson E, Sheridan R et al. 2012. Integrated analyses of microRNAs demonstrate their widespread influence on gene expression in high-grade serous ovarian carcinoma. *PLoSOne* **7**(3): e34546.
132. Crowley JJ, Hilliard CE, Kim Y, Morgan MB, Lewis LR, Muzny DM, Hawes AC, Sabo A, Wheeler DA, Lieberman JA et al. 2012. Deep resequencing and association analysis of schizophrenia candidate genes. *MolPsychiatry*.
133. Cuomo CA, Birren BW. 2010. The fungal genome initiative and lessons learned from genome sequencing. *Methods in enzymology* **470**: 833-855.
134. Daemen A, Griffith OL, Heiser LM, Wang NJ, Enache OM, Sanborn Z, Pepin F, Durinck S, Korkola JE, Griffith M et al. 2013. Modeling precision treatment of breast cancer. *Genome Biology* **14**(10).
135. Dalzell JJ, McVeigh P, Warnock ND, Mitreva M, Bird DM, Abad P, Fleming CC, Day TA, Mousley A, Marks NJ et al. 2011. RNAi Effector Diversity in Nematodes. *Plos Neglected Tropical Diseases* **5**(6).
136. Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, Handsaker RE, Lunter G, Marth GT, Sherry ST et al. 2011. The variant call format and VCFtools. *Bioinformatics* **27**(15): 2156-2158.
137. Davis EE, Zhang Q, Liu Q, Diplas BH, Davey LM, Hartley J, Stoetzel C, Szymanska K, Ramaswami G, Logan CV et al. 2011. TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum *NatGenet* **43**(3): 189-196.
138. De Graef J, Demeler J, Skuce P, Mitreva M, Von Samson-Himmelstjerna G, Vercruysse J, Claerebout E, Geldhof P. 2013. Gene expression analysis of ABC transporters in a resistant Cooperia oncophora isolate following in vivo and in vitro exposure to macrocyclic lactones. *Parasitology* **140**(4): 499-508.
139. de LJ, Boone PM, Pfundt R, Vissers LE, Richmond T, Geoghegan J, O'Moore K, de LN, Shaw C, Brunner HG et al. 2013. Detection of clinically relevant copy number variants with whole-exome sequencing *HumMutat* **34**(10): 1439-1448.
140. Dees ND, Miller CA, White BS, Schierding W, Vij R, Tomasson MH, Welch JS, Graubert TA, Walter MJ, Ley TJ et al. 2013. Tumor clonality detection using next generation sequencing data. *Cancer Research* **73**(8).
141. Dees ND, Zhang Q, Kandoth C, Wendl MC, Schierding W, Koboldt DC, Mooney TB, Callaway MB, Dooling D, Mardis ER et al. 2012. MuSiC: Identifying mutational significance in cancer genomes. *Genome Research* **22**(8): 1589-1598.
142. DeLuca DS, Levin JZ, Sivachenko A, Fennell T, Nazaire MD, Williams C, Reich M, Winckler W, Getz G. 2012. RNA-SeQC: RNA-seq metrics for quality control and process optimization. *Bioinformatics* **28**(11): 1530-1532.
143. Deng Z, Huang W, Bakkalbasi E, Brown NG, Adamski CJ, Rice K, Muzny D, Gibbs RA, Palzkill T. 2012. Deep Sequencing of Systematic Combinatorial Libraries Reveals beta-Lactamase Sequence Constraints at High Resolution *JMolBiol*.
144. Dennis MY, Nuttle X, Sudmant PH, Antonacci F, Graves TA, Nefedov M, Rosenfeld JA, Sajjadian S, Malig M, Kotkiewicz H et al. 2012. Evolution of Human-Specific Neural SRGAP2 Genes by Incomplete Segmental Duplication. *Cell* **149**(4).
145. DePristo MA, Banks E, Poplin R, Garimella KV, Maguire JR, Hartl C, Philippakis AA, del Angel G, Rivas MA, Hanna M et al. 2011. A framework for variation discovery and genotyping using next-generation DNA sequencing data. *Nat Genet* **43**(5): 491-498.
146. Desjardins CA, Gadau J, Lopez JA, Niehuis O, Avery AR, Loehlin DW, Richards S, Colbourne JK, Werren JH. 2013. Fine-scale mapping of the Nasonia genome to chromosomes using a high-density genotyping microarray *G3(Bethesda)* **3**(2): 205-215.
147. Ding L, Ley TJ, Larson DE, Miller CA, Koboldt DC, Welch JS, Ritchey JK, Young MA, Lamprecht T, McLellan MD et al. 2012. Clonal evolution in relapsed acute myeloid leukaemia revealed by whole-genome sequencing. *Nature* **481**(7382): 506-510.
148. Ding L, Raphael BJ, Chen F, Wendl MC. 2013. Advances for studying clonal evolution in cancer. *Cancer Letters* **340**(2): 212-219.
149. Ding L, Wendl MC. 2013. Differences that matter in cancer genomics. *Nature Biotechnology* **31**(10): 892-893.
150. Ding L, Wendl MC, Koboldt DC, Mardis ER. 2010. Analysis of next-generation genomic data in cancer: accomplishments and challenges. *Human Molecular Genetics* **19**: R188-R196.
151. Diogo D, Liao KP, Fulton RS, Graham RR, Cui J, Greenberg JD, Eyre S, Bowes J, Lee AT, Pappas DA et al. 2013. Potential Of Integrating Human Genetics and Electronic Medical Records For Drug Discovery: The Example Of TYK2 and Rheumatoid Arthritis. *Arthritis and Rheumatism* **65**: S1234-S1234.
152. Dixon-Salazar TJ, Silhavy JL, Udpa N, Schroth J, Bielas S, Schaffer AE, Olvera J, Bafna V, Zaki MS, Abdel-Salam GH et al. 2012. Exome sequencing can improve diagnosis and alter patient management. *Science translational medicine* **4**(138): 138ra178.
153. Dodman NH, Karlsson EK, Moon-Fanelli A, Galdzicka M, Perloski M, Shuster L, Lindblad-Toh K, Ginns EI. 2010. A canine chromosome 7 locus confers compulsive disorder susceptibility. *Molecular psychiatry* **15**(1): 8-10.
154. Donehower LA, Creighton CJ, Schultz N, Shinbrot E, Chang K, Gunaratne PH, Muzny D, Sander C, Hamilton SR, Gibbs RA et al. 2012. MLH1-Silenced and Non-Silenced Subgroups of Hypermutated Colorectal Carcinomas Have Distinct Mutational Landscapes. *JPathol*.
155. Downing JR, Wilson RK, Zhang J, Mardis ER, Pui C-H, Ding L, Ley TJ, Evans WE. 2012. The Pediatric Cancer Genome Project. *Nature Genetics* **44**(6): 619-622.
156. Doyle AJ, Doyle JJ, Bessling SL, Maragh S, Lindsay ME, Schepers D, Gillis E, Mortier G, Homfray T, Sauls K et al. 2012. Mutations in the TGF-beta repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. *NatGenet* **44**(11): 1249-1254.
157. Drier Y, Lawrence MS, Carter SL, Stewart C, Gabriel SB, Lander ES, Meyerson M, Beroukhim R, Getz G. 2013. Somatic rearrangements across cancer reveal classes of samples with distinct patterns of DNA breakage and rearrangement-induced hypermutability. *Genome Res* **23**(2): 228-235.
158. D'Souza CA, Kronstad JW, Taylor G, Warren R, Yuen M, Hu G, Jung WH, Sham A, Kidd SE, Tangen K et al. 2011. Genome variation in Cryptococcus gattii, an emerging pathogen of immunocompetent hosts. *mBio* **2**(1): e00342-00310.
159. Dulak AM, Schumacher SE, van Lieshout J, Imamura Y, Fox C, Shim B, Ramos AH, Saksena G, Baca SC, Baselga J et al. 2012. Gastrointestinal adenocarcinomas of the esophagus, stomach, and colon exhibit distinct patterns of genome instability and oncogenesis. *Cancer Res* **72**(17): 4383-4393.
160. Dulak AM, Stojanov P, Peng S, Lawrence MS, Fox C, Stewart C, Bandla S, Imamura Y, Schumacher SE, Shefler E et al. 2013. Exome and whole-genome sequencing of esophageal adenocarcinoma identifies recurrent driver events and mutational complexity. *Nat Genet* **45**(5): 478-486.
161. Duncavage EJ, Armstrong JR, Magrini VJ, Becker N, Demeter R, Marids ER, Pfeifer JD. 2010. Viral Insertion Site Discovery Using Next Generation Sequencing of Formalin Fixed Tissue. *Modern Pathology* **23**: 423A-423A.
162. Earl AM, Desjardins CA, Fitzgerald MG, Arachchi HM, Zeng Q, Mehta T, Griggs A, Birren BW, Toney NC, Carr J et al. 2011. High quality draft genome sequence of Segniliparus rugosus CDC 945(T)= (ATCC BAA-974(T)). *Stand Genomic Sci* **5**(3): 389-397.
163. Earl D, Bradnam K, St John J, Darling A, Lin D, Fass J, Yu HO, Buffalo V, Zerbino DR, Diekhans M et al. 2011. Assemblathon 1: a competitive assessment of de novo short read assembly methods. *Genome Res* **21**(12): 2224-2241.
164. Eckalbar WL, Hutchins ED, Markov GJ, Allen AN, Corneveaux JJ, Lindblad-Toh K, Di Palma F, Alfoldi J, Huentelman MJ, Kusumi K. 2013. Genome reannotation of the lizard Anolis carolinensis based on 14 adult and embryonic deep transcriptomes. *BMC Genomics* **14**: 49.
165. Ellis MJ, Ding L, Shen D, Luo J, Suman VJ, Goiffon RJ, Wallis JW, Goldstein TC, Chen K, Allred DC et al. 2012. Whole genome sequencing to characterize luminal-type breast cancer. *Journal of Clinical Oncology* **30**(15).
166. Ellis MJ, Ding L, Shen D, Luo J, Suman VJ, Wallis JW, Van Tine BA, Hoog J, Goiffon RJ, Goldstein TC et al. 2012. Whole-genome analysis informs breast cancer response to aromatase inhibition. *Nature* **486**(7403): 353-360.
167. Ellis MJ, Li S, Shen D, Ding L, Crowder R, Shao J, Goncalves R, Tao Y, Luo J, Prat A et al. 2013. Patient-derived xenografts from advanced luminal-type breast cancer: insights into endocrine therapy resistance. *Cancer Research* **73**(8).
168. Elsik CG, Worley KC, Bennett AK, Beye M, Camara F, Childers CP, de Graaf DC, Debyser G, Deng J, Devreese B et al. 2014. Finding the missing honey bee genes: lessons learned from a genome upgrade. *BMCGenomics* **15**: 86.
169. English AC, Richards S, Han Y, Wang M, Vee V, Qu J, Qin X, Muzny DM, Reid JG, Worley KC et al. 2012. Mind the Gap: Upgrading Genomes with Pacific Biosciences RS Long-Read Sequencing Technology *PLoSOne* **7**(11): e47768.
170. English AC, Salerno WJD, Reid JGD. 2014. PBHoney: Identifying Genomic Variants via Long-Read Discordance and Interrupted Mapping. *BMC Bioinformatics* **15**(1): 180.
171. Enns GM, Shashi V, Bainbridge M, Gambello MJ, Zahir FR, Bast T, Crimian R, Schoch K, Platt J, Cox R et al. 2014. Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway *GenetMed*.
172. Ewing AD, Ballinger TJ, Earl D, Harris CC, Ding L, Wilson RK, Haussler D, Broad Inst Genome Sequencing A. 2013. Retrotransposition of gene transcripts leads to structural variation in mammalian genomes. *Genome Biology* **14**(3).
173. Fairclough SR, Chen Z, Kramer E, Zeng Q, Young S, Robertson HM, Begovic E, Richter DJ, Russ C, Westbrook MJ et al. 2013. Premetazoan genome evolution and the regulation of cell differentiation in the choanoflagellate Salpingoeca rosetta. *Genome Biol* **14**(2): R15.
174. Fairfield H, Gilbert GJ, Barter M, Corrigan RR, Curtain M, Ding Y, D'Ascenzo M, Gerhardt DJ, He C, Huang W et al. 2011. Mutation discovery in mice by whole exome sequencing. *Genome Biol* **12**(9): R86.
175. Fang Y, Yao Q, Chen Z, Xiang J, William FE, Gibbs RA, Chen C. 2013. Genetic and molecular alterations in pancreatic cancer: implications for personalized medicine *MedSciMonit* **19**: 916-926.
176. Faust K, Sathirapongsasuti JF, Izard J, Segata N, Gevers D, Raes J, Huttenhower C. 2012. Microbial co-occurrence relationships in the human microbiome. *PLoS Comput Biol* **8**(7): e1002606.
177. Fawcett GL, Raveendran M, Rio DD, Chen D, Yu F, Harris RA, Ren Y, Muzny DM, Reid JG, Wheeler DA et al. 2011. Characterization of single-nucleotide variation in Indian-origin Rhesus Macaques(Macaca mulatta) *BMCGenomics* **12**(1): 311.
178. Fehniger TA, Wylie T, Germino E, Leong JW, Magrini VJ, Koul S, Keppel CR, Schneider SE, Koboldt DC, Sullivan RP et al. 2010. Next-generation sequencing identifies the natural killer cell microRNA transcriptome. *Genome Research* **20**(11): 1590-1604.
179. -. 2010. The NK Cell MicroRNA Transcriptome Defined by Next-Generation Sequencing Identifies IL-15-Signaled Alterations In Mature MiR-223 Expression, and MiR-223 as a Potential Regulator of Murine Granzyme B. *Blood* **116**(21): 51-52.
180. Fekete R, Bainbridge M, Baizabal-Carvallo JF, Rivera A, Miller B, Du P, Kholodovych V, Powell S, Ondo W. 2013. Exome sequencing in familial corticobasal degeneration *ParkinsonismRelat Disord* **19**(11): 1049-1052.
181. Fisher DAC, Miller CA, McLellan MD, Fulton RS, Martin KL, Kaiwar C, Moore DM, Ley TJ, Oh ST. 2012. Clonal Evolution Revealed by Whole Genome Sequencing in a Case of Primary Myelofibrosis Transformed to Secondary Acute Myeloid Leukemia. *Blood* **120**(21).
182. Fisher S, Barry A, Abreu J, Minie B, Nolan J, Delorey TM, Young G, Fennell TJ, Allen A, Ambrogio L et al. 2011. A scalable, fully automated process for construction of sequence-ready human exome targeted capture libraries. *Genome Biol* **12**(1): R1.
183. Flannick J, Beer NL, Bick AG, Agarwala V, Molnes J, Gupta N, Burtt NP, Florez JC, Meigs JB, Taylor H et al. 2013. Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. *Nat Genet* **45**(11): 1380-1385.
184. Flannick J, Thorleifsson G, Beer NL, Jacobs SB, Grarup N, Burtt NP, Mahajan A, Fuchsberger C, Atzmon G, Benediktsson R et al. 2014. Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. *Nat Genet* **46**(4): 357-363.
185. Fodor AA, DeSantis TZ, Wylie KM, Badger JH, Ye Y, Hepburn T, Hu P, Sodergren E, Liolios K, Huot-Creasy H et al. 2012. The "most wanted" taxa from the human microbiome for whole genome sequencing. *PLoS One* **7**(7): e41294.
186. Fondon JW, III, Martin A, Richards S, Gibbs RA, Mittelman D. 2012. Analysis of microsatellite variation in Drosophila melanogaster with population-scale genome sequencing *PLoSOne* **7**(3): e33036.
187. Forgetta V, Oughton MT, Marquis P, Brukner I, Blanchette R, Haub K, Magrini V, Mardis ER, Gerding DN, Loo VG et al. 2011. Fourteen-Genome Comparison Identifies DNA Markers for Severe-Disease-Associated Strains of Clostridium difficile. *Journal of Clinical Microbiology* **49**(6): 2230-2238.
188. Friedman J, Olvera J, Silhavy JL, Gabriel SB, Gleeson JG. 2012. Mild paroxysmal kinesigenic dyskinesia caused by PRRT2 missense mutation with reduced penetrance. *Neurology* **79**(9): 946-948.
189. Galloway-Pena JR, Bourgogne A, Qin X, Murray BE. 2011. Diversity of the fsr-gelE region of the Enterococcus faecalis genome but conservation in strains with partial deletions of the fsr operon *ApplEnvironMicrobiol* **77**(2): 442-451.
190. Gandolfi B, Alhaddad H, Affolter VK, Brockman J, Haggstrom J, Joslin SEK, Koehne AL, Mullikin JC, Outerbridge CA, Warren WC et al. 2013. To the Root of the Curl: A Signature of a Recent Selective Sweep Identifies a Mutation That Defines the Cornish Rex Cat Breed. *Plos One* **8**(6).
191. Gao X, Wang Z, Martin J, Abubucker S, Zhang X, Mitreva M, Hawdon JM. 2010. Identification of Hookworm DAF-16/FOXO Response Elements and Direct Gene Targets. *Plos One* **5**(8).
192. Garber M, Yosef N, Goren A, Raychowdhury R, Thielke A, Guttman M, Robinson J, Minie B, Chevrier N, Itzhaki Z et al. 2012. A high-throughput chromatin immunoprecipitation approach reveals principles of dynamic gene regulation in mammals. *Molecular cell* **47**(5): 810-822.
193. Gargis AS, Kalman L, Berry MW, Bick DP, Dimmock DP, Hambuch T, Lu F, Lyon E, Voelkerding KV, Zehnbauer BA et al. 2012. Assuring the quality of next-generation sequencing in clinical laboratory practice. *Nature Biotechnology* **30**(11): 1033-1036.
194. Garraway LA, Lander ES. 2013. Lessons from the cancer genome. *Cell* **153**(1): 17-37.
195. Gazave E, Ma L, Chang D, Coventry A, Gao F, Muzny D, Boerwinkle E, Gibbs RA, Sing CF, Clark AG et al. 2014. Neutral genomic regions refine models of recent rapid human population growth *ProcNatlAcadSciUSA* **111**(2): 757-762.
196. Genomes Project C, Abecasis GR, Altshuler D, Auton A, Brooks LD, Durbin RM, Gibbs RA, Hurles ME, McVean GA. 2010. A map of human genome variation from population-scale sequencing. *Nature* **467**(7319): 1061-1073.
197. Genomes Project C, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA. 2012. An integrated map of genetic variation from 1,092 human genomes. *Nature* **491**(7422): 56-65.
198. Gevers D, Knight R, Petrosino JF, Huang K, McGuire AL, Birren BW, Nelson KE, White O, Methe BA, Huttenhower C. 2012. The Human Microbiome Project: a community resource for the healthy human microbiome. *PLoS Biol* **10**(8): e1001377.
199. Gevers D, Pop M, Schloss PD, Huttenhower C. 2012. Bioinformatics for the Human Microbiome Project. *PLoS Comput Biol* **8**(11): e1002779.
200. Giannoukos G, Ciulla DM, Huang K, Haas BJ, Izard J, Levin JZ, Livny J, Earl AM, Gevers D, Ward DV et al. 2012. Efficient and robust RNA-seq process for cultured bacteria and complex community transcriptomes. *Genome Biol* **13**(3): R23.
201. Gibbs RA. 2011. Genome-sequencing anniversary. Bringing genomics and genetics back together *Science* **331**(6017): 548.
202. Gibbs RA, Rogers J. 2012. Genomics: Gorilla gorilla gorilla. *Nature* **483**(7388): 164-165.
203. Gnerre S, Maccallum I, Przybylski D, Ribeiro FJ, Burton JN, Walker BJ, Sharpe T, Hall G, Shea TP, Sykes S et al. 2011. High-quality draft assemblies of mammalian genomes from massively parallel sequence data. *Proc Natl Acad Sci U S A* **108**(4): 1513-1518.
204. Goldsmith JD, Allred DC, Beasley MB, Eisen R, Fulton RS, Gown AM, Hammond MEH. 2011. Fixation Time Does Not Affect Expression of HER2/neu. *American Journal of Clinical Pathology* **135**(3): 484-484.
205. Goldstein JI, Crenshaw A, Carey J, Grant GB, Maguire J, Fromer M, O'Dushlaine C, Moran JL, Chambert K, Stevens C et al. 2012. zCall: a rare variant caller for array-based genotyping: genetics and population analysis. *Bioinformatics* **28**(19): 2543-2545.
206. Gonzaga-Jauregui C, Lotze T, Jamal L, Penney S, Campbell IM, Pehlivan D, Hunter JV, Woodbury SL, Raymond G, Adesina AM et al. 2013. Mutations in VRK1 Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly *JAMA Neurol*.
207. Gonzaga-Jauregui C, Lupski JR, Gibbs RA. 2012. Human genome sequencing in health and disease. *AnnuRevMed* **63**: 35-61.
208. Gonzaga-Jauregui C, Mir S, Penney S, Jhangiani S, Midgen C, Finegold M, Muzny DM, Wang M, Bacino CA, Genomics BH et al. 2014. Whole-Exome Sequencing Reveals GPIHBP1 Mutations in A Case of Infantile Colitis with Severe Hypertriglyceridemia *JPediatrGastroenterolNutr*.
209. Gonzalez-Perez A, Mustonen V, Reva B, Ritchie GRS, Creixell P, Karchin R, Vazquez M, Fink JL, Kassahn KS, Pearson JV et al. 2013. Computational approaches to identify functional genetic variants in cancer genomes. *Nature Methods* **10**(8): 723-729.
210. Gordon A, Smith J, Metzker ML, Gibbs R, Mardis ER, Nickerson DA, Fulton R, Scherer S. 2012. PHARMACOGENETIC APPLICATIONS OF NEXT-GENERATION SEQUENCING. *Drug Metabolism Reviews* **44**: 24-25.
211. Govindan R, Ding L, Griffith M, Subramanian J, Dees ND, Kanchi KL, Maher CA, Fulton R, Fulton L, Wallis J et al. 2012. Genomic Landscape of Non-Small Cell Lung Cancer in Smokers and Never-Smokers. *Cell* **150**(6): 1121-1134.
212. Grabherr MG, Haas BJ, Yassour M, Levin JZ, Thompson DA, Amit I, Adiconis X, Fan L, Raychowdhury R, Zeng Q et al. 2011. Full-length transcriptome assembly from RNA-Seq data without a reference genome. *Nat Biotechnol* **29**(7): 644-652.
213. Grabherr MG, Pontiller J, Mauceli E, Ernst W, Baumann M, Biagi T, Swofford R, Russell P, Zody MC, Di Palma F et al. 2011. Exploiting nucleotide composition to engineer promoters. *PLoS One* **6**(5): e20136.
214. Grabherr MG, Russell P, Meyer M, Mauceli E, Alfoldi J, Di Palma F, Lindblad-Toh K. 2010. Genome-wide synteny through highly sensitive sequence alignment: Satsuma. *Bioinformatics* **26**(9): 1145-1151.
215. Grad YH, Lipsitch M, Feldgarden M, Arachchi HM, Cerqueira GC, Fitzgerald M, Godfrey P, Haas BJ, Murphy CI, Russ C et al. 2012. Genomic epidemiology of the Escherichia coli O104:H4 outbreaks in Europe, 2011. *Proc Natl Acad Sci U S A* **109**(8): 3065-3070.
216. Grad YH, Lipsitch M, Griggs AD, Haas BJ, Shea TP, McCowan C, Montmayeur A, FitzGerald M, Wortman JR, Krogfelt KA et al. 2012. Reply to Guy et al.: Support for a bottleneck in the 2011 Escherichia coli O104:H4 outbreak in Germany. *Proc Natl Acad Sci U S A* **109**(52): E3629-3630.
217. Graubert T, Ding L, Walter M, Larson D, Shen D, Dipersio J, Mardis E, Wilson R, Ley T. 2011. Discovery of Novel Mutations in Myelodysplastic Syndrome and Acute Myeloid Leukemia by whole Genome Sequencing. *Annals of Hematology* **90**: S49-S50.
218. Graubert TA, Mardis ER. 2011. Genomics of Acute Myeloid Leukemia. *Cancer Journal* **17**(6): 487-491.
219. Graubert TA, Shen D, Ding L, Okeyo-Owuor T, Lunn CL, Shao J, Krysiak K, Harris CC, Koboldt DC, Larson DE et al. 2012. Recurrent mutations in the U2AF1 splicing factor in myelodysplastic syndromes. *Nature Genetics* **44**(1): 53-U77.
220. Gravel S, Henn BM, Gutenkunst RN, Indap AR, Marth GT, Clark AG, Yu F, Gibbs RA, Bustamante CD. 2011. Demographic history and rare allele sharing among human populations *ProcNatlAcadSciUSA* **108**(29): 11983-11988.
221. Green RC, Lupski JR, Biesecker LG. 2013. Reporting genomic sequencing results to ordering clinicians: incidental, but not exceptional. *JAMA* **310**(4): 365-366.
222. Green RE, Krause J, Briggs AW, Maricic T, Stenzel U, Kircher M, Patterson N, Li H, Zhai W, Fritz MH et al. 2010. A draft sequence of the Neandertal genome. *Science* **328**(5979): 710-722.
223. Greulich H, Kaplan B, Mertins P, Chen TH, Tanaka KE, Yun CH, Zhang X, Lee SH, Cho J, Ambrogio L et al. 2012. Functional analysis of receptor tyrosine kinase mutations in lung cancer identifies oncogenic extracellular domain mutations of ERBB2. *Proc Natl Acad Sci U S A* **109**(36): 14476-14481.
224. Grewal PS, Bai X, Adams BJ, Ciche TA, Clifton S, Gaugler R, Kim K-s, Spieth J, Sternberg PW, Wilson RK. 2013. FIRST COMPLETE GENOME SEQUENCE OF AN ENTOMOPATHOGENIC NEMATODE GOES PUBLIC. *Journal of Nematology* **45**(4): 291-291.
225. Griffith M, Griffith OL, Coffman AC, Weible JV, McMichael JF, Spies NC, Koval J, Das I, Callaway MB, Eldred JM et al. 2013. DGIdb: mining the druggable genome. *Nature Methods* **10**(12): 1209-+.
226. Griffith OL, Pepin F, Enache OM, Heiser LM, Collisson EA, Spellman PT, Gray JW. 2013. A robust prognostic signature for hormone-positive node-negative breast cancer. *Genome Medicine* **5**.
227. Groenen MAM, Megens H-J, Zare Y, Warren WC, Hillier LW, Crooijmans RPMA, Vereijken A, Okimoto R, Muir WM, Cheng HH. 2011. The development and characterization of a 60K SNP chip for chicken. *Bmc Genomics* **12**.
228. Gronowski AM, Manson JE, Mardis ER, Mora S, Spong CY. 2014. What's Different about Women's Health? *Clinical Chemistry* **60**(1): 1-3.
229. Grover M, Campeau PM, Lietman CD, Lu JT, Gibbs RA, Schlesinger AE, Lee BH. 2013. Osteogenesis imperfecta without features of type V caused by a mutation in the IFITM5 gene. *JBone MinerRes*.
230. Gruber TA, Gedman AL, Ta HQ, Zhang J, Koss C, Chen S-C, Su X, Gupta V, Ogden S, Andersson AK et al. 2011. Transcriptome Sequence Analysis. of Pediatric Acute Megakaryoblastic Leukemia Identifies An Inv(16)(p13.3;q24.3)-Encoded CBFA2T3-GLIS2 Fusion Protein As a Recurrent Lesion in 39% of Non-Infant Cases: A Report From the St. Jude Children's Research Hospital - Washington University Pediatric Cancer Genome Project. *Blood* **118**(21): 344-344.
231. Gruber TA, Gedman AL, Zhang J, Koss CS, Marada S, Ta HQ, Chen S-C, Su X, Ogden SK, Dang J et al. 2012. An Inv(16)(p13.3q24.3)-Encoded CBFA2T3-GLIS2 Fusion Protein Defines an Aggressive Subtype of Pediatric Acute Megakaryoblastic Leukemia. *Cancer Cell* **22**(5): 683-697.
232. Gu H, Bock C, Mikkelsen TS, Jager N, Smith ZD, Tomazou E, Gnirke A, Lander ES, Meissner A. 2010. Genome-scale DNA methylation mapping of clinical samples at single-nucleotide resolution. *Nat Methods* **7**(2): 133-136.
233. Gutmann DH, McLellan MD, Hussain I, Wallis JW, Fulton LL, Fulton RS, Magrini V, Demeter R, Wylie T, Kandoth C et al. 2013. Somatic neurofibromatosis type 1 (NF1) inactivation characterizes NF1-associated pilocytic astrocytoma. *Genome Research* **23**(3): 431-439.
234. Guttman M, Donaghey J, Carey BW, Garber M, Grenier JK, Munson G, Young G, Lucas AB, Ach R, Bruhn L et al. 2011. lincRNAs act in the circuitry controlling pluripotency and differentiation. *Nature* **477**(7364): 295-300.
235. Guttman M, Garber M, Levin JZ, Donaghey J, Robinson J, Adiconis X, Fan L, Koziol MJ, Gnirke A, Nusbaum C et al. 2010. Ab initio reconstruction of cell type-specific transcriptomes in mouse reveals the conserved multi-exonic structure of lincRNAs. *Nat Biotechnol* **28**(5): 503-510.
236. Haas BJ, Chin M, Nusbaum C, Birren BW, Livny J. 2012. How deep is deep enough for RNA-Seq profiling of bacterial transcriptomes? *BMC Genomics* **13**: 734.
237. Haas BJ, Gevers D, Earl AM, Feldgarden M, Ward DV, Giannoukos G, Ciulla D, Tabbaa D, Highlander SK, Sodergren E et al. 2011. Chimeric 16S rRNA sequence formation and detection in Sanger and 454-pyrosequenced PCR amplicons. *Genome Res* **21**(3): 494-504.
238. Haas BJ, Zeng Q, Pearson MD, Cuomo CA, Wortman JR. 2011. Approaches to Fungal Genome Annotation. *Mycology* **2**(3): 118-141.
239. Hamilton MP, Rajapakshe K, Hartig SM, Reva B, McLellan MD, Kandoth C, Ding L, Zack TI, Gunaratne PH, Wheeler DA et al. 2013. Identification of a pan-cancer oncogenic microRNA superfamily anchored by a central core seed motif. *Nature Communications* **4**.
240. Hamilton PJ, Campbell NG, Sharma S, Erreger K, Herborg HF, Saunders C, Belovich AN, Sahai MA, Cook EH, Gether U et al. 2013. De novo mutation in the dopamine transporter gene associates dopamine dysfunction with autism spectrum disorder *MolPsychiatry* **18**(12): 1315-1323.
241. Hammerman PS Lawrence MS Voet D Jing R Cibulskis K Sivachenko A Stojanov P McKenna A Lander ES Gabriel S et al. 2012. Comprehensive genomic characterization of squamous cell lung cancers. *Nature* **489**(7417): 519-525.
242. Hamosh A, Sobreira N, Hoover-Fong J, Sutton VR, Boehm C, Schiettecatte F, Valle D. 2013. PhenoDB: a new web-based tool for the collection, storage, and analysis of phenotypic features *HumMutat* **34**(4): 566-571.
243. Hanchard N, Murdock D, Magoulas P, Bainbridge M, Muzny D, Wu Y, Wang M, McGuire A, Lupski J, Gibbs R et al. 2012. Exploring the utility of whole-exome sequencing as a diagnostic tool in a child with atypical episodic muscle weakness *ClinGenet*.
244. Hao Y-J, Montiel R, Abubucker S, Mitreva M, Simoes N. 2010. Transcripts analysis of the entomopathogenic nematode Steinernema carpocapsae induced in vitro with insect haemolymph. *Molecular and Biochemical Parasitology* **169**(2): 79-86.
245. Harring TR, Guiteau JJ, Nguyen NT, Cotton RT, Gingras MC, Wheeler DA, O'Mahony CA, Gibbs RA, Brunicardi FC, Goss JA. 2011. Building a comprehensive genomic program for hepatocellular carcinoma. *World JSurg* **35**(8): 1746-1750.
246. Harris RA, Tardif SD, Vinar T, Wildman DE, Rutherford JN, Rogers J, Worley KC, Aagaard KM. 2013. Evolutionary genetics and implications of small size and twinning in callitrichine primates *ProcNatlAcadSciUSA*.
247. Hartig G, Churakov G, Warren WC, Brosius J, Makalowski W, Schmitz J. 2013. Retrophylogenomics Place Tarsiers on the Evolutionary Branch of Anthropoids. *Scientific Reports* **3**.
248. He X, Sanders SJ, Liu L, De RS, Lim ET, Sutcliffe JS, Schellenberg GD, Gibbs RA, Daly MJ, Buxbaum JD et al. 2013. Integrated model of de novo and inherited genetic variants yields greater power to identify risk genes *PLoSGenet* **9**(8): e1003671.
249. Heizer E, Zarlenga DS, Rosa B, Gao X, Gasser RB, De Graef J, Geldhof P, Mitreva M. 2013. Transcriptome analyses reveal protein and domain families that delineate stage-related development in the economically important parasitic nematodes, Ostertagia ostertagi and Cooperia oncophora. *Bmc Genomics* **14**.
250. Hellsten U, Harland RM, Gilchrist MJ, Hendrix D, Jurka J, Kapitonov V, Ovcharenko I, Putnam NH, Shu S, Taher L et al. 2010. The Genome of the Western Clawed Frog Xenopus tropicalis. *Science* **328**(5978): 633-636.
251. Hernandez RD, Kelley JL, Elyashiv E, Melton SC, Auton A, McVean G, Sella G, Przeworski M. 2011. Classic selective sweeps were rare in recent human evolution. *Science* **331**(6019): 920-924.
252. Hjeij R, Lindstrand A, Francis R, Zariwala MA, Liu X, Li Y, Damerla R, Dougherty GW, Abouhamed M, Olbrich H et al. 2013. ARMC4 mutations cause primary ciliary dyskinesia with randomization of left/right body asymmetry *AmJHumGenet* **93**(2): 357-367.
253. Hodis E, Watson IR, Kryukov GV, Arold ST, Imielinski M, Theurillat JP, Nickerson E, Auclair D, Li L, Place C et al. 2012. A landscape of driver mutations in melanoma. *Cell* **150**(2): 251-263.
254. Hoeppner MP, Lundquist A, Pirun M, Meadows JR, Zamani N, Johnson J, Sundstrom G, Cook A, FitzGerald MG, Swofford R et al. 2014. An improved canine genome and a comprehensive catalogue of coding genes and non-coding transcripts. *PLoS One* **9**(3): e91172.
255. Holmfeldt L, Wei L, Diaz-Flores E, Walsh M, Zhang J, Ding L, Payne-Turner D, Churchman M, Andersson A, Chen S-C et al. 2013. The genomic landscape of hypodiploid acute lymphoblastic leukemia. *Nature Genetics* **45**(3): 242-252.
256. Hong LZ, Li J, Schmidt-Kuentzel A, Warren WC, Barsh GS. 2011. Digital gene expression for non-model organisms. *Genome Research* **21**(11): 1905-1915.
257. Huang W, Massouras A, Inoue Y, Peiffer J, Ramia M, Tarone A, Turlapati L, Zichner T, Zhu D, Lyman R et al. 2014. Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines *Genome Res*.
258. Huang W, Richards S, Carbone MA, Zhu D, Anholt RR, Ayroles JF, Duncan L, Jordan KW, Lawrence F, Magwire MM et al. 2012. Epistasis dominates the genetic architecture of Drosophila quantitative traits *ProcNatlAcadSciUSA* **109**(39): 15553-15559.
259. Huang Y, Li Y, Burt DW, Chen H, Zhang Y, Qian W, Kim H, Gan S, Zhao Y, Li J et al. 2013. The duck genome and transcriptome provide insight into an avian influenza virus reservoir species. *Nature Genetics* **45**(7): 776-+.
260. Huddleston J, Ranade S, Malig M, Antonacci F, Chaisson M, Hon L, Sudmant PH, Graves TA, Alkan C, Dennis MY et al. 2014. Reconstructing complex regions of genomes using long-read sequencing technology. *Genome Research* **24**(4): 688-696.
261. Hudson TJ Anderson W Aretz A Barker AD Bell C Bernabe RR Bhan MK Calvo F Eerola I Gerhard DS et al. 2010. International network of cancer genome projects. *Nature* **464**(7291): 993-998.
262. Huether R, Dong L, Chen X, Wu G, Parker M, Wei L, Ma J, Edmonson MN, Hedlund EK, Rusch MC et al. 2014. The landscape of somatic mutations in epigenetic regulators across 1,000 paediatric cancer genomes. *Nature Communications* **5**.
263. Hughes JF, Skaletsky H, Bellott DW, Chowdhary BP, Warren WC, Worley KC, Wilson RK, Gibbs RA, Page DC. 2013. No bull: Upholding community standards in public sharing of biological datasets. *Proceedings of the National Academy of Sciences of the United States of America* **110**(46): E4277-E4277.
264. Hughes JF, Skaletsky H, Brown LG, Pyntikova T, Graves T, Fulton RS, Dugan S, Ding Y, Buhay CJ, Kremitzki C et al. 2012. Strict evolutionary conservation followed rapid gene loss on human and rhesus Y chromosomes *Nature*.
265. Hughes JF, Skaletsky H, Brown LG, Pyntikova T, Graves T, Fulton RS, Dugan S, Ding Y, Buhay CJ, Kremitzki C et al. 2012. Strict evolutionary conservation followed rapid gene loss on human and rhesus Y chromosomes. *Nature* **483**(7387): 82-U124.
266. Hughes JF, Skaletsky H, Pyntikova T, Graves TA, van Daalen SKM, Minx PJ, Fulton RS, McGrath SD, Locke DP, Friedman C et al. 2010. Chimpanzee and human Y chromosomes are remarkably divergent in structure and gene content. *Nature* **463**(7280): 536-539.
267. Human Microbiome Jumpstart Reference Strains C, Nelson KE, Weinstock GM, Highlander SK, Worley KC, Creasy HH, Wortman JR, Rusch DB, Mitreva M, Sodergren E et al. 2010. A catalog of reference genomes from the human microbiome. *Science* **328**(5981): 994-999.
268. Human Microbiome Project C. 2012. A framework for human microbiome research. *Nature* **486**(7402): 215-221.
269. Imielinski M, Berger AH, Hammerman PS, Hernandez B, Pugh TJ, Hodis E, Cho J, Suh J, Capelletti M, Sivachenko A et al. 2012. Mapping the hallmarks of lung adenocarcinoma with massively parallel sequencing. *Cell* **150**(6): 1107-1120.
270. Iossifov I, Ronemus M, Levy D, Wang Z, Hakker I, Rosenbaum J, Yamrom B, Lee Y-h, Narzisi G, Leotta A et al. 2012. De Novo Gene Disruptions in Children on the Autistic Spectrum. *Neuron* **74**(2): 285-299.
271. Iraqi FA Mahajne M Salaymah Y Sandovski H Tayem H Vered K Balmer L Hall M Manship G Morahan G et al. 2012. The Genome Architecture of the Collaborative Cross Mouse Genetic Reference Population. *Genetics* **190**(2): 389-U159.
272. Ito K, Bick AG, Flannick J, Friedman DJ, Genovese G, Parfenov MG, Depalma SR, Gupta N, Gabriel SB, Taylor HA, Jr. et al. 2014. Increased burden of cardiovascular disease in carriers of APOL1 genetic variants. *Circulation research* **114**(5): 845-850.
273. Itsara A, Vissers LELM, Steinberg KM, Meyer KJ, Zody MC, Koolen DA, de Ligt J, Cuppen E, Baker C, Lee C et al. 2012. Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. *American Journal of Human Genetics* **90**(4): 599-613.
274. Jacoby MA, De Jesus Pizarro RE, Shao J, Koboldt DC, Fulton RS, Zhou G, Wilson RK, Walter MJ. 2014. The DNA double-strand break response is abnormal in myeloblasts from patients with therapy-related acute myeloid leukemia. *Leukemia* **28**(6): 1242-1251.
275. Jasinska AJ, Huang Y, Schmitt C, Jung Y, Svardal H, Wasserscheid J, Juretic N, Dewar K, Grobler P, Jacquelin B et al. 2013. SNP DISCOVERY IN AFRICAN GREEN MONKEY (CHLOROCEBUS AETHIOPS) THROUGH WHOLE GENOME SEQUENCING. *American Journal of Primatology* **75**: 68-68.
276. Jasinska AJ, Lin MK, Service S, Choi O-W, DeYoung J, Grujic O, Kong S-Y, Jung Y, Jorgensen MJ, Fairbanks LA et al. 2012. A non-human primate system for large-scale genetic studies of complex traits. *Human Molecular Genetics* **21**(15): 3307-3316.
277. Jasinska AJ, Ma D, Nam T, Schmitt C, Choi O-w, DeYoung J, Jung Y, Coppola G, Turner T, Grobler P et al. 2013. TRANSCRIPTOMIC CHARACTERIZATION OF AN AGM EXPOSED SERONEGATIVE MODEL. *Journal of Medical Primatology* **42**(5): 277-277.
278. Jasinska AJ, Schmitt CA, Ma D, Huang Y, Svardal H, Wassercheid J, Grobler P, Jorensen M, Muller-Trutwin M, Antonio M et al. 2014. Biological resources for genomic investigation in vervet monkey (Chlorocebus). *American Journal of Physical Anthropology* **153**: 151-152.
279. Jasinska AJ, Schmitt CA, Service SK, Cantor RM, Dewar K, Jentsch JD, Kaplan JR, Turner TR, Warren WC, Weinstock GM et al. 2013. Systems Biology of the Vervet Monkey. *Ilar Journal* **54**(2): 122-143.
280. Jiang RH, de Bruijn I, Haas BJ, Belmonte R, Lobach L, Christie J, van den Ackerveken G, Bottin A, Bulone V, Diaz-Moreno SM et al. 2013. Distinctive expansion of potential virulence genes in the genome of the oomycete fish pathogen Saprolegnia parasitica. *PLoS Genet* **9**(6): e1003272.
281. Jiang Y, Xie M, Chen W, Talbot R, Maddox JF, Faraut T, Wu C, Muzny DM, Li Y, Zhang W et al. 2014. The sheep genome illuminates biology of the rumen and lipid metabolism. *Science* **344**(6188): 1168-1173.
282. Jones FC, Grabherr MG, Chan YF, Russell P, Mauceli E, Johnson J, Swofford R, Pirun M, Zody MC, White S et al. 2012. The genomic basis of adaptive evolution in threespine sticklebacks. *Nature* **484**(7392): 55-61.
283. Jung S, Swart EC, Minx PJ, Magrini V, Mardis ER, Landweber LF, Eddy SR. 2011. Exploiting Oxytricha trifallax nanochromosomes to screen for non-coding RNA genes. *Nucleic Acids Research* **39**(17): 7529-7547.
284. Kaelin CB, Xu X, Hong LZ, David VA, McGowan KA, Schmidt-Kuentzel A, Roelke ME, Pino J, Pontius J, Cooper GM et al. 2012. Specifying and Sustaining Pigmentation Patterns in Domestic and Wild Cats. *Science* **337**(6101): 1536-1541.
285. Kanchi KL, Johnson KJ, Lu C, McLellan MD, Leiserson MDM, Wendl MC, Zhang Q, Koboldt DC, Xie M, Kandoth C et al. 2014. Integrated analysis of germline and somatic variants in ovarian cancer. *Nature Communications* **5**.
286. Kandoth C, McLellan MD, Vandin F, Ye K, Niu B, Lu C, Xie M, Zhang Q, McMichael JF, Wyczalkowski MA et al. 2013. Mutational landscape and significance across 12 major cancer types. *Nature* **502**(7471): 333-+.
287. Kantarci S, Ackerman KG, Russell MK, Longoni M, Sougnez C, Noonan KM, Hatchwell E, Zhang X, Pieretti Vanmarcke R, Anyane-Yeboa K et al. 2010. Characterization of the chromosome 1q41q42.12 region, and the candidate gene DISP1, in patients with CDH. *American journal of medical genetics Part A* **152A**(10): 2493-2504.
288. Karaca E, Weitzer S, Pehlivan D, Shiraishi H, Gogakos T, Hanada T, Jhangiani SN, Wiszniewski W, Withers M, Campbell IM et al. 2014. Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. *Cell* **157**(3): 636-650.
289. Karlsson EK, Sigurdsson S, Ivansson E, Thomas R, Elvers I, Wright J, Howald C, Tonomura N, Perloski M, Swofford R et al. 2013. Genome-wide analyses implicate 33 loci in heritable dog osteosarcoma, including regulatory variants near CDKN2A/B. *Genome Biol* **14**(12): R132.
290. Khajuria C, Williams CE, El BM, Whitworth RJ, Richards S, Stuart JJ, Chen MS. 2013. Deep sequencing and genome-wide analysis reveals the expansion of MicroRNA genes in the gall midge Mayetiola destructor. *BMCGenomics* **14**: 187.
291. Khurana E, Fu Y, Colonna V, Mu XJ, Kang HM, Lappalainen T, Sboner A, Lochovsky L, Chen J, Harmanci A et al. 2013. Integrative annotation of variants from 1092 humans: application to cancer genomics. *Science* **342**(6154): 1235587.
292. Kidd JM, Graves T, Newman TL, Fulton R, Hayden HS, Malig M, Kallicki J, Kaul R, Wilson RK, Eichler EE. 2010. A Human Genome Structural Variation Sequencing Resource Reveals Insights into Mutational Mechanisms. *Cell* **143**(5): 837-847.
293. Kidd JM, Sampas N, Antonacci F, Graves T, Fulton R, Hayden HS, Alkan C, Malig M, Ventura M, Giannuzzi G et al. 2010. Characterization of missing human genome sequences and copy-number polymorphic insertions. *Nature Methods* **7**(5): 365-U347.
294. Kirby A, Gnirke A, Jaffe DB, Baresova V, Pochet N, Blumenstiel B, Ye C, Aird D, Stevens C, Robinson JT et al. 2013. Mutations causing medullary cystic kidney disease type 1 lie in a large VNTR in MUC1 missed by massively parallel sequencing. *Nat Genet* **45**(3): 299-303.
295. Klco JM, Spencer DH, Lamprecht TL, Sarkaria SM, Wylie T, Magrini V, Hundal J, Walker J, Varghese N, Erdmann-Gilmore P et al. 2013. Genomic impact of transient low-dose decitabine treatment on primary AML cells. *Blood* **121**(9): 1633-1643.
296. Klco JM, Spencer DH, Miller C, Lamprecht T, Fulton RS, Ding L, Wilson RK, Ley TJ. 2013. Use of Targeted Deep Sequencing To Analyze the Genomic Similarities of Concurrent Acute Myeloid Leukemia in the Peripheral Blood and Bone Marrow. *Modern Pathology* **26**: 435A-435A.
297. Klco JM, Spencer DH, Miller C, Lamprecht T, Fulton RS, Welch JS, Ding L, Wilson RK, Ley TJ. 2012. Deep Digital Sequencing Identifies an AML Subclone with Enhanced in Vitro and in Vivo Growth Properties Associated with Disease Relapse. *Blood* **120**(21).
298. Klco JM, Spencer DH, Miller CA, Griffith M, Lamprecht TL, O'Laughlin M, Fronick C, Magrini V, Demeter RT, Fulton RS et al. 2014. Functional Heterogeneity of Genetically Defined Subclones in Acute Myeloid Leukemia. *Cancer Cell* **25**(3): 379-392.
299. Koboldt DC, Ding L, Mardis ER, Wilson RK. 2010. Challenges of sequencing human genomes. *Briefings in Bioinformatics* **11**(5): 484-498.
300. Koboldt DC Fulton RS McLellan MD Schmidt H Kalicki-Veizer J McMichael JF Fulton LL Dooling DJ Ding L Mardis ER et al. 2012. Comprehensive molecular portraits of human breast tumours. *Nature* **490**(7418): 61-70.
301. Koboldt DC, Larson DE, Chen K, Ding L, Wilson RK. 2012. Massively parallel sequencing approaches for characterization of structural variation. *Methods in molecular biology (Clifton, NJ)* **838**: 369-384.
302. Koboldt DC, Larson DE, Sullivan LS, Bowne SJ, Steinberg KM, Churchill JD, Buhr AC, Nutter N, Pierce EA, Blanton SH et al. 2014. Exome-Based Mapping and Variant Prioritization for Inherited Mendelian Disorders. *American Journal of Human Genetics* **94**(3): 373-384.
303. Koboldt DC, Staisch J, Thillainathan B, Haines K, Baird SE, Chamberlin HM, Haag ES, Miller RD, Gupta BP. 2010. A toolkit for rapid gene mapping in the nematode Caenorhabditis briggsae. *Bmc Genomics* **11**.
304. Koboldt DC, Steinberg KM, Larson DE, Wilson RK, Mardis ER. 2013. The Next-Generation Sequencing Revolution and Its Impact on Genomics. *Cell* **155**(1): 27-38.
305. Koboldt DC, Zhang Q, Larson DE, Shen D, McLellan MD, Lin L, Miller CA, Mardis ER, Ding L, Wilson RK. 2012. VarScan 2: Somatic mutation and copy number alteration discovery in cancer by exome sequencing. *Genome Research* **22**(3): 568-576.
306. Kostic AD, Gevers D, Pedamallu CS, Michaud M, Duke F, Earl AM, Ojesina AI, Jung J, Bass AJ, Tabernero J et al. 2012. Genomic analysis identifies association of Fusobacterium with colorectal carcinoma. *Genome Res* **22**(2): 292-298.
307. Kowalko JE, Rohner N, Linden TA, Rompani SB, Warren WC, Borowsky R, Tabin CJ, Jeffery WR, Yoshizawa M. 2013. Convergence in feeding posture occurs through different genetic loci in independently evolved cave populations of Astyanax mexicanus. *Proceedings of the National Academy of Sciences of the United States of America* **110**(42): 16933-16938.
308. Kucerova E, Clifton SW, Xia X-Q, Long F, Porwollik S, Fulton L, Fronick C, Minx P, Kyung K, Warren W et al. 2010. Genome Sequence of Cronobacter sakazakii BAA-894 and Comparative Genomic Hybridization Analysis with Other Cronobacter Species. *Plos One* **5**(3): A51-A60.
309. Kuczynski J, Lauber CL, Walters WA, Parfrey LW, Clemente JC, Gevers D, Knight R. 2012. Experimental and analytical tools for studying the human microbiome. *Nature reviews Genetics* **13**(1): 47-58.
310. Kunstner A, Wolf JBW, Backstrom N, Whitney O, Balakrishnan CN, Day L, Edwards SV, Janes DE, Schlinger BA, Wilson RK et al. 2010. Comparative genomics based on massive parallel transcriptome sequencing reveals patterns of substitution and selection across 10 bird species. *Molecular Ecology* **19**: 266-276.
311. Laine CM, Joeng KS, Campeau PM, Kiviranta R, Tarkkonen K, Grover M, Lu JT, Pekkinen M, Wessman M, Heino TJ et al. 2013. WNT1 mutations in early-onset osteoporosis and osteogenesis imperfecta *NEnglJMed* **368**(19): 1809-1816.
312. Landau DA, Carter SL, Stojanov P, McKenna A, Stevenson K, Lawrence MS, Sougnez C, Stewart C, Sivachenko A, Wang L et al. 2013. Evolution and impact of subclonal mutations in chronic lymphocytic leukemia. *Cell* **152**(4): 714-726.
313. Larman TC, DePalma SR, Hadjipanayis AG, Cancer Genome Atlas Research N, Protopopov A, Zhang J, Gabriel SB, Chin L, Seidman CE, Kucherlapati R et al. 2012. Spectrum of somatic mitochondrial mutations in five cancers. *Proc Natl Acad Sci U S A* **109**(35): 14087-14091.
314. Larson DE, Harris CC, Chen K, Koboldt DC, Abbott TE, Dooling DJ, Ley TJ, Mardis ER, Wilson RK, Ding L. 2012. SomaticSniper: identification of somatic point mutations in whole genome sequencing data. *Bioinformatics* **28**(3): 311-317.
315. Larson DE, Johnson RI, Swat M, Cordero JB, Glazier JA, Cagan RL. 2010. Computer Simulation of Cellular Patterning Within the Drosophila Pupal Eye. *Plos Computational Biology* **6**(7).
316. Larson G, Karlsson EK, Perri A, Webster MT, Ho SY, Peters J, Stahl PW, Piper PJ, Lingaas F, Fredholm M et al. 2012. Rethinking dog domestication by integrating genetics, archeology, and biogeography. *Proc Natl Acad Sci U S A* **109**(23): 8878-8883.
317. Lawrence MS, Stojanov P, Mermel CH, Robinson JT, Garraway LA, Golub TR, Meyerson M, Gabriel SB, Lander ES, Getz G. 2014. Discovery and saturation analysis of cancer genes across 21 tumour types. *Nature* **505**(7484): 495-501.
318. Lawrence MS, Stojanov P, Polak P, Kryukov GV, Cibulskis K, Sivachenko A, Carter SL, Stewart C, Mermel CH, Roberts SA et al. 2013. Mutational heterogeneity in cancer and the search for new cancer-associated genes. *Nature* **499**(7457): 214-218.
319. Lee E, Iskow R, Yang L, Gokcumen O, Haseley P, Luquette LJ, III, Lohr JG, Harris CC, Ding L, Wilson RK et al. 2012. Landscape of Somatic Retrotransposition in Human Cancers *Science*.
320. Lee E, Iskow R, Yang L, Gokcumen O, Haseley P, Luquette LJ, III, Lohr JG, Harris CC, Ding L, Wilson RK et al. 2012. Landscape of Somatic Retrotransposition in Human Cancers. *Science* **337**(6097): 967-971.
321. Lee JH, Huynh M, Silhavy JL, Kim S, Dixon-Salazar T, Heiberg A, Scott E, Bafna V, Hill KJ, Collazo A et al. 2012. De novo somatic mutations in components of the PI3K-AKT3-mTOR pathway cause hemimegalencephaly. *Nat Genet* **44**(8): 941-945.
322. Lee JH, Silhavy JL, Lee JE, Al-Gazali L, Thomas S, Davis EE, Bielas SL, Hill KJ, Iannicelli M, Brancati F et al. 2012. Evolutionarily assembled cis-regulatory module at a human ciliopathy locus. *Science* **335**(6071): 966-969.
323. Lee RS, Stewart C, Carter SL, Ambrogio L, Cibulskis K, Sougnez C, Lawrence MS, Auclair D, Mora J, Golub TR et al. 2012. A remarkably simple genome underlies highly malignant pediatric rhabdoid cancers. *The Journal of clinical investigation* **122**(8): 2983-2988.
324. Lennon NJ, Lintner RE, Anderson S, Alvarez P, Barry A, Brockman W, Daza R, Erlich RL, Giannoukos G, Green L et al. 2010. A scalable, fully automated process for construction of sequence-ready barcoded libraries for 454. *Genome Biol* **11**(2): R15.
325. Leung W, Shaffer CD, Cordonnier T, Wong J, Itano MS, Tempel EES, Kellmann E, Desruisseau DM, Cain C, Carrasquillo R et al. 2010. Evolution of a Distinct Genomic Domain in Drosophila: Comparative Analysis of the Dot Chromosome in Drosophila melanogaster and Drosophila virilis. *Genetics* **185**(4): 1519-U1629.
326. Levin JZ, Yassour M, Adiconis X, Nusbaum C, Thompson DA, Friedman N, Gnirke A, Regev A. 2010. Comprehensive comparative analysis of strand-specific RNA sequencing methods. *Nat Methods* **7**(9): 709-715.
327. Ley TJ, Ding L, Walter MJ, McLellan MD, Lamprecht T, Larson DE, Kandoth C, Payton JE, Baty J, Welch J et al. 2010. DNMT3A Mutations in Acute Myeloid Leukemia. *New England Journal of Medicine* **363**(25): 2424-2433.
328. Ley TJ, Ding L, Walter MJ, McLellan MD, Lamprecht T, Larson DE, Kandoth C, Payton JE, Baty J, Welch JS et al. 2010. Mutations In the DNA Methyltransferase Gene DNMT3A Are Highly Recurrent In Patients with Intermediate Risk Acute Myeloid Leukemia, and Predict Poor Outcomes. *Blood* **116**(21): 49-50.
329. Ley TJ Miller C Ding L Raphael BJ Mungall AJ Robertson AG Hoadley K Triche TJ, Jr. Laird PW Baty JD et al. 2013. Genomic and Epigenomic Landscapes of Adult De Novo Acute Myeloid Leukemia. *New England Journal of Medicine* **368**(22): 2059-2074.
330. Li B-W, Rush AC, Jiang D-J, Mitreva M, Abubucker S, Weil GJ. 2011. Gender-Associated Genes in Filarial Nematodes Are Important for Reproduction and Potential Intervention Targets. *Plos Neglected Tropical Diseases* **5**(1).
331. Li B-W, Rush AC, Jiang D-J, Mitreva M, Weil GJ. 2010. GENDER-REGULATED BRUGIA MALAYI GENES HAVE CAENORHABDITIS ELEGANS HOMOLOGUES WITH GERMLINE (SPERMATOGENESIS AND OOGENENSIS) OR EMBRYOGENESIS-ENRICHED EXPRESSION. *American Journal of Tropical Medicine and Hygiene* **83**(5): 66-67.
332. Li B-W, Wang Z, Rush AC, Mitreva M, Weil GJ. 2012. Transcription profiling reveals stage- and function-dependent expression patterns in the filarial nematode Brugia malayi. *Bmc Genomics* **13**.
333. Li D, Tanaka M, Brunicardi FC, Fisher WE, Gibbs RA, Gingras MC. 2011. Association between somatostatin receptor 5 gene polymorphisms and pancreatic cancer risk and survival *Cancer* **117**(13): 2863-2872.
334. Li S, Shen D, Shao J, Crowder R, Liu W, Prat A, He X, Liu S, Hoog J, Lu C et al. 2013. Endocrine-Therapy-Resistant ESR1 Variants Revealed by Genomic Characterization of Breast-Cancer-Derived Xenografts. *Cell Reports* **4**(6): 1116-1130.
335. Liao RG, Jung J, Tchaicha J, Wilkerson MD, Sivachenko A, Beauchamp EM, Liu Q, Pugh TJ, Pedamallu CS, Hayes DN et al. 2013. Inhibitor-sensitive FGFR2 and FGFR3 mutations in lung squamous cell carcinoma. *Cancer Res* **73**(16): 5195-5205.
336. Li-Byarlay H, Li Y, Stroud H, Feng S, Newman TC, Kaneda M, Hou KK, Worley KC, Elsik CG, Wickline SA et al. 2013. RNA interference knockdown of DNA methyl-transferase 3 affects gene alternative splicing in the honey bee. *ProcNatlAcadSciUSA* **110**(31): 12750-12755.
337. Lim ET, Raychaudhuri S, Sanders SJ, Stevens C, Sabo A, MacArthur DG, Neale BM, Kirby A, Ruderfer DM, Fromer M et al. 2013. Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders *Neuron* **77**(2): 235-242.
338. Lin H, Sinner MF, Brody JA, Arking DE, Lunetta KL, Rienstra M, Lubitz SA, Magnani JW, Sotoodehnia N, McKnight B et al. 2013. Targeted sequencing in candidate genes for atrial fibrillation: The Cohorts for Heart and Aging Research in Genomic Epidemiology Targeted Sequencing Study *Heart Rhythm*.
339. Lindblad-Toh K, Garber M, Zuk O, Lin MF, Parker BJ, Washietl S, Kheradpour P, Ernst J, Jordan G, Mauceli E et al. 2011. A high-resolution map of human evolutionary constraint using 29 mammals. *Nature* **478**(7370): 476-482.
340. Lindblad-Toh K, Garber M, Zuk O, Lin MF, Parker BJ, Washietl S, Kheradpour P, Ernst J, Jordan G, Mauceli E et al. 2011. A high-resolution map of human evolutionary constraint using 29 mammals. *Nature* **478**(7370): 476-482.
341. Lindsay ME, Schepers D, Bolar NA, Doyle JJ, Gallo E, Fert-Bober J, Kempers MJ, Fishman EK, Chen Y, Myers L et al. 2012. Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. *NatGenet* **44**(8): 922-927.
342. Lindstrand A, Davis EE, Carvalho CM, Pehlivan D, Willer JR, Tsai IC, Ramanathan S, Zuppan C, Sabo A, Muzny D et al. 2014. Recurrent CNVs and SNVs at the NPHP1 locus contribute pathogenic alleles to Bardet-Biedl syndrome. *Am J Hum Genet* **94**(5): 745-754.
343. Link DC, Schuettpelz LG, Shen D, Wang J, Walter MJ, Kulkarni S, Payton JE, Ivanovich J, Goodfellow PJ, Le Beau M et al. 2011. Identification of a Novel TP53 Cancer Susceptibility Mutation Through Whole-Genome Sequencing of a Patient With Therapy-Related AML. *Jama-Journal of the American Medical Association* **305**(15): 1568-1576.
344. Liu CT, Young KL, Brody JA, Olden M, Wojczynski MK, Heard-Costa N, Li G, Morrison AC, Muzny D, Gibbs RA et al. 2014. Sequence Variation in TMEM18 in Association With Body Mass Index: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Targeted Sequencing Study. *Circulation Cardiovascular genetics* **7**(3): 344-349.
345. Liu L, Sabo A, Neale BM, Nagaswamy U, Stevens C, Lim E, Bodea CA, Muzny D, Reid JG, Banks E et al. 2013. Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls *PLoSGenet* **9**(4): e1003443.
346. Liu SH, Patel S, Gingras MC, Nemunaitis J, Zhou G, Chen C, Li M, Fisher W, Gibbs RA, Brunicardi FC. 2010. PDX-1: demonstration of oncogenic properties in pancreatic cancer *Cancer*.
347. Liu SH, Rao DD, Nemunaitis J, Senzer N, Zhou G, Dawson D, Gingras MC, Wang Z, Gibbs R, Norman M et al. 2012. PDX-1 is a therapeutic target for pancreatic cancer, insulinoma and islet neoplasia using a novel RNA interference platform. *PLoSOne* **7**(8): e40452.
348. Locke DP Hillier LW Warren WC Worley KC Nazareth LV Muzny DM Yang SP Wang Z Chinwalla AT Minx P et al. 2011. Comparative and demographic analysis of orang-utan genomes *Nature* **469**(7331): 529-533.
349. Loh YH, Bezault E, Muenzel FM, Roberts RB, Swofford R, Barluenga M, Kidd CE, Howe AE, Di Palma F, Lindblad-Toh K et al. 2013. Origins of shared genetic variation in African cichlids. *Mol Biol Evol* **30**(4): 906-917.
350. Lohr JG, Stojanov P, Carter SL, Cruz-Gordillo P, Lawrence MS, Auclair D, Sougnez C, Knoechel B, Gould J, Saksena G et al. 2014. Widespread genetic heterogeneity in multiple myeloma: implications for targeted therapy. *Cancer Cell* **25**(1): 91-101.
351. Lohr JG, Stojanov P, Lawrence MS, Auclair D, Chapuy B, Sougnez C, Cruz-Gordillo P, Knoechel B, Asmann YW, Slager SL et al. 2012. Discovery and prioritization of somatic mutations in diffuse large B-cell lymphoma (DLBCL) by whole-exome sequencing. *Proc Natl Acad Sci U S A* **109**(10): 3879-3884.
352. London SJ, Gao W, Gharib SA, Hancock DB, Wilk JB, House JS, Gibbs RA, Muzny DM, Lumley T, Franceschini N et al. 2014. ADAM19 and HTR4 Variants and Pulmonary Function: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium Targeted Sequencing Study. *Circulation Cardiovascular genetics* **7**(3): 350-358.
353. Lotta LA, Tuana G, Yu J, Martinelli I, Wang M, Yu F, Passamonti SM, Pappalardo E, Valsecchi C, Scherer SE et al. 2013. Next-generation sequencing study finds an excess of rare, coding single-nucleotide variants of ADAMTS13 in patients with deep vein thrombosis *JThrombHaemost* **11**(7): 1228-1239.
354. Lotta LA, Wang M, Yu J, Martinelli I, Yu F, Passamonti SM, Consonni D, Pappalardo E, Menegatti M, Scherer SE et al. 2012. Identification of genetic risk variants for deep vein thrombosis by multiplexed next-generation sequencing of 186 hemostatic/pro-inflammatory genes. *BMCMedGenomics* **5**(1): 7.
355. Lotta LA, Wu HM, Mackie IJ, Noris M, Veyradier A, Scully MA, Remuzzi G, Coppo P, Liesner R, Donadelli R et al. 2012. Residual plasmatic activity of ADAMTS13 is correlated with phenotype severity in congenital thrombotic thrombocytopenic purpura. *Blood* **120**(2): 440-448.
356. Lowe CB, Kellis M, Siepel A, Raney BJ, Clamp M, Salama SR, Kingsley DM, Lindblad-Toh K, Haussler D. 2011. Three periods of regulatory innovation during vertebrate evolution. *Science* **333**(6045): 1019-1024.
357. Lozupone C, Cota-Gomez A, Palmer BE, Linderman DJ, Charlson ES, Sodergren E, Mitreva M, Abubucker S, Martin J, Yao G et al. 2013. Widespread Colonization of the Lung by Tropheryma whipplei in HIV Infection. *American Journal of Respiratory and Critical Care Medicine* **187**(10): 1110-1117.
358. Lu JT, Wang Y, Gibbs RA, Yu F. 2012. Characterizing linkage disequilibrium and evaluating imputation power of human genomic insertion-deletion polymorphisms *Genome Biol* **13**(2): R15.
359. Lucia Carbone RAH, Sante Gnerre, Krishna R. Veeramah, Belen Lorente-Galdos, John Huddleston, Thomas J. Meyer, Javier Herrero, Christian Roos, Bronwen Aken, Fabio Anaclerio, Nicoletta Archidiacono, Carl Baker, Daniel Barrell, Mark A. Batzer, Kathryn Beal, Antoine Blancher, Craig L. Bohrson, Markus Brameier, Michael S. Campbell, Oronzo Capozzi, Claudio Casola, Giorgia Chiatante, Andrew Cree, Annette Damert, Pieter J. de Jong, Laura Dumas, Marcos Fernandez-Callejo, Paul Flicek, Nina V. Fuchs, Marta Gut, Ivo Gut, Matthew W. Hahn, Jéssica Hernández-Rodríguez, LaDeana Hillier, Robert Hubley, Bianca Ianc, Zsuzsanna Izsvák, Nina G. Jablonski, Laurel Johnstone, Anis Karimpour-Fard, Miriam K. Konkel, Dennis Kostka, Nathan H. Lazar,, Sandra L. Lee, Lora R. Lewis, Yue Liu, Devin P. Locke,b, Swapan Mallick, Fernando L. Mendez, Matthieu Muffato, Lynne Nazareth, Kimberly A. Nevonen, Majesta O’Bleness, Cornelia Ochis, Duncan T. Odom, Katherine S. Pollard, Javier Quilez, David Reich0, Mariano Rocchi, Gerald G. Schumann, Stephen Searle, James M. Sikela, Gabriella Skollar, Arian Smit, Kemal Sonmez, Boudewijn ten Hallers0,c, Elizabeth Terhune, Gregg W.C. Thomas, Brygg Ullmer, Mario Ventura, Jerilyn A. Walker, Jeff D. Wall, Lutz Walter0, Michelle C. Ward,d, Sarah Wheelan, Christopher Whelan, Simon White, Larry J. Wilhelm, August E. Woerner, Mark Yandell, Baoli Zhu0,e, Michael Hammer, Tomas Marques-Bonet, Evan E. Eichler,, Lucinda Fulton, Catrina Fronick, Donna M. Muzny, Wesley C. Warren, Kim C. Worley, Jeffrey Rogers, Richard K. Wilson & Richard A. Gibbs. 2014. The gibbon genome gives insights on the fast karyotype evolution of small apes *Nature* **In Press**.
360. Lupski JR, Belmont JW, Boerwinkle E, Gibbs RA. 2011. Clan genomics and the complex architecture of human disease *Cell* **147**(1): 32-43.
361. Lupski JR, Gonzaga-Jauregui C, Yang Y, Bainbridge MN, Jhangiani S, Buhay CJ, Kovar CL, Wang M, Hawes AC, Reid JG et al. 2013. Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy *Genome Med* **5**(6): 57.
362. Lupski JR, Reid JG, Gonzaga-Jauregui C, Rio DD, Chen DC, Nazareth L, Bainbridge M, Dinh H, Jing C, Wheeler DA et al. 2010. Whole-genome sequencing in a patient with Charcot-Marie-Tooth neuropathy *NEnglJMed* **362**(13): 1181-1191.
363. Ma L, Huang DW, Cuomo CA, Sykes S, Fantoni G, Das B, Sherman BT, Yang J, Huber C, Xia Y et al. 2013. Sequencing and characterization of the complete mitochondrial genomes of three Pneumocystis species provide new insights into divergence between human and rodent Pneumocystis. *FASEB journal : official publication of the Federation of American Societies for Experimental Biology* **27**(5): 1962-1972.
364. MacArthur DG, Balasubramanian S, Frankish A, Huang N, Morris J, Walter K, Jostins L, Habegger L, Pickrell JK, Montgomery SB et al. 2012. A systematic survey of loss-of-function variants in human protein-coding genes *Science* **335**(6070): 823-828.
365. Mackay TF, Richards S, Stone EA, Barbadilla A, Ayroles JF, Zhu D, Casillas S, Han Y, Magwire MM, Cridland JM et al. 2012. The Drosophila melanogaster Genetic Reference Panel. *Nature* **482**(7384): 173-178.
366. Magnani JW, Brody JA, Prins BP, Arking DE, Lin H, Yin X, Liu CT, Morrison AC, Zhang F, Spector TD et al. 2014. Sequencing of SCN5A Identifies Rare and Common Variants Associated With Cardiac Conduction: Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium. *Circulation Cardiovascular genetics* **7**(3): 365-373.
367. Maher CA, Wilson RK. 2012. Chromothripsis and Human Disease: Piecing Together the Shattering Process. *Cell* **148**(1-2): 29-32.
368. Mangiola S, Young ND, Sternberg PW, Strube C, Korhonen PK, Mitreva M, Scheerlinck J-P, Hofmann A, Jex AR, Gasser RB. 2014. Analysis of the transcriptome of adult Dictyocaulus filaria and comparison with Dictyocaulus viviparus, with a focus on molecules involved in host-parasite interactions. *International Journal for Parasitology* **44**(3-4): 251-261.
369. Mannstadt M, Harris M, Bravenboer B, Chitturi S, Dreijerink KM, Lambright DG, Lim ET, Daly MJ, Gabriel S, Juppner H. 2013. Germline mutations affecting Galpha11 in hypoparathyroidism. *The New England journal of medicine* **368**(26): 2532-2534.
370. Manzini MC, Tambunan DE, Hill RS, Yu TW, Maynard TM, Heinzen EL, Shianna KV, Stevens CR, Partlow JN, Barry BJ et al. 2012. Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg syndrome. *Am J Hum Genet* **91**(3): 541-547.
371. Marchio C, Rodrigues DN, Wilkerson P, Lambros MB, Weigelt B, Sapino A, Mackay A, Maher C, Natrajan R, Reis-Filho JS. 2012. Breast Micropapillary Carcinomas: RNA-Seq and Mutation Profiling. *Laboratory Investigation* **92**: 53A-53A.
372. Mardis E, Ding L, Li S, Larson DE, Chen K, Wallis J, Hoog J, Deshryver K, Wilson R, Ellis MJ. 2010. Massive parallel sequencing of an African-American basal-like breast cancer: Comparison of primary tumor, metastasis, and xenograft. *Journal of Clinical Oncology* **28**(15).
373. Mardis ER. 2010. The $1,000 genome, the $100,000 analysis? *Genome Medicine* **2**.
374. -. 2010. Cancer genomics identifies determinants of tumor biology. *Genome Biology* **11**(5).
375. -. 2010. Next-generation cancer genomics. *Genome Biology* **11**.
376. -. 2011. A decade's perspective on DNA sequencing technology. *Nature* **470**(7333): 198-203.
377. -. 2011. Future of DNA Sequencing Technology. *Genetic Engineering & Biotechnology News* **31**(17): 40-+.
378. -. 2011. A glimpse at tumor genome evolution. *Genome Biology* **12**: 2-2.
379. -. 2012. Applying next-generation sequencing to pancreatic cancer treatment. *Nature Reviews Gastroenterology & Hepatology* **9**(8): 477-486.
380. -. 2012. Genome sequencing and cancer. *Current Opinion in Genetics & Development* **22**(3): 245-250.
381. -. 2013. Next-Generation Sequencing Platforms. *Annual Review of Analytical Chemistry, Vol 6* **6**: 287-303.
382. -. 2014. The translation of cancer genomics: time for a revolution in clinical cancer care. *Genome Medicine* **6**.
383. Mardis ER, Ding L, Westervelt P, Welch JS, Klco JM, DiPersio JF, Wilson RK, Ley TJ. 2012. Next-Generation Sequencing: A Discovery Tool for Blood Disorders. *Blood* **120**(21).
384. Margolin DH, Kousi M, Chan YM, Lim ET, Schmahmann JD, Hadjivassiliou M, Hall JE, Adam I, Dwyer A, Plummer L et al. 2013. Ataxia, dementia, and hypogonadotropism caused by disordered ubiquitination. *The New England journal of medicine* **368**(21): 1992-2003.
385. Marneros AG, Beck AE, Turner EH, McMillin MJ, Edwards MJ, Field M, de Macena Sobreira NL, Perez AB, Fortes JA, Lampe AK et al. 2013. Mutations in KCTD1 cause scalp-ear-nipple syndrome. *AmJHumGenet* **92**(4): 621-626.
386. Marth GT, Yu F, Indap AR, Garimella K, Gravel S, Leong WF, Tyler-Smith C, Bainbridge M, Blackwell T, Zheng-Bradley X et al. 2011. The functional spectrum of low-frequency coding variation *Genome Biol* **12**(9): R84.
387. Martin J, Abubucker S, Heizer E, Taylor CM, Mitreva M. 2012. Nematode.net update 2011: addition of data sets and tools featuring next-generation sequencing data. *Nucleic Acids Research* **40**(D1): D720-D728.
388. Martin J, Sykes S, Young S, Kota K, Sanka R, Sheth N, Orvis J, Sodergren E, Wang Z, Weinstock GM et al. 2012. Optimizing Read Mapping to Reference Genomes to Determine Composition and Species Prevalence in Microbial Communities. *Plos One* **7**(6).
389. Martinez DA, Oliver BG, Graser Y, Goldberg JM, Li W, Martinez-Rossi NM, Monod M, Shelest E, Barton RC, Birch E et al. 2012. Comparative genome analysis of Trichophyton rubrum and related dermatophytes reveals candidate genes involved in infection. *mBio* **3**(5): e00259-00212.
390. Martinez FJ, Lee JH, Lee JE, Blanco S, Nickerson E, Gabriel S, Frye M, Al-Gazali L, Gleeson JG. 2012. Whole exome sequencing identifies a splicing mutation in NSUN2 as a cause of a Dubowitz-like syndrome. *Journal of medical genetics* **49**(6): 380-385.
391. McCue ME, Bannasch DL, Petersen JL, Gurr J, Bailey E, Binns MM, Distl O, Guerin G, Hasegawa T, Hill EW et al. 2012. A high density SNP array for the domestic horse and extant Perissodactyla: utility for association mapping, genetic diversity, and phylogeny studies. *PLoS Genet* **8**(1): e1002451.
392. McDonald SA, Mardis ER, Ota D, Watson MA, Pfeifer JD, Green JM. 2012. Comprehensive Genomic Studies Emerging Regulatory, Strategic, and Quality Assurance Challenges for Biorepositories. *American Journal of Clinical Pathology* **138**(1): 31-41.
393. McEvoy J, Nagahawatte P, Finkelstein D, Richards-Yutz J, Valentine M, Ma J, Mullighan C, Song G, Chen X, Wilson M et al. 2014. RB1 gene inactivation by chromothripsis in human retinoblastoma. *Oncotarget* **5**(2): 438-450.
394. McGuire AL, Majumder MA, Halpern SD, Swindell JS, Yaeger LV, Gibbs RA, Wheeler TM. 2010. Taking DNA from the dead *NatRevGenet* **11**(5): 318.
395. McIntyre JC, Davis EE, Joiner A, Williams CL, Tsai IC, Jenkins PM, McEwen DP, Zhang L, Escobado J, Thomas S et al. 2012. Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model *NatMed*.
396. McKenna A, Hanna M, Banks E, Sivachenko A, Cibulskis K, Kernytsky A, Garimella K, Altshuler D, Gabriel S, Daly M et al. 2010. The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. *Genome Res* **20**(9): 1297-1303.
397. McNulty SN, Abubuckar S, Fischer K, Mitreva M, Weil GJ, Fischer PU. 2010. THE TRANSCRIPTOME OF THE WOLBACHIA-FREE FILARIAL NEMATODE ONCHOCERCA FLEXUOSA. *American Journal of Tropical Medicine and Hygiene* **83**(5): 246-246.
398. McNulty SN, Abubucker S, Simon GM, Mitreva M, McNulty NP, Fischer K, Curtis KC, Brattig NW, Weil GJ, Fischer PU. 2012. Transcriptomic and Proteomic Analyses of a Wolbachia-Free Filarial Parasite Provide Evidence of Trans-Kingdom Horizontal Gene Transfer. *Plos One* **7**(9).
399. McNulty SN, Foster JM, Mitreva M, Hotopp JCD, Martin J, Fischer K, Wu B, Davis PJ, Kumar S, Brattig NW et al. 2010. Endosymbiont DNA in Endobacteria-Free Filarial Nematodes Indicates Ancient Horizontal Genetic Transfer. *Plos One* **5**(6).
400. McNulty SN, Mitreva M, Weil GJ, Fischer PU. 2013. Inter and intra-specific diversity of parasites that cause lymphatic filariasis. *Infection Genetics and Evolution* **14**: 137-146.
401. Melnikov A, Galinsky K, Rogov P, Fennell T, Van Tyne D, Russ C, Daniels R, Barnes KG, Bochicchio J, Ndiaye D et al. 2011. Hybrid selection for sequencing pathogen genomes from clinical samples. *Genome Biol* **12**(8): R73.
402. Menon R, Gasser RB, Mitreva M, Ranganathan S. 2012. An analysis of the transcriptome of Teladorsagia circumcincta: its biological and biotechnological implications. *Bmc Genomics* **13**.
403. Methe BA Nelson KE Pop M Creasy HH Giglio MG Huttenhower C Gevers D Petrosino JF Abubucker S Badger JH et al. 2012. A framework for human microbiome research. *Nature* **486**(7402): 215-221.
404. Metzker ML. 2010. Sequencing technologies - the next generation *NatRevGenet* **11**(1): 31-46.
405. Meurs KM, Mauceli E, Lahmers S, Acland GM, White SN, Lindblad-Toh K. 2010. Genome-wide association identifies a deletion in the 3' untranslated region of striatin in a canine model of arrhythmogenic right ventricular cardiomyopathy. *Human genetics* **128**(3): 315-324.
406. Meyerson M, Gabriel S, Getz G. 2010. Advances in understanding cancer genomes through second-generation sequencing. *Nature reviews Genetics* **11**(10): 685-696.
407. Miller CA, Wilson RK, Ley TJ. 2013. Genomic Landscapes and Clonality of De Novo AML REPLY. *New England Journal of Medicine* **369**(15): 1473-1473.
408. Mills RE, Walter K, Stewart C, Handsaker RE, Chen K, Alkan C, Abyzov A, Yoon SC, Ye K, Cheetham RK et al. 2011. Mapping copy number variation by population-scale genome sequencing. *Nature* **470**(7332): 59-65.
409. Mitreva M. 2012. The genome of a blood fluke associated with human cancer. *Nature Genetics* **44**(2): 116-118.
410. Mitreva M, Jasmer DP. 2010. Trichinella spiralis: genomic application to control a zoonotic nematode. *Infectious disorders drug targets* **10**(5): 376-384.
411. Mitreva M, Jasmer DP, Zarlenga DS, Wang Z, Abubucker S, Martin J, Taylor CM, Yin Y, Fulton L, Minx P et al. 2011. The draft genome of the parasitic nematode Trichinella spiralis. *Nature Genetics* **43**(3): 228-U274.
412. Montemayor C, Montemayor OA, Ridgeway A, Lin F, Wheeler DA, Pletcher SD, Pereira FA. 2010. Genome-wide analysis of binding sites and direct target genes of the orphan nuclear receptor NR2F1/COUP-TFI. *PLoSOne* **5**(1): e8910.
413. Morrison AC, Voorman A, Johnson AD, Liu X, Yu J, Li A, Muzny D, Yu F, Rice K, Zhu C et al. 2013. Whole-genome sequence-based analysis of high-density lipoprotein cholesterol. *NatGenet* **45**(8): 899-901.
414. Moura AE, Janse van RC, Pilot M, Tehrani A, Best PB, Thornton M, Plon S, de Bruyn PJ, Worley KC, Gibbs RA et al. 2014. Killer Whale Nuclear Genome and mtDNA Reveal Widespread Population Bottleneck during the Last Glacial Maximum *MolBiolEvol* **31**(5): 1121-1131.
415. Mueller JL, Skaletsky H, Brown LG, Zaghlul S, Rock S, Graves T, Auger K, Warren WC, Wilson RK, Page DC. 2013. Independent specialization of the human and mouse X chromosomes for the male germ line. *Nature Genetics* **45**(9): 1083-+.
416. Mueller M, Barros P, Witherden AS, Roberts AL, Zhang Z, Schaschl H, Yu C-Y, Hurles ME, Schaffner C, Floto RA et al. 2013. Genomic Pathology of SLE-Associated Copy-Number Variation at the FCGR2C/FCGR3B/FCGR2B Locus. *American Journal of Human Genetics* **92**(1): 28-40.
417. Murdock DR, Clark GD, Bainbridge MN, Newsham I, Wu YQ, Muzny DM, Cheung SW, Gibbs RA, Ramocki MB. 2011. Whole-exome sequencing identifies compound heterozygous mutations in WDR62 in siblings with recurrent polymicrogyria *AmJMedGenetA* **155A**(9): 2071-2077.
418. Murphy JT, Bruinsma JJ, Schneider DL, Collier S, Guthrie J, Chinwalla A, Robertson JD, Mardis ER, Kornfeld K. 2011. Histidine Protects Against Zinc and Nickel Toxicity in Caenorhabditis elegans. *Plos Genetics* **7**(3).
419. Murphy SJ, Hart SN, Lima JF, Kipp BR, Klebig M, Winters JL, Szabo C, Zhang L, Eckloff BW, Petersen GM et al. 2013. Genetic alterations associated with progression from pancreatic intraepithelial neoplasia to invasive pancreatic tumor *Gastroenterology* **145**(5): 1098-1109.
420. Musunuru K, Pirruccello JP, Do R, Peloso GM, Guiducci C, Sougnez C, Garimella KV, Fisher S, Abreu J, Barry AJ et al. 2010. Exome sequencing, ANGPTL3 mutations, and familial combined hypolipidemia. *The New England journal of medicine* **363**(23): 2220-2227.
421. Muthappan V, Lee AY, Lamprecht TL, Akileswaran L, Dintzis SM, Lee C, Magrini V, Mardis ER, Shendure J, Van Gelder RN. 2011. Biome representational in silico karyotyping. *Genome Research* **21**(4): 626-633.
422. Muzny DM Bainbridge MN Chang K Dinh HH Drummond JA Fowler G Kovar CL Lewis LR Morgan MB Newsham IF et al. 2012. Comprehensive molecular characterization of human colon and rectal cancer. *Nature* **487**(7407): 330-337.
423. Nagaraja AK, Creighton CJ, Yu Z, Zhu H, Gunaratne PH, Reid JG, Olokpa E, Itamochi H, Ueno NT, Hawkins SM et al. 2010. A link between mir-100 and FRAP1/mTOR in clear cell ovarian cancer. *MolEndocrinol* **24**(2): 447-463.
424. Natrajan R, Wilkerson PM, Marchio C, Lambros MB, Ng CKY, Topfer C, Kozarewa I, Hakas J, Mitsopoulos K, Hardisson D et al. 2013. Functional Characterisation of Fusion Genes in Micropapillary Carcinomas of the Breast. *Modern Pathology* **26**: 57A-58A.
425. Natrajan R, Wilkerson PM, Marchio C, Piscuoglio S, Ng CKY, Wai P, Lambros MB, Samartzis EP, Dedes KJ, Frankum J et al. 2014. Characterization of the genomic features and expressed fusion genes in micropapillary carcinomas of the breast. *Journal of Pathology* **232**(5): 553-565.
426. Neafsey DE, Christophides GK, Collins FH, Emrich SJ, Fontaine MC, Gelbart W, Hahn MW, Howell PI, Kafatos FC, Lawson D et al. 2013. The evolution of the Anopheles 16 genomes project. *G3* **3**(7): 1191-1194.
427. Neafsey DE, Lawniczak MK, Park DJ, Redmond SN, Coulibaly MB, Traore SF, Sagnon N, Costantini C, Johnson C, Wiegand RC et al. 2010. SNP genotyping defines complex gene-flow boundaries among African malaria vector mosquitoes. *Science* **330**(6003): 514-517.
428. Neale BM, Kou Y, Liu L, Ma'ayan A, Samocha KE, Sabo A, Lin CF, Stevens C, Wang LS, Makarov V et al. 2012. Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature* **485**(7397): 242-245.
429. Nguyen NT, Cotton RT, Harring TR, Guiteau JJ, Gingras MC, Wheeler DA, O'Mahony CA, Gibbs RA, Brunicardi FC, Goss JA. 2011. A primer on a hepatocellular carcinoma bioresource bank using the cancer genome atlas guidelines: practical issues and pitfalls *World JSurg* **35**(8): 1732-1737.
430. Nielsen TO, Parker JS, Leung S, Voduc D, Ebbert M, Vickery T, Davies SR, Snider J, Stijleman IJ, Reed J et al. 2010. A Comparison of PAM50 Intrinsic Subtyping with Immunohistochemistry and Clinical Prognostic Factors in Tamoxifen-Treated Estrogen Receptor-Positive Breast Cancer. *Clinical Cancer Research* **16**(21): 5222-5232.
431. Niu B, Ye K, Zhang Q, Lu C, Xie M, McLellan MD, Wendl MC, Ding L. 2014. MSIsensor: microsatellite instability detection using paired tumor-normal sequence data. *Bioinformatics* **30**(7): 1015-1016.
432. Niu T, Smith DL, Yang Z, Gao S, Yin T, Jiang ZH, You M, Gibbs RA, Petrosino JF, Hu M. 2012. Bioactivity and Bioavailability of Ginsenosides are Dependent on the Glycosidase Activities of the A/J Mouse Intestinal Microbiome Defined by Pyrosequencing *PharmRes*.
433. Noushmehr H, Weisenberger DJ, Diefes K, Phillips HS, Pujara K, Berman BP, Pan F, Pelloski CE, Sulman EP, Bhat KP et al. 2010. Identification of a CpG Island Methylator Phenotype that Defines a Distinct Subgroup of Glioma. *Cancer Cell* **17**(5): 510-522.
434. Novarino G, El-Fishawy P, Kayserili H, Meguid NA, Scott EM, Schroth J, Silhavy JL, Kara M, Khalil RO, Ben-Omran T et al. 2012. Mutations in BCKD-kinase lead to a potentially treatable form of autism with epilepsy. *Science* **338**(6105): 394-397.
435. Novarino G, Fenstermaker AG, Zaki MS, Hofree M, Silhavy JL, Heiberg AD, Abdellateef M, Rosti B, Scott E, Mansour L et al. 2014. Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. *Science* **343**(6170): 506-511.
436. Ober U, Ayroles JF, Stone EA, Richards S, Zhu D, Gibbs RA, Stricker C, Gianola D, Schlather M, Mackay TF et al. 2012. Using whole-genome sequence data to predict quantitative trait phenotypes in Drosophila melanogaster. *PLoSGenet* **8**(5): e1002685.
437. O'Bleness M, Searles VB, Dickens CM, Astling D, Albracht D, Mak ACY, Lai YYY, Lin C, Chu C, Graves T et al. 2014. Finished sequence and assembly of the DUF1220-rich 1q21 region using a haploid human genome. *Bmc Genomics* **15**.
438. Okada Y, Diogo D, Greenberg JD, Mouassess F, Achkar WAL, Fulton RS, Denny JC, Gupta N, Mirel D, Gabriel S et al. 2014. Integration of Sequence Data from a Consanguineous Family with Genetic Data from an Outbred Population Identifies PLB1 as a Candidate Rheumatoid Arthritis Risk Gene. *Plos One* **9**(2).
439. Okamoto Y, Goksungur MT, Pehlivan D, Beck CR, Gonzaga-Jauregui C, Muzny DM, Atik MM, Carvalho CM, Matur Z, Bayraktar S et al. 2013. Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D *GenetMed*.
440. Olsson M, Meadows JR, Truve K, Rosengren Pielberg G, Puppo F, Mauceli E, Quilez J, Tonomura N, Zanna G, Docampo MJ et al. 2011. A novel unstable duplication upstream of HAS2 predisposes to a breed-defining skin phenotype and a periodic fever syndrome in Chinese Shar-Pei dogs. *PLoS Genet* **7**(3): e1001332.
441. Ondrovics M, Silbermayr K, Mitreva M, Young ND, Razzazi-Fazeli E, Gasser RB, Joachim A. 2013. Proteomic Analysis of Oesophagostomum dentatum (Nematoda) during Larval Transition, and the Effects of Hydrolase Inhibitors on Development. *Plos One* **8**(5).
442. Pabuwal V, Boswell M, Pasquali A, Wise SS, Kumar S, Shen Y, Garcia T, LaCerte C, Wise JP, Jr., Wise JP, Sr. et al. 2013. Transcriptomic analysis of cultured whale skin cells exposed to hexavalent chromium Cr(VI). *Aquatic Toxicology* **134**: 74-81.
443. Paciorkowski AR, Keppler-Noreuil K, Robinson L, Sullivan C, Sajan S, Christian SL, Bukshpun P, Gabriel SB, Gleeson JG, Sherr EH et al. 2013. Deletion 16p13.11 uncovers NDE1 mutations on the non-deleted homolog and extends the spectrum of severe microcephaly to include fetal brain disruption. *American journal of medical genetics Part A* **161A**(7): 1523-1530.
444. Page DC, Hughes JF, Bellott DW, Mueller JL, Gill ME, Larracuente A, Graves T, Muzny D, Warren WC, Gibbs RA et al. 2010. Reconstructing sex chromosome evolution. *Genome Biology* **11**.
445. Parikh A, Miranda ER, Katoh-Kurasawa M, Fuller D, Rot G, Zagar L, Curk T, Sucgang R, Chen R, Zupan B et al. 2010. Conserved developmental transcriptomes in evolutionarily divergent species. *Genome Biol* **11**(3): R35.
446. Parikh N, Hilsenbeck S, Creighton CJ, Dayaram T, Shuck R, Shinbrot E, Xi L, Gibbs RA, Wheeler DA, Donehower LA. 2014. Effects of TP53 mutational status on gene expression patterns across 10 human cancer types. *The Journal of pathology* **232**(5): 522-533.
447. Parker M, Chen X, Bahrami A, Dalton J, Rusch M, Wu G, Easton J, Cheung N-K, Dyer M, Mardis ER et al. 2012. Assessing telomeric DNA content in pediatric cancers using whole-genome sequencing data. *Genome Biology* **13**(12).
448. Parker M, Mohankumar KM, Punchihewa C, Weinlich R, Dalton JD, Li Y, Lee R, Tatevossian RG, Phoenix TN, Thiruvenkatam R et al. 2014. C11orf95-RELA fusions drive oncogenic NF-kappa B signalling in ependymoma. *Nature* **506**(7489): 451-+.
449. Pavan MG, Mesquita RD, Lawrence GG, Lazoski C, Dotson EM, Abubucker S, Mitreva M, Randall-Maher J, Monteiro FA. 2013. A nuclear single-nucleotide polymorphism (SNP) potentially useful for the separation of Rhodnius prolixus from members of the Rhodnius robustus cryptic species complex (Hemiptera: Reduviidae). *Infection Genetics and Evolution* **14**: 426-433.
450. Peacock WF, Chandra A, Char D, Collins S, Sahakian GD, Ding L, Dunbar L, Fermann G, Fonarow GC, Garrison N et al. 2014. Clevidipine in acute heart failure: Results of the A Study of Blood Pressure Control in Acute Heart Failure-A Pilot Study (PRONTO). *American Heart Journal* **167**(4): 529-536.
451. Pehlivan D, Karaca E, Aydin H, Beck CR, Gambin T, Muzny DM, Bilge GB, Karaman A, Jhangiani SN, Gibbs RA et al. 2014. Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia *EurJHumGenet*.
452. Pickering CR, Zhang J, Neskey DM, Zhao M, Jasser SA, Wang J, Ward A, Tsai CJ, Ortega Alves MV, Zhou JH et al. 2014. Squamous cell carcinoma of the oral tongue in young non-smokers is genomically similar to tumors in older smokers. *Clinical cancer research : an official journal of the American Association for Cancer Research*.
453. Pickering CR, Zhang J, Yoo SY, Bengtsson L, Moorthy S, Neskey DM, Zhao M, Ortega Alves MV, Chang K, Drummond J et al. 2013. Integrative genomic characterization of oral squamous cell carcinoma identifies frequent somatic drivers. *Cancer Discov*.
454. Plon SE, Wheeler DA, Strong LC, Tomlinson GE, Pirics M, Meng Q, Cheung HC, Begin PR, Muzny DM, Lewis L et al. 2011. Identification of genetic susceptibility to childhood cancer through analysis of genes in parallel. *Cancer Genet* **204**(1): 19-25.
455. Pong W, Ding L, McLellan M, Hussain I, Emnett R, Gianino S, Higer S, Leonard J, Guha A, Mardis E et al. 2013. WHOLE GENOME SEQUENCING REVEALSMICROGLIA AS KEY MICROENVIRONMENTAL DRIVERS OF NEUROFIBROMATOSIS-1 GLIOMA FORMATION AND GROWTH. *Neuro-Oncology* **15**: 175-175.
456. Pong WW, Walker J, Wylie T, Magrini V, Luo J, Emnett RJ, Choi J, Cooper ML, Griffith M, Griffith OL et al. 2013. F11R Is a Novel Monocyte Prognostic Biomarker for Malignant Glioma. *Plos One* **8**(10).
457. Powell BC, Jiang L, Muzny DM, Trevino LR, Dreyer ZE, Strong LC, Wheeler DA, Gibbs RA, Plon SE. 2012. Identification of TP53 as an acute lymphocytic leukemia susceptibility gene through exome sequencing *PediatrBlood Cancer*.
458. Prado-Martinez J, Sudmant PH, Kidd JM, Li H, Kelley JL, Lorente-Galdos B, Veeramah KR, Woerner AE, O'Connor TD, Santpere G et al. 2013. Great ape genetic diversity and population history. *Nature* **499**(7459): 471-475.
459. Puffenberger EG, Jinks RN, Sougnez C, Cibulskis K, Willert RA, Achilly NP, Cassidy RP, Fiorentini CJ, Heiken KF, Lawrence JJ et al. 2012. Genetic mapping and exome sequencing identify variants associated with five novel diseases. *PLoS One* **7**(1): e28936.
460. Puffenberger EG, Jinks RN, Wang H, Xin B, Fiorentini C, Sherman EA, Degrazio D, Shaw C, Sougnez C, Cibulskis K et al. 2012. A homozygous missense mutation in HERC2 associated with global developmental delay and autism spectrum disorder. *Hum Mutat* **33**(12): 1639-1646.
461. Pugh TJ, Morozova O, Attiyeh EF, Asgharzadeh S, Wei JS, Auclair D, Carter SL, Cibulskis K, Hanna M, Kiezun A et al. 2013. The genetic landscape of high-risk neuroblastoma. *Nat Genet* **45**(3): 279-284.
462. Pugh TJ, Weeraratne SD, Archer TC, Pomeranz Krummel DA, Auclair D, Bochicchio J, Carneiro MO, Carter SL, Cibulskis K, Erlich RL et al. 2012. Medulloblastoma exome sequencing uncovers subtype-specific somatic mutations. *Nature* **488**(7409): 106-110.
463. Purcell SM, Moran JL, Fromer M, Ruderfer D, Solovieff N, Roussos P, O'Dushlaine C, Chambert K, Bergen SE, Kahler A et al. 2014. A polygenic burden of rare disruptive mutations in schizophrenia. *Nature* **506**(7487): 185-190.
464. Qin X, Galloway-Pena JR, Sillanpaa J, Hyeob RJ, Nallapareddy SR, Chowdhury S, Bourgogne A, Choudhury T, Munzy DM, Buhay CJ et al. 2012. Complete genome sequence of Enterococcus faecium strain TX16 and comparative genomic analysis of Enterococcus faecium genomes *BMCMicrobiol* **12**(1): 135.
465. Quesada V, Velasco G, Puente XS, Warren WC, Lopez-Otin C. 2010. Comparative genomic analysis of the zebra finch degradome provides new insights into evolution of proteases in birds and mammals. *Bmc Genomics* **11**.
466. Rabani M, Levin JZ, Fan L, Adiconis X, Raychowdhury R, Garber M, Gnirke A, Nusbaum C, Hacohen N, Friedman N et al. 2011. Metabolic labeling of RNA uncovers principles of RNA production and degradation dynamics in mammalian cells. *Nat Biotechnol* **29**(5): 436-442.
467. Radmanesh F, Caglayan AO, Silhavy JL, Yilmaz C, Cantagrel V, Omar T, Rosti B, Kaymakcalan H, Gabriel S, Li M et al. 2013. Mutations in LAMB1 cause cobblestone brain malformation without muscular or ocular abnormalities. *Am J Hum Genet* **92**(3): 468-474.
468. Rainger J, Pehlivan D, Johansson S, Bengani H, Sanchez-Pulido L, Williamson KA, Ture M, Barker H, Rosendahl K, Spranger J et al. 2014. Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. *Am J Hum Genet* **94**(6): 915-923.
469. Ramsingh G, Koboldt DC, Trissal M, Chiappinelli KB, Wylie T, Koul S, Chang L-W, Nagarajan R, Fehniger TA, Goodfellow P et al. 2010. Complete characterization of the microRNAome in a patient with acute myeloid leukemia. *Blood* **116**(24): 5316-5326.
470. Ramsingh G, Shen D, Lamprecht T, Heath S, Fulton RS, Mardis ER, Ding L, Westervelt P, Welch JS, Walter MJ et al. 2012. Whole Genome Sequencing of Therapy-Related Acute Myeloid Leukemia. *Blood* **120**(21).
471. Rao RU, Huang Y, Abubucker S, Heinz M, Crosby SD, Mitreva M, Weil GJ. 2012. Effects of doxycycline on gene expression in Wolbachia and Brugia malayi adult female worms in vivo. *Journal of biomedical science* **19**: 21-21.
472. Reekie K, Metspalu A, Chanock SJ, Liu ET, Mardis ER, Scherer SW, Kwok P-Y, Brookes AJ. 2010. HGV2009 Meeting: Bigger and Better Studies Provide More Answers and More Questions. *Human Mutation* **31**(7): 886-888.
473. Reid JG, Carroll A, Veeraraghavan N, Dahdouli M, Sundquist A, English A, Bainbridge M, White S, Salerno W, Buhay C et al. 2014. Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline *BMCBioinformatics* **15**: 30.
474. Reis-Filho JS, Mackay A, Wilkerson PM, Lambros MB, Gauthier A, Mariani O, Duprez R, Rodrigues DN, Mandour M, Maher C et al. 2012. Identification of Fusion Genes in Papillary Carcinomas of the Breast. *Modern Pathology* **25**: 61A-62A.
475. Renfree MB Papenfuss AT Deakin JE Lindsay J Heider T Belov K Rens W Waters PD Pharo EA Shaw G et al. 2011. Genome sequence of an Australian kangaroo, Macropus eugenii, provides insight into the evolution of mammalian reproduction and development *Genome Biol* **12**(12): 414.
476. Rhind N, Chen Z, Yassour M, Thompson DA, Haas BJ, Habib N, Wapinski I, Roy S, Lin MF, Heiman DI et al. 2011. Comparative functional genomics of the fission yeasts. *Science* **332**(6032): 930-936.
477. Ribeiro FJ, Przybylski D, Yin S, Sharpe T, Gnerre S, Abouelleil A, Berlin AM, Montmayeur A, Shea TP, Walker BJ et al. 2012. Finished bacterial genomes from shotgun sequence data. *Genome Res* **22**(11): 2270-2277.
478. Rivas MA, Beaudoin M, Gardet A, Stevens C, Sharma Y, Zhang CK, Boucher G, Ripke S, Ellinghaus D, Burtt N et al. 2011. Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. *Nat Genet* **43**(11): 1066-1073.
479. Roberts RJ, Carneiro MO, Schatz MC. 2013. The advantages of SMRT sequencing. *Genome Biol* **14**(6): 405.
480. Robinson G, Parker M, Kranenburg TA, Lu C, Chen X, Ding L, Phoenix TN, Hedlund E, Wei L, Zhu X et al. 2012. Novel mutations target distinct subgroups of medulloblastoma. *Nature* **488**(7409): 43-48.
481. Robinson GW, Parker M, Kranenburg T, Lu C, Chen X, Ding L, Phoenix T, Huether R, Thiruvenkatam R, Wang J et al. 2012. Use of whole genome sequencing to identify novel mutations in distinct subgroups of medulloblastoma. *Journal of Clinical Oncology* **30**(15).
482. Roca H, Hernandez J, Weidner S, McEachin RC, Fuller D, Sud S, Schumann T, Wilkinson JE, Zaslavsky A, Li H et al. 2013. Transcription Factors OVOL1 and OVOL2 Induce the Mesenchymal to Epithelial Transition in Human Cancer. *Plos One* **8**(10).
483. Rogers AJ, Maher CO, Schunk JE, Quayle K, Jacobs E, Lichenstein R, Powell E, Miskin M, Dayan P, Holmes JF et al. 2013. Incidental Findings in Children With Blunt Head Trauma Evaluated With Cranial CT Scans. *Pediatrics* **132**(2): E356-E363.
484. Rogers J. 2013. In transition: primate genomics at a time of rapid change. *ILARJ* **54**(2): 224-233.
485. Rogers J, Gibbs RA. 2014. Comparative primate genomics: emerging patterns of genome content and dynamics. *NatRevGenet*.
486. Rogers J, Raveendran M, Fawcett GL, Fox AS, Shelton SE, Oler JA, Cheverud J, Muzny DM, Gibbs RA, Davidson RJ et al. 2013. CRHR1 genotypes, neural circuits and the diathesis for anxiety and depression *MolPsychiatry* **18**(6): 700-707.
487. Rosa BA, Jasmer DP, Mitreva M. 2014. Genome-Wide Tissue-Specific Gene Expression, Co-expression and Regulation of Co-expressed Genes in Adult Nematode Ascaris suum. *Plos Neglected Tropical Diseases* **8**(2).
488. Rosen LB, Freeman AF, Yang LM, Jutivorakool K, Olivier KN, Angkasekwinai N, Suputtamongkol Y, Bennett JE, Pyrgos V, Williamson PR et al. 2013. Anti-GM-CSF Autoantibodies in Patients with Cryptococcal Meningitis. *Journal of Immunology* **190**(8): 3959-3966.
489. Rosenberg JE, Bambury RM, Van Allen EM, Drabkin HA, Lara PN, Jr., Harzstark AL, Wagle N, Figlin RA, Smith GW, Garraway LA et al. 2014. A phase II trial of AS1411 (a novel nucleolin-targeted DNA aptamer) in metastatic renal cell carcinoma. *Investigational new drugs* **32**(1): 178-187.
490. Ross JA, Koboldt DC, Staisch JE, Chamberlin HM, Gupta BP, Miller RD, Baird SE, Haag ES. 2011. Caenorhabditis briggsae Recombinant Inbred Line Genotypes Reveal Inter-Strain Incompatibility and the Evolution of Recombination. *Plos Genetics* **7**(7).
491. Ross MG, Russ C, Costello M, Hollinger A, Lennon NJ, Hegarty R, Nusbaum C, Jaffe DB. 2013. Characterizing and measuring bias in sequence data. *Genome Biol* **14**(5): R51.
492. Rubin CJ, Zody MC, Eriksson J, Meadows JR, Sherwood E, Webster MT, Jiang L, Ingman M, Sharpe T, Ka S et al. 2010. Whole-genome resequencing reveals loci under selection during chicken domestication. *Nature* **464**(7288): 587-591.
493. Russler-Germain DA, Spencer DH, Young MA, Lamprecht TL, Miller CA, Fulton R, Meyer MR, Erdmann-Gilmore P, Townsend RR, Wilson RK et al. 2014. The R882H DNMT3A Mutation Associated with AML Dominantly Inhibits Wild-Type DNMT3A by Blocking Its Ability to Form Active Tetramers. *Cancer Cell* **25**(4): 442-454.
494. Sankaran VG, Ghazvinian R, Do R, Thiru P, Vergilio JA, Beggs AH, Sieff CA, Orkin SH, Nathan DG, Lander ES et al. 2012. Exome sequencing identifies GATA1 mutations resulting in Diamond-Blackfan anemia. *The Journal of clinical investigation* **122**(7): 2439-2443.
495. Sano D, Xie TX, Ow TJ, Zhao M, Pickering CR, Zhou G, Sandulache VC, Wheeler DA, Gibbs RA, Caulin C et al. 2011. Disruptive TP53 mutation is associated with aggressive disease characteristics in an orthotopic murine model of oral tongue cancer. *ClinCancer Res* **17**(21): 6658-6670.
496. Saraf SR, Kempler KE, Dugger DR, Speiser DI, Oakely TH, Wilson RK, Battelle BA. 2014. Opsin expression in the Limulus visual system: Too many opsins? *Integrative and Comparative Biology* **54**: E344-E344.
497. Saulnier DM, Riehle K, Mistretta TA, Diaz MA, Mandal D, Raza S, Weidler EM, Qin X, Coarfa C, Milosavljevic A et al. 2011. Gastrointestinal microbiome signatures of pediatric patients with irritable bowel syndrome *Gastroenterology* **141**(5): 1782-1791.
498. Scally A, Dutheil JY, Hillier LW, Jordan GE, Goodhead I, Herrero J, Hobolth A, Lappalainen T, Mailund T, Marques-Bonet T et al. 2012. Insights into hominid evolution from the gorilla genome sequence. *Nature* **483**(7388): 169-175.
499. Schaaf CP, Sabo A, Sakai Y, Crosby J, Muzny D, Hawes A, Lewis L, Akbar H, Varghese R, Boerwinkle E et al. 2011. Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. *HumMolGenet*.
500. Schafer CM, Campbell NG, Cai G, Yu F, Makarov V, Yoon S, Daly MJ, Gibbs RA, Schellenberg GD, Devlin B et al. 2013. Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. *Genomics*.
501. Schartl M, Walter RB, Shen Y, Garcia T, Catchen J, Amores A, Braasch I, Chalopin D, Volff J-N, Lesch K-P et al. 2013. The genome of the platyfish, Xiphophorus maculatus, provides insights into evolutionary adaptation and several complex traits. *Nature Genetics* **45**(5): 567-U150.
502. Schloissnig S, Arumugam M, Sunagawa S, Mitreva M, Tap J, Zhu A, Waller A, Mende DR, Kultima JR, Martin J et al. 2013. Genomic variation landscape of the human gut microbiome. *Nature* **493**(7430): 45-50.
503. Schuettpelz L, Link DC, Shen D, Walter MJ, Koboldt DC, Dooling DJ, Fulton RS, Schmidt H, Maupin R, O'Laughlin M et al. 2010. DNA Sequence of the Cancer Genome of a Patient with Therapy-Related Acute Myeloid Leukemia. *Blood* **116**(21): 256-257.
504. Schuster SC, Miller W, Ratan A, Tomsho LP, Giardine B, Kasson LR, Harris RS, Petersen DC, Zhao F, Qi J et al. 2010. Complete Khoisan and Bantu genomes from southern Africa. *Nature* **463**(7283): 943-947.
505. Sczesnak A, Segata N, Qin X, Gevers D, Petrosino JF, Huttenhower C, Littman DR, Ivanov II. 2011. The genome of th17 cell-inducing segmented filamentous bacteria reveals extensive auxotrophy and adaptations to the intestinal environment *Cell HostMicrobe* **10**(3): 260-272.
506. Sebe-Pedros A, Irimia M, Del Campo J, Parra-Acero H, Russ C, Nusbaum C, Blencowe BJ, Ruiz-Trillo I. 2013. Regulated aggregative multicellularity in a close unicellular relative of metazoa. *eLife* **2**: e01287.
507. Segata N, Haake SK, Mannon P, Lemon KP, Waldron L, Gevers D, Huttenhower C, Izard J. 2012. Composition of the adult digestive tract bacterial microbiome based on seven mouth surfaces, tonsils, throat and stool samples. *Genome Biol* **13**(6): R42.
508. Service SK, Teslovich TM, Fuchsberger C, Ramensky V, Yajnik P, Koboldt DC, Larson DE, Zhang Q, Lin L, Welch R et al. 2014. Re-sequencing Expands Our Understanding of the Phenotypic Impact of Variants at GWAS Loci. *Plos Genetics* **10**(1).
509. Severance S, Rajagopal A, Rao AU, Cerqueira GC, Mitreva M, El-Sayed NM, Krause M, Hamza I. 2010. Genome-Wide Analysis Reveals Novel Genes Essential for Heme Homeostasis in Caenorhabditis elegans. *Plos Genetics* **6**(7).
510. Shaaban S, Ramos-Platt L, Gilles FH, Chan WM, Andrews C, De Girolami U, Demer J, Engle EC. 2013. RYR1 mutations as a cause of ophthalmoplegia, facial weakness, and malignant hyperthermia. *JAMA ophthalmology* **131**(12): 1532-1540.
511. Shaffer CD, Alvarez C, Bailey C, Barnard D, Bhalla S, Chandrasekaran C, Chandrasekaran V, Chung H-M, Dorer DR, Du C et al. 2010. The Genomics Education Partnership: Successful Integration of Research into Laboratory Classes at a Diverse Group of Undergraduate Institutions. *Cbe-Life Sciences Education* **9**(1): 55-69.
512. Shaffer CD, Alvarez CJ, Bednarski AE, Dunbar D, Goodman AL, Reinke C, Rosenwald AG, Wolyniak MJ, Bailey C, Barnard D et al. 2014. A Course-Based Research Experience: How Benefits Change with Increased Investment in Instructional Time. *Cbe-Life Sciences Education* **13**(1): 111-130.
513. Shaffer HB, Minx P, Warren DE, Shedlock AM, Thomson RC, Valenzuela N, Abramyan J, Amemiya CT, Badenhorst D, Biggar KK et al. 2013. The western painted turtle genome, a model for the evolution of extreme physiological adaptations in a slowly evolving lineage. *Genome Biology* **14**(3).
514. Shalev SA, Tenenbaum-Rakover Y, Horovitz Y, Paz VP, Ye H, Carmody D, Highland HM, Boerwinkle E, Hanis CL, Muzny DM et al. 2013. Microcephaly, epilepsy, and neonatal diabetes due to compound heterozygous mutations in IER3IP1: insights into the natural history of a rare disorder *PediatrDiabetes*.
515. Shapiro JR, Lietman C, Grover M, Lu JT, Nagamani SC, Dawson BC, Baldridge DM, Bainbridge MN, Cohn DH, Blazo M et al. 2013. Phenotypic variability of osteogenesis imperfecta type V caused by an IFITM5 mutation *JBone MinerRes* **28**(7): 1523-1530.
516. Shen Y, Catchen J, Garcia T, Amores A, Beldorth I, Wagner J, Zhang Z, Postlethwait J, Warren W, Schartl M et al. 2012. Identification of transcriptome SNPs between Xiphophorus lines and species for assessing allele specific gene expression within F-1 interspecies hybrids. *Comparative Biochemistry and Physiology C-Toxicology & Pharmacology* **155**(1): 102-108.
517. Shen Y, Garcia T, Pabuwal V, Boswell M, Pasquali A, Beldorth I, Warren W, Schartl M, Cresko WA, Walter RB. 2013. Alternative strategies for development of a reference transcriptome for quantification of allele specific expression in organisms having sparse genomic resources. *Comparative Biochemistry and Physiology D-Genomics & Proteomics* **8**(1): 11-16.
518. Shen Y, Wan Z, Coarfa C, Drabek R, Chen L, Ostrowski EA, Liu Y, Weinstock GM, Wheeler DA, Gibbs RA et al. 2010. A SNP discovery method to assess variant allele probability from next-generation resequencing data. *Genome Res* **20**(2): 273-280.
519. Shirak A, Grabherr M, Di Palma F, Lindblad-Toh K, Hulata G, Ron M, Kocher TD, Seroussi E. 2010. Identification of repetitive elements in the genome of Oreochromis niloticus: tilapia repeat masker. *Marine biotechnology* **12**(2): 121-125.
520. Singh R, Ong-Abdullah M, Low E-TL, Manaf MAA, Rosli R, Nookiah R, Ooi LC-L, Ooi S-E, Chan K-L, Halim MA et al. 2013. Oil palm genome sequence reveals divergence of interfertile species in Old and New Worlds. *Nature* **500**(7462): 335-+.
521. Slavoff SA, Mitchell AJ, Schwaid AG, Cabili MN, Ma J, Levin JZ, Karger AD, Budnik BA, Rinn JL, Saghatelian A. 2013. Peptidomic discovery of short open reading frame-encoded peptides in human cells. *Nature chemical biology* **9**(1): 59-64.
522. Sloan DB, Nakabachi A, Richards S, Qu J, Murali SC, Gibbs RA, Moran NA. 2014. Parallel histories of horizontal gene transfer facilitated extreme reduction of endosymbiont genomes in sap-feeding insects *MolBiolEvol*.
523. Smajs D, Zobanikova M, Strouhal M, Cejkova D, Dugan-Rocha S, Pospisilova P, Norris SJ, Albert T, Qin X, Hallsworth-Pepin K et al. 2011. Complete Genome Sequence of Treponema paraluiscuniculi, Strain Cuniculi A: The Loss of Infectivity to Humans Is Associated with Genome Decay *PLoSOne* **6**(5): e20415.
524. Smith JJ, Kuraku S, Holt C, Sauka-Spengler T, Jiang N, Campbell MS, Yandell MD, Manousaki T, Meyer A, Bloom OE et al. 2013. Sequencing of the sea lamprey (Petromyzon marinus) genome provides insights into vertebrate evolution. *Nature Genetics* **45**(4): 415-421.
525. Soblik H, Younis AE, Mitreva M, Renard BY, Kirchner M, Geisinger F, Steen H, Brattig NW. 2011. Life Cycle Stage-resolved Proteomic Analysis of the Excretome/Secretome from Strongyloides ratti-Identification of Stage-specific Proteases. *Molecular & Cellular Proteomics* **10**(12).
526. Stark M, Lukaszuk J, Prawitz A, Salacinski A. 2012. Protein timing and its effects on muscular hypertrophy and strength in individuals engaged in weight-training. *JIntSocSports Nutr* **9**(1): 54.
527. Stong N, Deng Z, Gupta R, Hu S, Paul S, Weiner AK, Eichler EE, Graves T, Fronick CC, Courtney L et al. 2014. Subtelomeric CTCF and cohesin binding site organization using improved subtelomere assemblies and a novel annotation pipeline. *Genome Research* **24**(6): 1039-1050.
528. Stransky N, Egloff AM, Tward AD, Kostic AD, Cibulskis K, Sivachenko A, Kryukov GV, Lawrence MS, Sougnez C, McKenna A et al. 2011. The mutational landscape of head and neck squamous cell carcinoma. *Science* **333**(6046): 1157-1160.
529. Stray-Pedersen A, Backe PH, Sorte HS, Morkrid L, Chokshi NY, Erichsen HC, Gambin T, Elgstoen KB, Bjoras M, Wlodarski MW et al. 2014. PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. *Am J Hum Genet*.
530. Sudmant PH, Kitzman JO, Antonacci F, Alkan C, Malig M, Tsalenko A, Sampas N, Bruhn L, Shendure J, Eichler EE. 2010. Diversity of human copy number variation and multicopy genes *Science* **330**(6004): 641-646.
531. Suen G, Teiling C, Li L, Holt C, Abouheif E, Bornberg-Bauer E, Bouffard P, Caldera EJ, Cash E, Cavanaugh A et al. 2011. The Genome Sequence of the Leaf-Cutter Ant Atta cephalotes Reveals Insights into Its Obligate Symbiotic Lifestyle. *Plos Genetics* **7**(2).
532. Suga H, Chen Z, de Mendoza A, Sebe-Pedros A, Brown MW, Kramer E, Carr M, Kerner P, Vervoort M, Sanchez-Pons N et al. 2013. The Capsaspora genome reveals a complex unicellular prehistory of animals. *Nat Commun* **4**: 2325.
533. Sule G, Campeau PM, Zhang VW, Nagamani SC, Dawson BC, Grover M, Bacino CA, Sutton VR, Brunetti-Pierri N, Lu JT et al. 2013. Next-generation sequencing for disorders of low and high bone mineral density *OsteoporosInt*.
534. Svardal H, Huang YS, Schmitt CA, Jasinska AJ, Jung Y, Wasserscheid J, Jureticx N, Muller-Trutwin M, Jacquelin B, Antonio M et al. 2014. The evolutionary history of the genus Chlorocebus inferred from whole genome sequencing. *American Journal of Physical Anthropology* **153**: 250-250.
535. Swart EC, Bracht JR, Magrini V, Minx P, Chen X, Zhou Y, Khurana JS, Goldman AD, Nowacki M, Schotanus K et al. 2013. The Oxytricha trifallax Macronuclear Genome: A Complex Eukaryotic Genome with 16,000 Tiny Chromosomes. *Plos Biology* **11**(1).
536. Swart EC, Nowacki M, Shum J, Stiles H, Higgins BP, Doak TG, Schotanus K, Magrini VJ, Minx P, Mardis ER et al. 2012. The Oxytricha trifallax Mitochondrial Genome. *Genome Biology and Evolution* **4**(2): 136-154.
537. Tamborero D, Gonzalez-Perez A, Perez-Llamas C, Deu-Pons J, Kandoth C, Reimand J, Lawrence MS, Getz G, Bader GD, Ding L et al. 2013. Comprehensive identification of mutational cancer driver genes across 12 tumor types. *Scientific Reports* **3**.
538. Tang YT, Gao X, Rosa BA, Abubucker S, Hallsworth-Pepin K, Martin J, Tyagi R, Heizer E, Zhang X, Bhonagiri-Palsikar V et al. 2014. Genome of the human hookworm Necator americanus. *Nature Genetics* **46**(3): 261-+.
539. Taylor CM, Abubucker S, Wang Z, Martin J, Mitreva M. 2010. PAN PHYLUM ANALYSIS OF PROTEIN-PROTEIN INTERACTIONS REVEALS POTENTIAL DRUG TARGETS FOR HELMINTHES. *American Journal of Tropical Medicine and Hygiene* **83**(5): 353-353.
540. Taylor CM, Fischer K, Abubucker S, Wang Z, Martin J, Jiang D, Magliano M, Rosso M-N, Li B-W, Fischer PU et al. 2011. Targeting Protein-Protein Interactions for Parasite Control. *Plos One* **6**(4).
541. Taylor CM, Martin J, Rao RU, Powell K, Abubucker S, Mitreva M. 2013. Using Existing Drugs as Leads for Broad Spectrum Anthelmintics Targeting Protein Kinases. *Plos Pathogens* **9**(2).
542. Taylor CM, Wang Q, Rosa BA, Huang SC-C, Powell K, Schedl T, Pearce EJ, Abubucker S, Mitreva M. 2013. Discovery of Anthelmintic Drug Targets and Drugs Using Chokepoints in Nematode Metabolic Pathways. *Plos Pathogens* **9**(8).
543. Tengvall K, Kierczak M, Bergvall K, Olsson M, Frankowiack M, Farias FH, Pielberg G, Carlborg O, Leeb T, Andersson G et al. 2013. Genome-wide analysis in German shepherd dogs reveals association of a locus on CFA 27 with atopic dermatitis. *PLoS Genet* **9**(5): e1003475.
544. The TG, Hdl Working Group of the Exome Sequencing Project NHL, Blood I. 2014. Loss-of-Function Mutations in APOC3, Triglycerides, and Coronary Disease. *The New England journal of medicine*.
545. Thorvaldsdottir H, Robinson JT, Mesirov JP. 2013. Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. *Brief Bioinform* **14**(2): 178-192.
546. Tomasson MH, Shen D, Hucthagowder V, Schierding W, Mullins CD, Fiala M, Hall IM, Wallis J, Fulton RS, Fulton LA et al. 2012. Whole Genome Sequencing Reveals Novel Recurring Somatic Mutations Affecting HUWE1 and DIAPH2 Genes in Multiple Myeloma. *Blood* **120**(21).
547. Tripathi P, Wang Y, Coussens M, Manda KR, Casey AM, Lin C, Poyo E, Pfeifer JD, Basappa N, Bates CM et al. 2014. Activation of NFAT signaling establishes a tumorigenic microenvironment through cell autonomous and non-cell autonomous mechanisms. *Oncogene* **33**(14): 1840-1849.
548. Tu Q, Cameron RA, Worley KC, Gibbs RA, Davidson EH. 2012. Gene structure in the sea urchin Strongylocentrotus purpuratus based on transcriptome analysis. *Genome Res*.
549. Turabelidze G, Lawrence SJ, Gao H, Sodergren E, Weinstock GM, Abubucker S, Wylie T, Mitreva M, Shaikh N, Gautom R et al. 2013. Precise Dissection of an Escherichia coli O157:H7 Outbreak by Single Nucleotide Polymorphism Analysis. *Journal of Clinical Microbiology* **51**(12): 3950-3954.
550. Valente EM, Logan CV, Mougou-Zerelli S, Lee JH, Silhavy JL, Brancati F, Iannicelli M, Travaglini L, Romani S, Illi B et al. 2010. Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. *Nat Genet* **42**(7): 619-625.
551. Van Allen EM, Foye A, Wagle N, Kim W, Carter SL, McKenna A, Simko JP, Garraway LA, Febbo PG. 2014. Successful whole-exome sequencing from a prostate cancer bone metastasis biopsy. *Prostate cancer and prostatic diseases* **17**(1): 23-27.
552. Van Allen EM, Wagle N, Stojanov P, Perrin DL, Cibulskis K, Marlow S, Jane-Valbuena J, Friedrich DC, Kryukov G, Carter SL et al. 2014. Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin-embedded tumor samples to guide precision cancer medicine. *Nature medicine* **20**(6): 682-688.
553. Van Allen EM, Wagle N, Sucker A, Treacy DJ, Johannessen CM, Goetz EM, Place CS, Taylor-Weiner A, Whittaker S, Kryukov GV et al. 2014. The genetic landscape of clinical resistance to RAF inhibition in metastatic melanoma. *Cancer Discov* **4**(1): 94-109.
554. van Berkum NL, Lieberman-Aiden E, Williams L, Imakaev M, Gnirke A, Mirny LA, Dekker J, Lander ES. 2010. Hi-C: a method to study the three-dimensional architecture of genomes. *Journal of visualized experiments : JoVE*(39).
555. Vatta M, Niu Z, Lupski JR, Putnam P, Spoonamore KG, Fang P, Eng CM, Willis AS. 2013. Evidence for replicative mechanism in a CHD7 rearrangement in a patient with CHARGE syndrome. *AmJMedGenetA* **161A**(12): 3182-3186.
556. Vaysse A, Ratnakumar A, Derrien T, Axelsson E, Rosengren Pielberg G, Sigurdsson S, Fall T, Seppala EH, Hansen MS, Lawley CT et al. 2011. Identification of genomic regions associated with phenotypic variation between dog breeds using selection mapping. *PLoS Genet* **7**(10): e1002316.
557. Velusamy T, Palanisamy N, Kalyana-Sundaram S, Sahasrabuddhe AA, Maher CA, Robinson DR, Bahler DW, Cornell TT, Wilson TE, Lim MS et al. 2013. Recurrent reciprocal RNA chimera involving YPEL5 and PPP1CB in chronic lymphocytic leukemia. *Proceedings of the National Academy of Sciences of the United States of America* **110**(8): 3035-3040.
558. Venkatesh B, Lee AP, Ravi V, Maurya AK, Lian MM, Swann JB, Ohta Y, Flajnik MF, Sutoh Y, Kasahara M et al. 2014. Elephant shark genome provides unique insights into gnathostome evolution. *Nature* **505**(7482): 174-179.
559. Venkatesh B, Ravi V, Lee AP, Warren WC, Brenner S. 2013. Basal Vertebrates Clarify the Evolutionary History of Ciliopathy-Associated Genes Tmem138 and Tmem216. *Molecular Biology and Evolution* **30**(1): 62-65.
560. Ventura M, Catacchio CR, Alkan C, Marques-Bonet T, Sajjadian S, Graves TA, Hormozdiari F, Navarro A, Malig M, Baker C et al. 2011. Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Research* **21**(10): 1640-1649.
561. Ventura M, Catacchio CR, Sajjadian S, Vives L, Sudmant PH, Marques-Bonet T, Graves TA, Wilson RK, Eichler EE. 2012. The evolution of African great ape subtelomeric heterochromatin and the fusion of human chromosome 2. *Genome Research* **22**(6): 1036-1049.
562. Verhaak RG, Hoadley KA, Purdom E, Wang V, Qi Y, Wilkerson MD, Miller CR, Ding L, Golub T, Mesirov JP et al. 2010. Integrated genomic analysis identifies clinically relevant subtypes of glioblastoma characterized by abnormalities in PDGFRA, IDH1, EGFR, and NF1 *Cancer Cell* **17**(1): 98-110.
563. Verhaak RG, Tamayo P, Yang JY, Hubbard D, Zhang H, Creighton CJ, Fereday S, Lawrence M, Carter SL, Mermel CH et al. 2013. Prognostically relevant gene signatures of high-grade serous ovarian carcinoma. *The Journal of clinical investigation* **123**(1): 517-525.
564. Verhaak RGW, Hoadley KA, Purdom E, Wang V, Qi Y, Wilkerson MD, Miller CR, Ding L, Golub T, Mesirov JP et al. 2010. Integrated Genomic Analysis Identifies Clinically Relevant Subtypes of Glioblastoma Characterized by Abnormalities in PDGFRA, IDH1, EGFR, and NF1. *Cancer Cell* **17**(1): 98-110.
565. Vesely MD, Matsushita H, Koboldt DC, Rickert CG, Mardis ER, Schreiber RD. 2012. Cancer exome analysis reveals a T cell dependent mechanism of cancer immunoediting. *Journal of Investigative Dermatology* **132**: S97-S97.
566. Voora D, Koboldt DC, King CR, Lenzini PA, Eby CS, Porche-Sorbet R, Deych E, Crankshaw M, Milligan PE, McLeod HL et al. 2010. A Polymorphism in the VKORC1 Regulator Calumenin Predicts Higher Warfarin Dose Requirements in African Americans. *Clinical Pharmacology & Therapeutics* **87**(4): 445-451.
567. Wagle N, Berger MF, Davis MJ, Blumenstiel B, Defelice M, Pochanard P, Ducar M, Van Hummelen P, Macconaill LE, Hahn WC et al. 2012. High-throughput detection of actionable genomic alterations in clinical tumor samples by targeted, massively parallel sequencing. *Cancer Discov* **2**(1): 82-93.
568. Wagle N, Van Allen EM, Treacy DJ, Frederick DT, Cooper ZA, Taylor-Weiner A, Rosenberg M, Goetz EM, Sullivan RJ, Farlow DN et al. 2014. MAP kinase pathway alterations in BRAF-mutant melanoma patients with acquired resistance to combined RAF/MEK inhibition. *Cancer Discov* **4**(1): 61-68.
569. Walter MJ, Ding L, Shen D, Shao J, Grillot M, McLellan M, Fulton R, Schmidt H, Kalicki-Veizer J, O'Laughlin M et al. 2011. Recurrent DNMT3A mutations in patients with myelodysplastic syndromes. *Leukemia* **25**(7): 1153-1158.
570. Walter MJ, Shen D, Ding L, Shao J, Koboldt DC, Chen K, Larson DE, McLellan MD, Dooling D, Abbott R et al. 2012. Clonal Architecture of Secondary Acute Myeloid Leukemia. *New England Journal of Medicine* **366**(12): 1090-1098.
571. Walter MJ, Shen D, Ding L, Shao J, Witowski S, Chen K, Koboldt DC, Dooling DJ, Maupin R, Fulton RS et al. 2010. Detection of Novel Mutations In MDS/AML by Whole Genome Sequencing. *Blood* **116**(21): 136-136.
572. Walter MJ, Shen D, Shao J, Ding L, Grillot M, McLellan M, Fulton R, Schmidt H, Kalicki-Veizer J, O'Laughlin M et al. 2010. Recurrent DNMT3A Mutations In Patients with Myelodysplastic Syndrome. *Blood* **116**(21): 267-268.
573. Walter MJ, Shen D, Shao J, Ding L, White BS, Kandoth C, Miller CA, Niu B, McLellan MD, Dees ND et al. 2013. Clonal diversity of recurrently mutated genes in myelodysplastic syndromes. *Leukemia* **27**(6): 1275-1282.
574. Wang C, Zhan X, Bragg-Gresham J, Kang HM, Stambolian D, Chew EY, Branham KE, Heckenlively J, Fulton R, Wilson RK et al. 2014. Ancestry estimation and control of population stratification for sequence-based association studies. *Nature Genetics* **46**(4): 409-+.
575. Wang H, Chattopadhyay A, Li Z, Daines B, Li Y, Gao C, Gibbs RA, Zhang K, Chen R. 2010. Rapid identification of heterozygous mutations in Drosophila melanogaster using genomic capture sequencing *Genome Res* **20**(7): 981-988.
576. Wang H, Chen X, Dudinsky L, Patenia C, Chen Y, Li Y, Wei Y, Abboud EB, Al-Rajhi AA, Lewis RA et al. 2011. Exome capture sequencing identifies a novel mutation in BBS4 *MolVis* **17**: 3529-3540.
577. Wang J, Czech B, Crunk A, Wallace A, Mitreva M, Hannon GJ, Davis RE. 2011. Deep small RNA sequencing from the nematode Ascaris reveals conservation, functional diversification, and novel developmental profiles. *Genome Research* **21**(9): 1462-1477.
578. Wang J, Mitreva M, Berriman M, Thorne A, Magrini V, Koutsovoulos G, Kumar S, Blaxter ML, Davis RE. 2012. Silencing of Germline-Expressed Genes by DNA Elimination in Somatic Cells. *Developmental Cell* **23**(5): 1072-1080.
579. Wang J, Mullighan CG, Easton J, Roberts S, Heatley SL, Ma J, Rusch MC, Chen K, Harris CC, Ding L et al. 2011. CREST maps somatic structural variation in cancer genomes with base-pair resolution. *Nature Methods* **8**(8): 652-U669.
580. Wang L, Lawrence MS, Wan Y, Stojanov P, Sougnez C, Stevenson K, Werner L, Sivachenko A, DeLuca DS, Zhang L et al. 2011. SF3B1 and other novel cancer genes in chronic lymphocytic leukemia. *The New England journal of medicine* **365**(26): 2497-2506.
581. Wang L, Swierczek SI, Drummond J, Hickman K, Walker K, Doddapaneni H, Muzny DM, Gibbs RA, Wheeler DA, Prchal JT. 2014. Whole-exome sequencing of polycythemia vera revealed novel driver genes and somatic mutation shared by T-cells and granulocytes *Leukemia*.
582. Wang L, Swierczek SI, Lanikova L, Kim SJ, Hickman K, Walker K, Wang K, Drummond J, Doddapaneni H, Reid JG et al. 2014. The relationship of JAK2(V617F) and acquired UPD at chromosome 9p in polycythemia vera *Leukemia* **28**(4): 938-941.
583. Wang L, Yamaguchi S, Burstein MD, Terashima K, Chang K, Ng HK, Nakamura H, He Z, Doddapaneni H, Lewis L et al. 2014. Novel somatic and germline mutations in intracranial germ cell tumours. *Nature*.
584. Wang QY, Song J, Gibbs RA, Boerwinkle E, Dong JF, Yu FL. 2012. Characterizing Polymorphisms and Allelic Diversity of von Willebrand Factor Gene in the 1000 Genomes. *JThrombHaemost*.
585. Wang X, Wang H, Cao M, Li Z, Chen X, Patenia C, Gore A, Abboud EB, Al-Rajhi AA, Lewis RA et al. 2011. Whole-exome sequencing identifies ALMS1, IQCB1, CNGA3, and MYO7A mutations in patients with Leber congenital amaurosis. *HumMutat* **32**(12): 1450-1459.
586. Wang Y, Lu J, Yu J, Gibbs RA, Yu F. 2013. An integrative variant analysis pipeline for accurate genotype/haplotype inference in population NGS data *Genome Res*.
587. Wang Y, Zhang Y, Yang J, Ni X, Liu S, Li Z, Hodges SE, Fisher WE, Brunicardi FC, Gibbs RA et al. 2012. Genomic Sequencing of Key Genes in Mouse Pancreatic Cancer Cells *CurrMolMed*.
588. Wang Z, Abubucker S, Martin J, Wilson RK, Hawdon J, Mitreva M. 2010. Characterizing Ancylostoma caninum transcriptome and exploring nematode parasitic adaptation. *Bmc Genomics* **11**.
589. Wang Z, Gao X, Martin J, Yin Y, Abubucker S, Rash AC, Li B-W, Nash B, Hallsworth-Pepin K, Jasmer DP et al. 2013. Gene expression analysis distinguishes tissue-specific and gender-related functions among adult Ascaris suum tissues. *Molecular Genetics and Genomics* **288**(5-6): 243-260.
590. Wang Z, Zarlenga D, Martin J, Abubucker S, Mitreva M. 2012. Exploring metazoan evolution through dynamic and holistic changes in protein families and domains. *Bmc Evolutionary Biology* **12**.
591. Wangler MF, Gonzaga-Jauregui C, Gambin T, Penney S, Moss T, Chopra A, Probst FJ, Xia F, Yang Y, Werlin S et al. 2014. Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. *PLoSGenet* **10**(3): e1004258.
592. Warren WC, Clayton DF, Ellegren H, Arnold AP, Hillier LW, Kuenstner A, Searle S, White S, Vilella AJ, Fairley S et al. 2010. The genome of a songbird. *Nature* **464**(7289): 757-762.
593. Wartman LD, Larson DE, Xiang Z, Ding L, Chen K, Lin L, Cahan P, Klco JM, Welch JS, Li C et al. 2011. Sequencing a mouse acute promyelocytic leukemia genome reveals genetic events relevant for disease progression. *Journal of Clinical Investigation* **121**(4): 1445-1455.
594. Watson CT, Steinberg KM, Huddleston J, Warren RL, Malig M, Schein J, Willsey AJ, Joy JB, Scott JK, Graves TA et al. 2013. Complete Haplotype Sequence of the Human Immunoglobulin Heavy-Chain Variable, Diversity, and Joining Genes and Characterization of Allelic and Copy-Number Variation. *American Journal of Human Genetics* **92**(4): 530-546.
595. Waubant E, Pelletier D, Mass M, Cohen JA, Kita M, Cross A, Bar-Or A, Vollmer T, Racke M, Stueve O et al. 2012. Randomized controlled trial of atorvastatin in clinically isolated syndrome The STAyCIS study. *Neurology* **78**(15): 1171-1178.
596. Weakley SM, Jiang J, Kougias P, Lin PH, Yao Q, Brunicardi FC, Gibbs RA, Chen C. 2010. Role of somatic mutations in vascular disease formation *ExpertRevMolDiagn* **10**(2): 173-185.
597. Webb BD, Shaaban S, Gaspar H, Cunha LF, Schubert CR, Hao K, Robson CD, Chan WM, Andrews C, MacKinnon S et al. 2012. HOXB1 founder mutation in humans recapitulates the phenotype of Hoxb1-/- mice. *Am J Hum Genet* **91**(1): 171-179.
598. Weeke P, Mosley JD, Hanna D, Delaney JT, Shaffer C, Wells QS, Van Driest S, Karnes JH, Ingram C, Guo Y et al. 2014. Exome Sequencing Implicates an Increased Burden of Rare Potassium Channel Variants in the Risk of Drug-Induced Long QT Interval Syndrome. *Journal of the American College of Cardiology* **63**(14): 1430-1437.
599. Welch JS, Ding L, Chen K, Larson DE, Kulkarni S, Payton JE, Wallis J, Veizer J, McLellan MD, Vickery TL et al. 2010. Resolution of a Clinical Dilemma with Whole Genome Sequencing, and Discovery of a New Mechanism for Generating PML-Rara: Insertional Fusion. *Blood* **116**(21): 1136-1136.
600. Welch JS, Larson D, Ding L, McLellan MD, Lamprecht T, Kandoth C, Payton JE, Baty J, Harris CC, Lichti CF et al. 2011. Complete Sequencing and Comparison of 12 Normal Karyotype M1 AML Genomes with 12 t(15;17) Positive M3-APL Genomes. *Blood* **118**(21): 185-185.
601. Welch JS, Ley TJ, Link DC, Miller CA, Larson DE, Koboldt DC, Wartman LD, Lamprecht TL, Liu F, Xia J et al. 2012. The Origin and Evolution of Mutations in Acute Myeloid Leukemia. *Cell* **150**(2): 264-278.
602. Welch JS, Westervelt P, Ding L, Larson DE, Klco JM, Kulkarni S, Wallis J, Chen K, Payton JE, Fulton RS et al. 2011. Use of Whole-Genome Sequencing to Diagnose a Cryptic Fusion Oncogene. *Jama-Journal of the American Medical Association* **305**(15): 1577-1584.
603. Wendl MC, Kota K, Weinstock GM, Mitreva M. 2013. Coverage theories for metagenomic DNA sequencing based on a generalization of Stevens' theorem. *Journal of Mathematical Biology* **67**(5): 1141-1161.
604. Wendl MC, Wallis JW, Lin L, Kandoth C, Mardis ER, Wilson RK, Ding L. 2011. PathScan: a tool for discerning mutational significance in groups of putative cancer genes. *Bioinformatics* **27**(12): 1595-1602.
605. Werren JH Richards S Desjardins CA Niehuis O Gadau J Colbourne JK Werren JH Richards S Desjardins CA Niehuis O et al. 2010. Functional and evolutionary insights from the genomes of three parasitoid Nasonia species. *Science* **327**(5963): 343-348.
606. White NM, Feng FY, Maher CA. 2013. Recurrent Rearrangements in Prostate Cancer: Causes and Therapeutic Potential. *Current Drug Targets* **14**(4): 450-459.
607. Whittington CM, Papenfuss AT, Locke DP, Mardis ER, Wilson RK, Abubucker S, Mitreva M, Wong ESW, Hsu AL, Kuchel PW et al. 2010. Novel venom gene discovery in the platypus. *Genome Biology* **11**(9).
608. Wilbe M, Jokinen P, Truve K, Seppala EH, Karlsson EK, Biagi T, Hughes A, Bannasch D, Andersson G, Hansson-Hamlin H et al. 2010. Genome-wide association mapping identifies multiple loci for a canine SLE-related disease complex. *Nat Genet* **42**(3): 250-254.
609. Williams LJ, Tabbaa DG, Li N, Berlin AM, Shea TP, Maccallum I, Lawrence MS, Drier Y, Getz G, Young SK et al. 2012. Paired-end sequencing of Fosmid libraries by Illumina. *Genome Res* **22**(11): 2241-2249.
610. Wiszniewski W, Hunter JV, Hanchard NA, Willer JR, Shaw C, Tian Q, Illner A, Wang X, Cheung SW, Patel A et al. 2013. TM4SF20 ancestral deletion and susceptibility to a pediatric disorder of early language delay and cerebral white matter hyperintensities. *AmJHumGenet* **93**(2): 197-210.
611. Wong E, Whittington C, Papenfuss T, Nicol S, Warren WC, Belov K. 2012. The Evolutionary Origins of Monotreme Crural Glands. *Toxicon* **60**(2): 122-123.
612. Wong ES, Papenfuss AT, Heger A, Hsu AL, Ponting CP, Miller RD, Fenelon JC, Renfree MB, Gibbs RA, Belov K. 2011. Transcriptomic analysis supports similar functional roles for the two thymuses of the tammar wallaby *BMCGenomics* **12**: 420.
613. Wong ESW, Morgenstern D, Mofiz E, Gombert S, Morris KM, Temple-Smith P, Renfree MB, Whittington CM, King GF, Warren WC et al. 2012. Proteomics and Deep Sequencing Comparison of Seasonally Active Venom Glands in the Platypus Reveals Novel Venom Peptides and Distinct Expression Profiles. *Molecular & Cellular Proteomics* **11**(11): 1354-1364.
614. Wong ESW, Nicol S, Warren WC, Belov K. 2013. Echidna Venom Gland Transcriptome Provides Insights into the Evolution of Monotreme Venom. *Plos One* **8**(11).
615. Wong ESW, Papenfuss AT, Whittington CM, Warren WC, Belov K. 2012. A Limited Role for Gene Duplications in the Evolution of Platypus Venom. *Molecular Biology and Evolution* **29**(1): 167-177.
616. Worley KC, Gibbs RA. 2010. Genetics: Decoding a national treasure. *Nature* **463**(7279): 303-304.
617. Worley KC Warren WC Rogers J Locke D Muzny DM Mardis ER Weinstock GM Tardif SD Aagaard KM Archidiacono N et al. 2014. The genome of the common marmoset: A comparative analysis of an extraordinary South American primate. *Nature Genetics*.
618. Wu G, Broniscer A, McEachron TA, Lu C, Paugh BS, Becksfort J, Qu C, Ding L, Huether R, Parker M et al. 2012. Somatic histone H3 alterations in pediatric diffuse intrinsic pontine gliomas and non-brainstem glioblastomas. *Nature Genetics* **44**(3): 251-253.
619. Wu G, Diaz AK, Paugh BS, Rankin SL, Ju B, Li Y, Zhu X, Qui C, Chen X, Zhang J et al. 2014. The genomic landscape of diffuse intrinsic pontine glioma and pediatric non-brainstem high-grade glioma. *Nature Genetics* **46**(5): 444-450.
620. Xia F, Bainbridge MN, Tan TY, Wangler MF, Scheuerle AE, Zackai EH, Harr MH, Sutton VR, Nalam RL, Zhu W et al. 2014. De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. *AmJHumGenet* **94**(5): 784-789.
621. Xie YA, Lee W, Cai C, Gambin T, Noupuu K, Sujirakul T, Ayuso C, Jhangiani S, Muzny D, Boerwinkle E et al. 2014. New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. *Hum Mol Genet*.
622. Xu J, Yanagisawa Y, Tsankov AM, Hart C, Aoki K, Kommajosyula N, Steinmann KE, Bochicchio J, Russ C, Regev A et al. 2012. Genome-wide identification and characterization of replication origins by deep sequencing. *Genome Biol* **13**(4): R27.
623. Yang Y, Muzny DM, Reid JG, Bainbridge MN, Willis A, Ward PA, Braxton A, Beuten J, Xia F, Niu Z et al. 2013. Clinical whole-exome sequencing for the diagnosis of mendelian disorders *NEnglJMed* **369**(16): 1502-1511.
624. Yao G, Ye L, Gao H, Minx P, Warren WC, Weinstock GM. 2012. Graph accordance of next-generation sequence assemblies. *Bioinformatics* **28**(1): 13-16.
625. Yassour M, Pfiffner J, Levin JZ, Adiconis X, Gnirke A, Nusbaum C, Thompson DA, Friedman N, Regev A. 2010. Strand-specific RNA sequencing reveals extensive regulated long antisense transcripts that are conserved across yeast species. *Genome Biol* **11**(8): R87.
626. Ye L, Hillier LW, Minx P, Thane N, Locke DP, Martin JC, Chen L, Mitreva M, Miller JR, Haub KV et al. 2011. A vertebrate case study of the quality of assemblies derived from next-generation sequences. *Genome Biology* **12**(3).
627. Yeoman CJ, Yildirim S, Thomas SM, Durkin AS, Torralba M, Sutton G, Buhay CJ, Ding Y, Dugan-Rocha SP, Muzny DM et al. 2010. Comparative genomics of Gardnerella vaginalis strains reveals substantial differences in metabolic and virulence potential *PLoSOne* **5**(8): e12411.
628. Yilmaz P, Kottmann R, Field D, Knight R, Cole JR, Amaral-Zettler L, Gilbert JA, Karsch-Mizrachi I, Johnston A, Cochrane G et al. 2011. Minimum information about a marker gene sequence (MIMARKS) and minimum information about any (x) sequence (MIxS) specifications *NatBiotechnol* **29**(5): 415-420.
629. Young MA, Larson DE, Sun C-W, George DR, Ding L, Miller CA, Lin L, Pawlik KM, Chen K, Fan X et al. 2012. Background Mutations in Parental Cells Account for Most of the Genetic Heterogeneity of Induced Pluripotent Stem Cells. *Cell Stem Cell* **10**(5): 570-582.
630. Younis AE, Geisinger F, Ajonina-Ekoti I, Soblik H, Steen H, Mitreva M, Erttmann KD, Perbandt M, Liebau E, Brattig NW. 2011. Stage-specific excretory-secretory small heat shock proteins from the parasitic nematode Strongyloides ratti - putative links to host's intestinal mucosal defense system. *Febs Journal* **278**(18): 3319-3336.
631. Zack TI, Schumacher SE, Carter SL, Cherniack AD, Saksena G, Tabak B, Lawrence MS, Zhang CZ, Wala J, Mermel CH et al. 2013. Pan-cancer patterns of somatic copy number alteration. *Nat Genet* **45**(10): 1134-1140.
632. Zamani N, Russell P, Lantz H, Hoeppner MP, Meadows JR, Vijay N, Mauceli E, di Palma F, Lindblad-Toh K, Jern P et al. 2013. Unsupervised genome-wide recognition of local relationship patterns. *BMC Genomics* **14**: 347.
633. Zhan X, Larson DE, Wang C, Koboldt DC, Sergeev YV, Fulton RS, Fulton LL, Fronick CC, Branham KE, Bragg-Gresham J et al. 2013. Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. *Nature Genetics* **45**(11): 1375-+.
634. Zhang J, Benavente CA, McEvoy J, Flores-Otero J, Ding L, Chen X, Ulyanov A, Wu G, Wilson M, Wang J et al. 2012. A novel retinoblastoma therapy from genomic and epigenetic analyses. *Nature* **481**(7381): 329-334.
635. Zhang J, Ding L, Holmfeldt L, Wu G, Heatley SL, Payne-Turner D, Easton J, Chen X, Wang J, Rusch M et al. 2011. Discovery of Novel Recurrent Mutations in Childhood Early T-Cell Precursor Acute Lymphoblastic Leukemia by Whole Genome Sequencing - a Report From the St Jude Children's Research Hospital - Washington University Pediatric Cancer Genome Project. *Blood* **118**(21): 32-33.
636. Zhang J, Ding L, Holmfeldt L, Wu G, Heatley SL, Payne-Turner D, Easton J, Chen X, Wang J, Rusch M et al. 2012. The genetic basis of early T-cell precursor acute lymphoblastic leukaemia. *Nature* **481**(7380): 157-163.
637. Zhang J, Wu G, Miller CP, Tatevossian RG, Dalton JD, Tang B, Orisme W, Punchihewa C, Parker M, Qaddoumi I et al. 2013. Whole-genome sequencing identifies genetic alterations in pediatric low-grade gliomas. *Nature Genetics* **45**(6): 602-+.
638. Zhang Q, Ding L, Larson DE, Koboldt DC, McLellan MD, Chen K, Shi X, Kraja A, Mardis ER, Wilson RK et al. 2010. CMDS: a population-based method for identifying recurrent DNA copy number aberrations in cancer from high-resolution data. *Bioinformatics* **26**(4): 464-469.
639. Zhang Z, Wang Y, Wang S, Liu J, Warren W, Mitreva M, Walter RB. 2011. Transcriptome Analysis of Female and Male Xiphophorus maculatus Jp 163 A. *Plos One* **6**(4).
640. Zhao W, Petit E, Gafni RI, Collins MT, Robey PG, Seton M, Miller KK, Mannstadt M. 2013. Mutations in NOTCH2 in patients with Hajdu-Cheney syndrome. *Osteoporosis international : a journal established as result of cooperation between the European Foundation for Osteoporosis and the National Osteoporosis Foundation of the USA* **24**(8): 2275-2281.
641. Zhou G, Gingras MC, Liu SH, Li D, Li Z, Catania RL, Stehling KM, Li M, Paganelli G, Gibbs RA et al. 2011. The hypofunctional effect of P335L single nucleotide polymorphism on SSTR5 function. *World JSurg* **35**(8): 1715-1724.
642. Zhou G, Gingras MC, Liu SH, Sanchez R, Edwards D, Dawson D, Christensen K, Paganelli G, Gibbs R, Fisher W et al. 2011. SSTR5 P335L monoclonal antibody differentiates pancreatic neuroendocrine neuroplasms with different SSTR5 genotypes. *Surgery* **150**(6): 1136-1142.
643. Zhou Z, Yu F, Buchanan A, Fu Y, Campos M, Wu KK, Chambless LE, Folsom AR, Boerwinkle E, Dong JF. 2014. Possible race and gender divergence in association of genetic variations with plasma von Willebrand factor: a study of ARIC and 1000 genome cohorts. *PLoSOne* **9**(1): e84810.
644. Zighelboim I, Mutch DG, Knapp A, Ding L, Xie M, Cohn DE, Goodfellow PJ. 2014. High Frequency Strand Slippage Mutations in CTCF in MSI-Positive Endometrial Cancers. *Human Mutation* **35**(1): 63-65.
645. Zuk O, Hechter E, Sunyaev SR, Lander ES. 2012. The mystery of missing heritability: Genetic interactions create phantom heritability. *Proc Natl Acad Sci U S A* **109**(4): 1193-1198.
646. Zuk O, Schaffner SF, Samocha K, Do R, Hechter E, Kathiresan S, Daly MJ, Neale BM, Sunyaev SR, Lander ES. 2014. Searching for missing heritability: designing rare variant association studies. *Proc Natl Acad Sci U S A* **111**(4): E455-464.