Publications

Selected Recent Papers

1. McGinty RJ, Balick DJ, Mirkin SM, Sunyaev SR. Inherent instability of simple DNA repeats shapes an evolutionarily stable distribution of repeat lengths. Nat Commun. 2025 Dec 9. doi: 10.1038/s41467-025-66725-5.
[PMID: 41365873] [DOI: 10.1038/s41467-025-66725-5]
2. Seplyarskiy V, Moldovan MA, Koch E, Kar P, Neville MDC, Rahbari R, Sunyaev S. Hotspots of human mutation point to clonal expansions in spermatogonia. Nature. 2025 Oct 8. doi: 10.1038/s41586-025-09579-7.
[PMID: 41062699] [DOI: 10.1038/s41586-025-09579-7]
3. Kobren SN, Moldovan MA, Reimers R, Traviglia D, Li X, Barnum D, Veit A, Corona RI, Carvalho Neto GdV, Willett J, Berselli M, Ronchetti W, Nelson SF, Martinez-Agosto JA, Sherwood R, Krier J, Kohane IS, Sunyaev SR, Undiagnosed Diseases Network. Joint, multifaceted genomic analysis enables diagnosis of diverse, ultra-rare monogenic presentations. Nat Commun. 2025 Aug 7;16(1):7267. doi: 10.1038/s41467-025-61712-2.
[PMID: 40770127] [DOI: 10.1038/s41467-025-61712-2]
4. Maury EA, Jones A, Seplyarskiy V, Nguyen TTL, Rosenbluh C, Bae T, Wang Y, Abyzov A, Khoshkhoo S, Chahine Y, Zhao S, Venkatesh S, Root E, Voloudakis G, Roussos P, Park PJ, Akbarian S, Brennand K, Reilly S, Lee EA, Sunyaev SR, Walsh CA, Chess A, Brain Somatic Mosaicism Network‡. Somatic mosaicism in schizophrenia brains reveals prenatal mutational processes. Science. 2024 Oct 11;386(6718):217-224. doi: 10.1126/science.adq1456. Epub 2024 Oct 10.
[PMID: 39388546] [DOI: 10.1126/science.adq1456]
5. Seplyarskiy V, Koch EM, Lee DJ, Lichtman JS, Luan HH, Sunyaev SR. A mutation rate model at the basepair resolution identifies the mutagenic effect of polymerase III transcription. Nat Genet. 2023 Dec;55(12):2235-2242. doi: 10.1038/s41588-023-01562-0. Epub 2023 Nov 30.
[PMID: 38036792] [DOI: 10.1038/s41588-023-01562-0]
6. McGinty RJ, Sunyaev SR. Revisiting mutagenesis at non-B DNA motifs in the human genome. Nat Struct Mol Biol. 2023 Apr;30(4):417-424. doi: 10.1038/s41594-023-00936-6. Epub 2023 Mar 13.
[PMID: 36914796] [PMCID: 10225297] [DOI: 10.1038/s41594-023-00936-6]
7. Connally NJ, Nazeen S, Lee D, Shi H, Stamatoyannopoulos J, Chun S, Cotsapas C, Cassa CA, Sunyaev SR. The missing link between genetic association and regulatory function. Elife. 2022 Dec 14;11:e74970. doi: 10.7554/eLife.74970.

[PMID: 36515579] [PMCID: 9842386] [DOI: 10.7554/eLife.74970]

8. Seplyarskiy VB, Sunyaev S. The origin of human mutation in light of genomic data. Nat Rev Genet. 2021 Oct;22(10):672-686. doi: 10.1038/s41576-021-00376-2. Epub 2021 Jun 23. [PMID: 34163020] [DOI: 10.1038/s41576-021-00376-2]
9. Koch EM, Sunyaev SR. Maintenance of Complex Trait Variation: Classic Theory and Modern Data. Front Genet. 2021 Nov 12;12:763363. doi: 10.3389/fgene.2021.763363. eCollection 2021. [PMID: 34868244] [PMCID: 8636146] [DOI: 10.3389/fgene.2021.763363]
10. Seplyarskiy VB, Soldatov RA, Koch E, McGinty RJ, Goldmann JM, Hernandez RD, Barnes K, Correa A, Burchard EG, Ellinor PT, McGarvey ST, Mitchell BD, Vasan RS, Redline S, Silverman E, Weiss ST, Arnett DK, Blangero J, Boerwinkle E, He J, Montgomery C, Rao DC, Rotter JI, Taylor KD, Brody JA, Chen YI, de Las Fuentes L, Hwu C, Rich SS, Manichaikul AW, Mychaleckyj JC, Palmer ND, Smith JA, Kardia SLR, Peyser PA, Bielak LF, O'Connor TD, Emery LS, Gilissen C, Wong WSW, Kharchenko PV, Sunyaev S, TOPMed Population Genetics Working Group. Population sequencing data reveal a compendium of mutational processes in the human germ line. Science. 2021 Aug 27;373(6558):1030-1035. doi: 10.1126/science.aba7408. Epub 2021 Aug 12. [PMID: 34385354] [PMCID: 9217108] [DOI: 10.1126/science.aba7408]
11. Radke DW, Sul JH, Balick DJ, Akle S, Green RC, Sunyaev SR, Alzheimer's Disease Neuroimaging Initiative. Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. Genome Res. 2021 Jun;31(6):935-946. doi: 10.1101/gr.275263.121. Epub 2021 May 7. [PMID: 33963077] [PMCID: 8168579] [DOI: 10.1101/gr.275263.121]
12. Dietlein F, Weghorn D, Taylor-Weiner A, Richters A, Reardon B, Liu D, Lander ES, Van Allen EM, Sunyaev SR. Identification of cancer driver genes based on nucleotide context. Nat Genet. 2020 Feb;52(2):208-218. doi: 10.1038/s41588-019-0572-y. Epub 2020 Feb 3. [PMID: 32015527] [PMCID: 7031046] [DOI: 10.1038/s41588-019-0572-y]
13. Chun S, Imakaev M, Hui D, Patsopoulos NA, Neale BM, Kathiresan S, Stitziel NO, Sunyaev SR. Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. Am J Hum Genet. 2020 Jul 2;107(1):46-59. doi: 10.1016/j.ajhg.2020.05.004. Epub 2020 May 28. [PMID: 32470373] [PMCID: 7332650] [DOI: 10.1016/j.ajhg.2020.05.004]
14. Weghorn D, Balick DJ, Cassa C, Kosmicki JA, Daly MJ, Beier DR, Sunyaev SR. Applicability of the Mutation-Selection Balance Model to Population Genetics of Heterozygous Protein-Truncating Variants in Humans. Mol Biol Evol. 2019 Aug 1;36(8):1701-1710. doi: 10.1093/molbev/msz092. [PMID: 31004148] [PMCID: 6738481] [DOI: 10.1093/molbev/msz092]
15. Sohail M, Maier RM, Ganna A, Bloemendal A, Martin AR, Turchin MC, Chiang CW, Hirschhorn J, Daly MJ, Patterson N, Neale B, Mathieson I, Reich D, Sunyaev SR. Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. Elife. 2019 Mar 21;8:e39702. doi: 10.7554/eLife.39702. [PMID: 30895926] [PMCID: 6428571] [DOI: 10.7554/eLife.39702]
16. Seplyarskiy VB, Akkuratov EE, Akkuratova N, Andrianova MA, Nikolaev SI, Bazykin GA, Adameyko I, Sunyaev SR. Error-prone bypass of DNA lesions during lagging-strand replication is a common source of germline and cancer mutations. Nat Genet. 2019 Jan;51(1):36-41. doi: 10.1038/s41588-018-0285-7. Epub 2018 Dec 3. [PMID: 30510240] [PMCID: 6317876] [DOI: 10.1038/s41588-018-0285-7]
17. Weghorn D, Sunyaev S. Bayesian inference of negative and positive selection in human cancers. Nat Genet. 2017 Dec;49(12):1785-1788. doi: 10.1038/ng.3987. Epub 2017 Nov 6.

[PMID: 29106416] [DOI: 10.1038/ng.3987]

18. Sohail M, Vakhrusheva OA, Sul JH, Pulit SL, Francioli LC, van den Berg LH, Veldink JH, de Bakker PIW, Bazykin GA, Kondrashov AS, Sunyaev SR, Alzheimer's Disease Neuroimaging Initiative. Negative selection in humans and fruit flies involves synergistic epistasis. Science. 2017 May 5;356(6337):539-542. doi: 10.1126/science.aah5238.
[PMID: 28473589] [PMCID: 6200135] [DOI: 10.1126/science.aah5238]
19. Cassa CA, Weghorn D, Balick DJ, Jordan DM, Nusinow D, Samocha KE, O'Donnell-Luria A, MacArthur DG, Daly MJ, Beier DR, Sunyaev SR. Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nat Genet. 2017 May;49(5):806-810. doi: 10.1038/ng.3831. Epub 2017 Apr 3.
[PMID: 28369035] [PMCID: 5618255] [DOI: 10.1038/ng.3831]
20. Chun S, Casparino A, Patsopoulos NA, Croteau-Chonka DC, Raby BA, De Jager PL, Sunyaev SR, Cotsapas C. Limited statistical evidence for shared genetic effects of eQTLs and autoimmune-disease-associated loci in three major immune-cell types. Nat Genet. 2017 Apr;49(4):600-605. doi: 10.1038/ng.3795. Epub 2017 Feb 20.
[PMID: 28218759] [PMCID: 5374036] [DOI: 10.1038/ng.3795]
21. Polak P, Karlić R, Koren A, Thurman R, Sandstrom R, Lawrence M, Reynolds A, Rynes E, Vlahoviček K, Stamatoyannopoulos JA, Sunyaev SR. Cell-of-origin chromatin organization shapes the mutational landscape of cancer. Nature. 2015 Feb 19;518(7539):360-364. doi: 10.1038/nature14221.
[PMID: 25693567] [PMCID: 4405175] [DOI: 10.1038/nature14221]
22. Francioli LC, Polak PP, Koren A, Menelaou A, Chun S, Renkens I, van Duijn CM, Swertz M, Wijmenga C, van Ommen G, Slagboom PE, Boomsma DI, Ye K, Guryev V, Arndt PF, Kloosterman WP, de Bakker PIW, Sunyaev SR, Genome of the Netherlands Consortium. Genome-wide patterns and properties of de novo mutations in humans. Nat Genet. 2015 Jul;47(7):822-826. doi: 10.1038/ng.3292. Epub 2015 May 18.
[PMID: 25985141] [PMCID: 4485564] [DOI: 10.1038/ng.3292]
23. Balick DJ, Do R, Cassa CA, Reich D, Sunyaev SR. Dominance of Deleterious Alleles Controls the Response to a Population Bottleneck. PLoS Genet. 2015 Aug 28;11(8):e1005436. doi: 10.1371/journal.pgen.1005436. eCollection 2015 Aug.
[PMID: 26317225] [PMCID: 4552954] [DOI: 10.1371/journal.pgen.1005436]
24. Jordan DM, Frangakis SG, Golzio C, Cassa CA, Kurtzberg J, Davis EE, Sunyaev SR, Katsanis N, Task Force for Neonatal Genomics. Identification of cis-suppression of human disease mutations by comparative genomics. Nature. 2015 Aug 13;524(7564):225-9. doi: 10.1038/nature14497. Epub 2015 Jun 29.
[PMID: 26123021] [PMCID: 4537371] [DOI: 10.1038/nature14497]

Other Papers

1. Zhou H, Verma V, Li X, Li Z, Shedd N, Li TC, Yang H, Zhang A, Borsari B, Buyske S, Gerstein M, Matise T, Zody MC, Neale B, Weng Z, Sunyaev SR, Lin X. FAVOR 2.0: A reengineered functional annotation of variants online resource for interpreting genomic variation. Nucleic Acids Res. 2025 Dec 3:gkaf1217. doi: 10.1093/nar/gkaf1217.
[PMID: 41335103] [DOI: 10.1093/nar/gkaf1217]

2. Fazal S, Dashnow H, Dohrn MF, Raposo J, Hiatt L, Danzi MC, Xu IRL, Toro C, Adams DR, Usdin K, Hayward B, Kobren SN, Sunyaev SR, Spillmann RC, Shashi V, Rebelo A, Bademci G, Tekin M, Quinlan AR, Züchner S, Undiagnosed Diseases Network. A genome-wide approach for the discovery of novel repeat expansion disorders in the Undiagnosed Diseases Network cohort. Genet Med. 2025 May 22:101462. doi: 10.1016/j.gim.2025.101462.
[PMID: 40417743] [DOI: 10.1016/j.gim.2025.101462]
3. Nazeen S, Wang X, Morrow A, Strom R, Ethier E, Ritter D, Henderson A, Afroz J, Stitziel NO, Gupta RM, Luk K, Studer L, Khurana V, Sunyaev SR. NERINE reveals rare variant associations in gene networks across multiple phenotypes and implicates an SNCA-PRL-LRRK2 subnetwork in Parkinson's disease. bioRxiv [Preprint]. 2025 Jan 10:2025.01.07.631688. doi: 10.1101/2025.01.07.631688.
[PMID: 39829934] [PMCID: 11741352] [DOI: 10.1101/2025.01.07.631688]
4. Hakim A, Connally NJ, Schnitzler GR, Cho MH, Jiang ZG, Sunyaev SR, Gupta RM. Missing Regulation Between Genetic Association and Transcriptional Abundance for Hypercholesterolemia Genes. Genes (Basel). 2025 Jan 15;16(1):84. doi: 10.3390/genes16010084.
[PMID: 39858631] [PMCID: 11764661] [DOI: 10.3390/genes16010084]
5. Hajheideri M, Sunyaev S, de Meaux J. Are complex traits underpinned by polygenic molecular traits? A reflection on the complexity of gene expression. Plant Cell Physiol. 2024 Nov 29:pcae140. doi: 10.1093/pcp/pcae140.
[PMID: 39626022] [DOI: 10.1093/pcp/pcae140]
6. Lake NJ, Ma K, Liu W, Battle SL, Laricchia KM, Tiao G, Puiu D, Ng KK, Cohen J, Compton AG, Cowie S, Christodoulou J, Thorburn DR, Zhao H, Arking DE, Sunyaev SR, Lek M. Quantifying constraint in the human mitochondrial genome. Nature. 2024 Oct 16. doi: 10.1038/s41586-024-08048-x.
[PMID: 39415008] [DOI: 10.1038/s41586-024-08048-x]
7. Li TC, Zhou H, Verma V, Tang X, Shao Y, Van Buren E, Weng Z, Gerstein M, Neale B, Sunyaev SR, Lin X. FAVOR-GPT: a generative natural language interface to whole genome variant functional annotations. Bioinform Adv. 2024 Sep 28;4(1):vbae143. doi: 10.1093/bioadv/vbae143. eCollection 2024.
[PMID: 39387060] [PMCID: 11461909] [DOI: 10.1093/bioadv/vbae143]
8. Yu T, Fife JD, Bhat V, Adzhubey I, Sherwood R, Cassa CA. FUSE: Improving the estimation and imputation of variant impacts in functional screening. Cell Genom. 2024 Oct 9;4(10):100667. doi: 10.1016/j.xgen.2024.100667.
[PMID: 39389016] [DOI: 10.1016/j.xgen.2024.100667]
9. Lincoln MR, Connally N, Axisa P, Gasperi C, Mitrovic M, van Heel D, Wijmenga C, Withoff S, Jonkers IH, Padyukov L, Rich SS, Graham RR, Gaffney PM, Langefeld CD, Vyse TJ, Hafler DA, Chun S, Sunyaev SR, Cotsapas C, International Multiple Sclerosis Genetics Consortium. Genetic mapping across autoimmune diseases reveals shared associations and mechanisms. Nat Genet. 2024 May;56(5):838-845. doi: 10.1038/s41588-024-01732-8. Epub 2024 May 13.
[PMID: 38741015] [DOI: 10.1038/s41588-024-01732-8]
10. Bendapudi PK, Nazeen S, Ryu J, Söylemez O, Robbins A, Rouaisnel B, O'Neil JK, Pokhriyal R, Yang M, Colling M, Pasko B, Bouzinier M, Tomczak L, Collier L, Barrios D, Ram S, Toth-Petroczy A, Krier J, Fieg E, Dzik WH, Hudspeth JC, Pozdnyakova O, Nardi V, Knight J, Maas R, Sunyaev S, Losman J. Low-frequency inherited complement receptor variants are associated with purpura fulminans. Blood. 2024 Mar 14;143(11):1032-1044. doi: 10.1182/blood.2023021231.
[PMID: 38096369] [PMCID: 10950473] [DOI: 10.1182/blood.2023021231]

11. Nazeen S, Wang X, Zielinski D, Lam I, Hallacli E, Xu P, Ethier E, Strom R, Zanella CA, Nithianandam V, Ritter D, Henderson A, Saurat N, Afroz J, Nutter-Upham A, Benyamini H, Copty J, Ravishankar S, Morrow A, Mitchel J, Neavin D, Gupta R, Farbehi N, Grundman J, Myers RH, Scherzer CR, Trojanowski JQ, Van Deerlin VM, Cooper AA, Lee EB, Erlich Y, Lindquist S, Peng J, Geschwind DH, Powell J, Studer L, Feany MB, Sunyaev SR, Khurana V. Deep sequencing of proteotoxicity modifier genes uncovers a Presenilin-2/beta-amyloid-actin genetic risk module shared among alpha-synucleinopathies. bioRxiv [Preprint]. 2024 Mar 7:2024.03.03.583145. doi: 10.1101/2024.03.03.583145.
[PMID: 38496508] [PMCID: 10942362] [DOI: 10.1101/2024.03.03.583145]
12. Critical Assessment of Genome Interpretation Consortium. CAGI, the Critical Assessment of Genome Interpretation, establishes progress and prospects for computational genetic variant interpretation methods. Genome Biol. 2024 Feb 22;25(1):53. doi: 10.1186/s13059-023-03113-6.
[PMID: 38389099] [PMCID: 10882881] [DOI: 10.1186/s13059-023-03113-6]
13. Fazal S, Danzi MC, Xu I, Kobren SN, Sunyaev S, Reuter C, Marwaha S, Wheeler M, Dolzhenko E, Lucas F, Wuchty S, Tekin M, Züchner S, Aguiar-Pulido V. RExPRT: a machine learning tool to predict pathogenicity of tandem repeat loci. Genome Biol. 2024 Jan 31;25(1):39. doi: 10.1186/s13059-024-03171-4.
[PMID: 38297326] [PMCID: 10832122] [DOI: 10.1186/s13059-024-03171-4]
14. Danilov SM, Adzhubei IA, Kozuch AJ, Petukhov PA, Popova IA, Choudhury A, Sengupta D, Dudek SM. Carriers of Heterozygous Loss-of-Function ACE Mutations Are at Risk for Alzheimer's Disease. Biomedicines. 2024 Jan 12;12(1):162. doi: 10.3390/biomedicines12010162.
[PMID: 38255267] [PMCID: 10813023] [DOI: 10.3390/biomedicines12010162]
15. Gao H, Hamp T, Ede J, Schraiber JG, McRae J, Singer-Berk M, Yang Y, Dietrich ASD, Fiziev PP, Kuderna LFK, Sundaram L, Wu Y, Adhikari A, Field Y, Chen C, Batzoglou S, Aguet F, Lemire G, Reimers R, Balick D, Janiak MC, Kuhlwilm M, Orkin JD, Manu S, Valenzuela A, Bergman J, Rousselle M, Silva FE, Agueda L, Blanc J, Gut M, de Vries D, Goodhead I, Harris RA, Raveendran M, Jensen A, Chuma IS, Horvath JE, Hvilsom C, Juan D, Frandsen P, de Melo FR, Bertuol F, Byrne H, Sampaio I, Farias I, do Amaral JV, Messias M, da Silva MNF, Trivedi M, Rossi R, Hrbek T, Andriaholinirina N, Rabarivola CJ, Zaramody A, Jolly CJ, Phillips-Conroy J, Wilkerson G, Abee C, Simmons JH, Fernandez-Duque E, Kanthaswamy S, Shiferaw F, Wu D, Zhou L, Shao Y, Zhang G, Keyyu JD, Knauf S, Le MD, Lizano E, Merker S, Navarro A, Bataillon T, Nadler T, Khor CC, Lee J, Tan P, Lim WK, Kitchener AC, Zinner D, Gut I, Melin A, Guschanski K, Schierup MH, Beck RMD, Umapathy G, Roos C, Boubli JP, Lek M, Sunyaev S, O'Donnell-Luria A, Rehm HL, Xu J, Rogers J, Marques-Bonet T, Farh KK. The landscape of tolerated genetic variation in humans and primates. Science. 2023 Jun 2;380(6648):eabn8153. doi: 10.1126/science.abn8197. Epub 2023 Jun 2.
[PMID: 37262156] [PMCID: 10713091] [DOI: 10.1126/science.abn8197]
16. Jain N, Richter F, Adzhubei I, Sharp AJ, Gelb BD. Small open reading frames: a comparative genetics approach to validation. BMC Genomics. 2023 May 1;24(1):226. doi: 10.1186/s12864-023-09311-7.
[PMID: 37127568] [PMCID: 10152738] [DOI: 10.1186/s12864-023-09311-7]
17. Luppino F, Adzhubei IA, Cassa CA, Toth-Petroczy A. DeMAG predicts the effects of variants in clinically actionable genes by integrating structural and evolutionary epistatic features. Nat Commun. 2023 Apr 19;14(1):2230. doi: 10.1038/s41467-023-37661-z.
[PMID: 37076482] [PMCID: 10115847] [DOI: 10.1038/s41467-023-37661-z]
18. Wakeley J, Fan WL, Koch E, Sunyaev S. Recurrent mutation in the ancestry of a rare variant. Genetics. 2023 Jul 6;224(3):iyad049. doi: 10.1093/genetics/iyad049.
[PMID: 36967220] [PMCID: 10324944] [DOI: 10.1093/genetics/iyad049]

19. Bhat V, Adzhubei IA, Fife JD, Lebo M, Cassa CA. Informing variant assessment using structured evidence from prior classifications (PS1, PM5, and PVS1 sequence variant interpretation criteria). Genet Med. 2023 Jan;25(1):16-26. doi: 10.1016/j.gim.2022.09.009. Epub 2022 Oct 28. [PMID: 36305854] [PMCID: 10223563] [DOI: 10.1016/j.gim.2022.09.009]
20. Zhou H, Arapoglou T, Li X, Li Z, Zheng X, Moore J, Asok A, Kumar S, Blue EE, Buyske S, Cox N, Felsenfeld A, Gerstein M, Kenny E, Li B, Matise T, Philippakis A, Rehm HL, Sofia HJ, Snyder G, Weng Z, Neale B, Sunyaev SR, Lin X,
NHGRI Genome Sequencing Program Variant Functional Annotation Working Group. FAVOR: functional annotation of variants online resource and annotator for variation across the human genome. Nucleic Acids Res. 2023 Jan 6;51(D1):D1300-D1311. doi: 10.1093/nar/gkac966. [PMID: 36350676] [PMCID: 9825437] [DOI: 10.1093/nar/gkac966]
21. Chun S, Akle S, Teodosiadis A, Cade BE, Wang H, Sofer T, Evans DS, Stone KL, Gharib SA, Mukherjee S, Palmer LJ, Hillman D, Rotter JI, Hanis CL, Stamatoyannopoulos JA, Redline S, Cotsapas C, Sunyaev SR. Leveraging pleiotropy to discover and interpret GWAS results for sleep-associated traits. PLoS Genet. 2022 Dec 27;18(12):e1010557. doi: 10.1371/journal.pgen.1010557. eCollection 2022 Dec.
[PMID: 36574455] [PMCID: 9829185] [DOI: 10.1371/journal.pgen.1010557] [data]
22. Bouzinier MA, Etin D, Trifonov SI, Evdokimova VN, Ulitin V, Shen J, Kokorev A, Ghazani AA, Chekaluk Y, Albertyn Z, Giersch A, Morton CC, Abraamyan F, Bendapudi PK, Sunyaev S, Undiagnosed D, Brigham G, SEQuencing A, Quantori , Krier JB. AnFiSA: An open-source computational platform for the analysis of sequencing data for rare genetic disease. J Biomed Inform. 2022 Sep;133:104174. doi: 10.1016/j.jbi.2022.104174. Epub 2022 Aug 23.
[PMID: 35998814] [DOI: 10.1016/j.jbi.2022.104174]
23. Koch EM, Du J, Dressner M, Alwasti HE, Al Taif Z, Shehab F, Mohamed AM, Ghanem A, Alhajeri A, Alawadhi A, Almoamen N, Ashoor K, Hasan S, Haghighi A, Sunyaev S, Farhat M. Demographic and Viral-Genetic Analyses of COVID-19 Severity in Bahrain Identify Local Risk Factors and a Protective Effect of Polymerase Mutations. medRxiv [Preprint]. 2023 Oct 12:2022.08.13.22278740. doi: 10.1101/2022.08.13.22278740.
[PMID: 36032980] [PMCID: 9413726] [DOI: 10.1101/2022.08.13.22278740]
24. Collins RL, Glessner JT, Porcu E, Lepamets M, Brandon R, Lauricella C, Han L, Morley T, Niestroj L, Ulirsch J, Everett S, Howrigan DP, Boone PM, Fu J, Karczewski KJ, Kellaris G, Lowther C, Lucente D, Mohajeri K, Nõukas M, Nuttle X, Samocha KE, Trinh M, Ullah F, Võsa U, Hurles ME, Aradhya S, Davis EE, Finucane H, Gusella JF, Janze A, Katsanis N, Matyakhina L, Neale BM, Sanders D, Warren S, Hodge JC, Lal D, Ruderfer DM, Meck J, Mägi R, Esko T, Reymond A, Kutalik Z, Hakonarson H, Sunyaev S, Brand H, Talkowski ME, Estonian Biobank Research Team. A cross-disorder dosage sensitivity map of the human genome. Cell. 2022 Aug 4;185(16):3041-3055.e25. doi: 10.1016/j.cell.2022.06.036. Epub 2022 Aug 1.
[PMID: 35917817] [PMCID: 9742861] [DOI: 10.1016/j.cell.2022.06.036]
25. Hallacli E, Kayatekin C, Nazeen S, Wang XH, Sheinkopf Z, Sathyakumar S, Sarkar S, Jiang X, Dong X, Di Maio R, Wang W, Keeney MT, Felsky D, Sandoe J, Vahdatshoar A, Udeshi ND, Mani DR, Carr SA, Lindquist S, De Jager PL, Bartel DP, Myers CL, Greenamyre JT, Feany MB, Sunyaev SR, Chung CY, Khurana V. The Parkinson's disease protein alpha-synuclein is a modulator of processing bodies and mRNA stability. Cell. 2022 Jun 9;185(12):2035-2056.e33. doi: 10.1016/j.cell.2022.05.008.
[PMID: 35688132] [PMCID: 9394447] [DOI: 10.1016/j.cell.2022.05.008]
26. Dietlein F, Wang AB, Fagre C, Tang A, Besselink NJM, Cuppen E, Li C, Sunyaev SR, Neal JT, Van Allen EM. Genome-wide analysis of somatic noncoding mutation patterns in cancer. Science. 2022 Apr

8;376(6589):eabg5601. doi: 10.1126/science.abg5601. Epub 2022 Apr 8.
[PMID: 35389777] [PMCID: 9092060] [DOI: 10.1126/science.abg5601]

27. Sharo AG, Hu Z, Sunyaev SR, Brenner SE. StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants. Am J Hum Genet. 2022 Feb 3;109(2):195-209. doi: 10.1016/j.ajhg.2021.12.007. Epub 2022 Jan 14.
[PMID: 35032432] [PMCID: 8874149] [DOI: 10.1016/j.ajhg.2021.12.007]
28. Balick DJ, Jordan DM, Sunyaev S, Do R. Overcoming constraints on the detection of recessive selection in human genes from population frequency data. Am J Hum Genet. 2022 Jan 6;109(1):33-49. doi: 10.1016/j.ajhg.2021.12.001. Epub 2021 Dec 23.
[PMID: 34951958] [PMCID: 8764206] [DOI: 10.1016/j.ajhg.2021.12.001]
29. Seplyarskiy VB, Sunyaev S. Author Correction: The origin of human mutation in light of genomic data. Nat Rev Genet. 2021 Oct;22(10):687. doi: 10.1038/s41576-021-00410-3.
[PMID: 34376821] [DOI: 10.1038/s41576-021-00410-3]
30. Mendelevich A, Vinogradova S, Gupta S, Mironov AA, Sunyaev SR, Gimelbrant AA. Replicate sequencing libraries are important for quantification of allelic imbalance. Nat Commun. 2021 Jun 7;12(1):3370. doi: 10.1038/s41467-021-23544-8.
[PMID: 34099647] [PMCID: 8184992] [DOI: 10.1038/s41467-021-23544-8]
31. Kobren SN, Baldridge D, Velinder M, Krier JB, LeBlanc K, Esteves C, Pusey BN, Züchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner DJ, Paul AJ, Sunyaev SR, Kohane IS, Undiagnosed Diseases Network. Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genet Med. 2021 Jun;23(6):1075-1085. doi: 10.1038/s41436-020-01084-8. Epub 2021 Feb 12.
[PMID: 33580225] [PMCID: 8187147] [DOI: 10.1038/s41436-020-01084-8]
32. Takou M, Hämälä T, Koch EM, Steige KA, Dittberner H, Yant L, Genete M, Sunyaev S, Castric V, Vekemans X, Savolainen O, Meaux Jd. Maintenance of Adaptive Dynamics and No Detectable Load in a Range-Edge Outcrossing Plant Population. Mol Biol Evol. 2021 May 4;38(5):1820-1836. doi: 10.1093/molbev/msaa322.
[PMID: 33480994] [PMCID: 8097302] [DOI: 10.1093/molbev/msaa322]
33. Gasperi C, Chun S, Sunyaev SR, Cotsapas C. Shared associations identify causal relationships between gene expression and immune cell phenotypes. Commun Biol. 2021 Mar 4;4(1):279. doi: 10.1038/s42003-021-01823-w.
[PMID: 33664438] [PMCID: 7933159] [DOI: 10.1038/s42003-021-01823-w]
34. Shi H, Gazal S, Kanai M, Koch EM, Schoech AP, Siewert KM, Kim SS, Luo Y, Amariuta T, Huang H, Okada Y, Raychaudhuri S, Sunyaev SR, Price AL. Population-specific causal disease effect sizes in functionally important regions impacted by selection. Nat Commun. 2021 Feb 17;12(1):1098. doi: 10.1038/s41467-021-21286-1.
[PMID: 33597505] [PMCID: 7889654] [DOI: 10.1038/s41467-021-21286-1]
35. Wieters B, Steige KA, He F, Koch EM, Ramos-Onsins SE, Gu H, Guo Y, Sunyaev S, de Meaux J. Polygenic adaptation of rosette growth in Arabidopsis thaliana. PLoS Genet. 2021 Jan 25;17(1):e1008748. doi: 10.1371/journal.pgen.1008748. eCollection 2021 Jan.
[PMID: 33493157] [PMCID: 7861555] [DOI: 10.1371/journal.pgen.1008748]
36. Kousi M, Söylemez O, Ozanturk A, Mourtzi N, Akle S, Jungreis I, Muller J, Cassa CA, Brand H, Mokry JA, Wolf MY, Sadeghpour A, McFadden K, Lewis RA, Talkowski ME, Dollfus H, Kellis M, Davis EE, Sunyaev SR, Katsanis N. Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nat Genet. 2020

Nov;52(11):1145-1150. doi: 10.1038/s41588-020-0707-1. Epub 2020 Oct 12.
[PMID: 33046855] [PMCID: 8272915] [DOI: 10.1038/s41588-020-0707-1]

37. Li X, Li Z, Zhou H, Gaynor SM, Liu Y, Chen H, Sun R, Dey R, Arnett DK, Aslibekyan S, Ballantyne CM, Bielak LF, Blangero J, Boerwinkle E, Bowden DW, Broome JG, Conomos MP, Correa A, Cupples LA, Curran JE, Freedman BI, Guo X, Hindy G, Irvin MR, Kardia SLR, Kathiresan S, Khan AT, Kooperberg CL, Laurie CC, Liu XS, Mahaney MC, Manichaikul AW, Martin LW, Mathias RA, McGarvey ST, Mitchell BD, Montasser ME, Moore JE, Morrison AC, O'Connell JR, Palmer ND, Pampana A, Peralta JM, Peyser PA, Psaty BM, Redline S, Rice KM, Rich SS, Smith JA, Tiwari HK, Tsai MY, Vasan RS, Wang FF, Weeks DE, Weng Z, Wilson JG, Yanek LR, Neale BM, Sunyaev SR, Abecasis GR, Rotter JI, Willer CJ, Peloso GM, Natarajan P, Lin X, TOPMed Lipids Working Group. Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. Nat Genet. 2020 Sep;52(9):969-983. doi: 10.1038/s41588-020-0676-4. Epub 2020 Aug 24.
[PMID: 32839606] [PMCID: 7483769] [DOI: 10.1038/s41588-020-0676-4]
38. Arnold B, Sohail M, Wadsworth C, Corander J, Hanage WP, Sunyaev S, Grad YH. Fine-Scale Haplotype Structure Reveals Strong Signatures of Positive Selection in a Recombining Bacterial Pathogen. Mol Biol Evol. 2020 Feb 1;37(2):417-428. doi: 10.1093/molbev/msz225.
[PMID: 31589312] [PMCID: 6993868] [DOI: 10.1093/molbev/msz225]
39. Nissim S, Leshchiner I, Mancias JD, Greenblatt MB, Maertens O, Cassa CA, Rosenfeld JA, Cox AG, Hedgepeth J, Wucherpfennig JI, Kim AJ, Henderson JE, Gonyo P, Brandt A, Lorimer E, Unger B, Prokop JW, Heidel JR, Wang X, Ukaegbu CI, Jennings BC, Paulo JA, Gableske S, Fierke CA, Getz G, Sunyaev SR, Wade Harper J, Cichowski K, Kimmelman AC, Houvras Y, Syngal S, Williams C, Goessling W. Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nat Genet. 2019 Sep;51(9):1308-1314. doi: 10.1038/s41588-019-0475-y. Epub 2019 Aug 12.
[PMID: 31406347] [PMCID: 7159804] [DOI: 10.1038/s41588-019-0475-y]
40. Farhat MR, Freschi L, Calderon R, Ioerger T, Snyder M, Meehan CJ, de Jong B, Rigouts L, Sloutsky A, Kaur D, Sunyaev S, van Soolingen D, Shendure J, Sacchettini J, Murray M. GWAS for quantitative resistance phenotypes in Mycobacterium tuberculosis reveals resistance genes and regulatory regions. Nat Commun. 2019 May 13;10(1):2128. doi: 10.1038/s41467-019-10110-6.
[PMID: 31086182] [PMCID: 6513847] [DOI: 10.1038/s41467-019-10110-6]
41. Cade BE, Chen H, Stilp AM, Louie T, Ancoli-Israel S, Arens R, Barfield R, Below JE, Cai J, Conomos MP, Evans DS, Frazier-Wood AC, Gharib SA, Gleason KJ, Gottlieb DJ, Hillman DR, Johnson WC, Lederer DJ, Lee J, Loredo JS, Mei H, Mukherjee S, Patel SR, Post WS, Purcell SM, Ramos AR, Reid KJ, Rice K, Shah NA, Sofer T, Taylor KD, Thornton TA, Wang H, Yaffe K, Zee PC, Hanis CL, Palmer LJ, Rotter JI, Stone KL, Tranah GJ, Wilson JG, Sunyaev SR, Laurie CC, Zhu X, Saxena R, Lin X, Redline S. Associations of variants In the hexokinase 1 and interleukin 18 receptor regions with oxyhemoglobin saturation during sleep. PLoS Genet. 2019 Apr 16;15(4):e1007739. doi: 10.1371/journal.pgen.1007739. eCollection 2019 Apr.
[PMID: 30990817] [PMCID: 6467367] [DOI: 10.1371/journal.pgen.1007739]
42. Mohanty AK, Vuzman D, Francioli L, Cassa C, Toth-Petroczy A, Sunyaev S, Brigham and Women’s Hospital FaceBase Project. novoCaller: a Bayesian network approach for de novo variant calling from pedigree and population sequence data. Bioinformatics. 2019 Apr 1;35(7):1174-1180. doi: 10.1093/bioinformatics/bty749.
[PMID: 30169785] [PMCID: 6449753] [DOI: 10.1093/bioinformatics/bty749]
43. Schoech AP, Jordan DM, Loh P, Gazal S, O'Connor LJ, Balick DJ, Palamara PF, Finucane HK, Sunyaev

SR, Price AL. Quantification of frequency-dependent genetic architectures in 25 UK Biobank traits reveals action of negative selection. Nat Commun. 2019 Feb 15;10(1):790. doi: 10.1038/s41467-019-08424-6.
[PMID: 30770844] [PMCID: 6377669] [DOI: 10.1038/s41467-019-08424-6]

44. Wang H, Cade BE, Sofer T, Sands SA, Chen H, Browning SR, Stilp AM, Louie TL, Thornton TA, Johnson WC, Below JE, Conomos MP, Evans DS, Gharib SA, Guo X, Wood AC, Mei H, Yaffe K, Loredo JS, Ramos AR, Barrett-Connor E, Ancoli-Israel S, Zee PC, Arens R, Shah NA, Taylor KD, Tranah GJ, Stone KL, Hanis CL, Wilson JG, Gottlieb DJ, Patel SR, Rice K, Post WS, Rotter JI, Sunyaev SR, Cai J, Lin X, Purcell SM, Laurie CC, Saxena R, Redline S, Zhu X. Admixture mapping identifies novel loci for obstructive sleep apnea in Hispanic/Latino Americans. Hum Mol Genet. 2019 Feb 15;28(4):675-687. doi: 10.1093/hmg/ddy387.
[PMID: 30403821] [PMCID: 6360325] [DOI: 10.1093/hmg/ddy387]
45. Cassa CA, Weghorn D, Balick DJ, Jordan DM, Nusinow D, Samocha KE, O'Donnell-Luria A, MacArthur DG, Daly MJ, Beier DR, Sunyaev SR. Reply to 'Selective effects of heterozygous protein-truncating variants'. Nat Genet. 2019 Jan;51(1):3-4. doi: 10.1038/s41588-018-0301-y.
[PMID: 30478437] [PMCID: 9196138] [DOI: 10.1038/s41588-018-0301-y]
46. Gazal S, Loh P, Finucane HK, Ganna A, Schoech A, Sunyaev S, Price AL. Functional architecture of low-frequency variants highlights strength of negative selection across coding and non-coding annotations. Nat Genet. 2018 Nov;50(11):1600-1607. doi: 10.1038/s41588-018-0231-8. Epub 2018 Oct 8.
[PMID: 30297966] [PMCID: 6236676] [DOI: 10.1038/s41588-018-0231-8]
47. Bodea CA, Mitchell AA, Bloemendal A, Day-Williams AG, Runz H, Sunyaev SR. PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. Genome Biol. 2018 Oct 25;19(1):173. doi: 10.1186/s13059-018-1546-6.
[PMID: 30359302] [PMCID: 6203199] [DOI: 10.1186/s13059-018-1546-6]
48. Cassa CA, Jordan DM, Adzhubei I, Sunyaev S. A literature review at genome scale: improving clinical variant assessment. Genet Med. 2018 Sep;20(9):936-941. doi: 10.1038/gim.2017.230. Epub 2018 Feb 1.
[PMID: 29388949] [PMCID: 6070443] [DOI: 10.1038/gim.2017.230]
49. Haghighi A, Krier JB, Toth-Petroczy A, Cassa CA, Frank NY, Carmichael N, Fieg E, Bjonnes A, Mohanty A, Briere LC, Lincoln S, Lucia S, Gupta VA, Söylemez O, Sutti S, Kooshesh K, Qiu H, Fay CJ, Perroni V, Valerius J, Hanna M, Frank A, Ouahed J, Snapper SB, Pantazi A, Chopra SS, Leshchiner I, Stitziel NO, Feldweg A, Mannstadt M, Loscalzo J, Sweetser DA, Liao E, Stoler JM, Nowak CB, Sanchez-Lara PA, Klein OD, Perry H, Patsopoulos NA, Raychaudhuri S, Goessling W, Green RC, Seidman CE, MacRae CA, Sunyaev SR, Maas RL, Vuzman D, Undiagnosed Diseases Network, Brigham and Women’s Hospital FaceBase Project, Brigham Genomic Medicine (BGM). An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. NPJ Genom Med. 2018 Aug 13;3:21. doi: 10.1038/s41525-018-0060-9. eCollection 2018.
[PMID: 30131872] [PMCID: 6089983] [DOI: 10.1038/s41525-018-0060-9]
50. Savova V, Pearl EJ, Boke E, Nag A, Adzhubei I, Horb ME, Peshkin L. Transcriptomic insights into genetic diversity of protein-coding genes in X. laevis. Dev Biol. 2017 Apr 15;424(2):181-188. doi: 10.1016/j.ydbio.2017.02.019. Epub 2017 Mar 7.
[PMID: 28283406] [PMCID: 5405699] [DOI: 10.1016/j.ydbio.2017.02.019]
51. Chen H, Cade BE, Gleason KJ, Bjonnes AC, Stilp AM, Sofer T, Conomos MP, Ancoli-Israel S, Arens R, Azarbarzin A, Bell GI, Below JE, Chun S, Evans DS, Ewert R, Frazier-Wood AC, Gharib SA, Haba-Rubio J, Hagen EW, Heinzer R, Hillman DR, Johnson WC, Kutalik Z, Lane JM, Larkin EK, Lee SK, Liang J,

Loredo JS, Mukherjee S, Palmer LJ, Papanicolaou GJ, Penzel T, Peppard PE, Post WS, Ramos AR, Rice K, Rotter JI, Sands SA, Shah NA, Shin C, Stone KL, Stubbe B, Sul JH, Tafti M, Taylor KD, Teumer A, Thornton TA, Tranah GJ, Wang C, Wang H, Warby SC, Wellman DA, Zee PC, Hanis CL, Laurie CC, Gottlieb DJ, Patel SR, Zhu X, Sunyaev SR, Saxena R, Lin X, Redline S. Multiethnic Meta-Analysis Identifies RAI1 as a Possible Obstructive Sleep Apnea-related Quantitative Trait Locus in Men. Am J Respir Cell Mol Biol. 2018 Mar;58(3):391-401. doi: 10.1165/rcmb.2017-0237OC.
[PMID: 29077507] [PMCID: 5854957] [DOI: 10.1165/rcmb.2017-0237OC]

52. D Antonio M, Weghorn D, D Antonio-Chronowska A, Coulet F, Olson KM, DeBoever C, Drees F, Arias A, Alakus H, Richardson AL, Schwab RB, Farley EK, Sunyaev SR, Frazer KA. Identifying DNase I hypersensitive sites as driver distal regulatory elements in breast cancer. Nat Commun. 2017 Sep 5;8(1):436. doi: 10.1038/s41467-017-00100-x.
[PMID: 28874753] [PMCID: 5585396] [DOI: 10.1038/s41467-017-00100-x]
53. Chandonia J, Adhikari A, Carraro M, Chhibber A, Cutting GR, Fu Y, Gasparini A, Jones DT, Kramer A, Kundu K, Lam HYK, Leonardi E, Moult J, Pal LR, Searls DB, Shah S, Sunyaev S, Tosatto SCE, Yin Y, Buckley BA. Lessons from the CAGI-4 Hopkins clinical panel challenge. Hum Mutat. 2017 Sep;38(9):1155-1168. doi: 10.1002/humu.23225. Epub 2017 Jun 12.
[PMID: 28397312] [PMCID: 5600166] [DOI: 10.1002/humu.23225]
54. Oetting WS, Béroud C, Brenner SE, Greenblatt M, Karchin R, Mooney SD, Sunyaev S. Non-Coding Variation: The 2016 Annual Scientific Meeting of the Human Genome Variation Society. Hum Mutat. 2017 Apr;38(4):460-463. doi: 10.1002/humu.23169. Epub 2017 Feb 14.
[PMID: 28054414] [PMCID: 6546288] [DOI: 10.1002/humu.23169]
55. Wang H, Cade BE, Chen H, Gleason KJ, Saxena R, Feng T, Larkin EK, Vasan RS, Lin H, Patel SR, Tracy RP, Liu Y, Gottlieb DJ, Below JE, Hanis CL, Petty LE, Sunyaev SR, Frazier-Wood AC, Rotter JI, Post W, Lin X, Redline S, Zhu X. Variants in angiopoietin-2 (ANGPT2) contribute to variation in nocturnal oxyhaemoglobin saturation level. Hum Mol Genet. 2016 Dec 1;25(23):5244-5253. doi: 10.1093/hmg/ddw324.
[PMID: 27798093] [PMCID: 6078634] [DOI: 10.1093/hmg/ddw324]
56. Zou J, Valiant G, Valiant P, Karczewski K, Chan SO, Samocha K, Lek M, Sunyaev S, Daly M, MacArthur DG. Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nat Commun. 2016 Oct 31;7:13293. doi: 10.1038/ncomms13293.
[PMID: 27796292] [PMCID: 5095512] [DOI: 10.1038/ncomms13293]
57. Sul JH, Cade BE, Cho MH, Qiao D, Silverman EK, Redline S, Sunyaev S. Increasing Generality and Power of Rare-Variant Tests by Utilizing Extended Pedigrees. Am J Hum Genet. 2016 Oct 6;99(4):846-859. doi: 10.1016/j.ajhg.2016.08.015. Epub 2016 Sep 22.
[PMID: 27666371] [PMCID: 5065687] [DOI: 10.1016/j.ajhg.2016.08.015]
58. Lenz TL, Spirin V, Jordan DM, Sunyaev SR. Excess of Deleterious Mutations around HLA Genes Reveals Evolutionary Cost of Balancing Selection. Mol Biol Evol. 2016 Oct;33(10):2555-64. doi: 10.1093/molbev/msw127. Epub 2016 Jun 28.
[PMID: 27436009] [PMCID: 5026253] [DOI: 10.1093/molbev/msw127]
59. Cade BE, Chen H, Stilp AM, Gleason KJ, Sofer T, Ancoli-Israel S, Arens R, Bell GI, Below JE, Bjonnes AC, Chun S, Conomos MP, Evans DS, Johnson WC, Frazier-Wood AC, Lane JM, Larkin EK, Loredo JS, Post WS, Ramos AR, Rice K, Rotter JI, Shah NA, Stone KL, Taylor KD, Thornton TA, Tranah GJ, Wang C, Zee PC, Hanis CL, Sunyaev SR, Patel SR, Laurie CC, Zhu X, Saxena R, Lin X, Redline S. Genetic Associations with Obstructive Sleep Apnea Traits in Hispanic/Latino Americans. Am J Respir Crit Care Med. 2016 Oct 1;194(7):886-897. doi: 10.1164/rccm.201512-2431OC.

[PMID: 26977737] [PMCID: 5074655] [DOI: 10.1164/rccm.201512-2431OC]

60. Oetting WS, Brenner SE, Brookes AJ, Greenblatt MS, Hart RK, Karchin R, Sunyaev SR, Taschner PE. Pathogenicity Interpretation in the Age of Precision Medicine: The 2015 Annual Scientific Meeting of the Human Genome Variation Society. Hum Mutat. 2016 Apr;37(4):406-11. doi: 10.1002/humu.22958. Epub 2016 Feb 12.
[PMID: 26791113] [PMCID: 4783187] [DOI: 10.1002/humu.22958]
61. Savova V, Chun S, Sohail M, McCole RB, Witwicki R, Gai L, Lenz TL, Wu C, Sunyaev SR, Gimelbrant AA. Genes with monoallelic expression contribute disproportionately to genetic diversity in humans. Nat Genet. 2016 Mar;48(3):231-237. doi: 10.1038/ng.3493. Epub 2016 Jan 25.
[PMID: 26808112] [PMCID: 4942303] [DOI: 10.1038/ng.3493]
62. Cassa CA, Smith SE, Docken W, Hoffman E, McLaughlin H, Chun S, Leshchiner I, Miraoui H, Raychaudhuri S, Frank NY, Wilson BJ, Sunyaev SR, Maas RL, Vuzman D, Brigham Genomic Medicine. An argument for early genomic sequencing in atypical cases: a WISP3 variant leads to diagnosis of progressive pseudorheumatoid arthropathy of childhood. Rheumatology (Oxford). 2016 Mar;55(3):586-9. doi: 10.1093/rheumatology/kev367. Epub 2015 Oct 22.
[PMID: 26493744] [PMCID: 5009447] [DOI: 10.1093/rheumatology/kev367]
63. Palamara PF, Francioli LC, Wilton PR, Genovese G, Gusev A, Finucane HK, Sankararaman S, Sunyaev SR, de Bakker PIW, Wakeley J, Pe'er I, Price AL, Genome of the Netherlands Consortium. Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. Am J Hum Genet. 2015 Dec 3;97(6):775-89. doi: 10.1016/j.ajhg.2015.10.006. Epub 2015 Nov 12.
[PMID: 26581902] [PMCID: 4678427] [DOI: 10.1016/j.ajhg.2015.10.006]
64. Kazanov MD, Roberts SA, Polak P, Stamatoyannopoulos J, Klimczak LJ, Gordenin DA, Sunyaev SR. APOBEC-Induced Cancer Mutations Are Uniquely Enriched in Early-Replicating, Gene-Dense, and Active Chromatin Regions. Cell Rep. 2015 Nov 10;13(6):1103-1109. doi: 10.1016/j.celrep.2015.09.077. Epub 2015 Oct 29.
[PMID: 26527001] [PMCID: 4644490] [DOI: 10.1016/j.celrep.2015.09.077]
65. Chopra SS, Leshchiner I, Duzkale H, McLaughlin H, Giovanni M, Zhang C, Stitziel N, Fingeroth J, Joyce RM, Lebo M, Rehm H, Vuzman D, Maas R, Sunyaev SR, Murray M, Cassa CA. Inherited CHST11/MIR3922 deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. Mol Genet Genomic Med. 2015 Sep;3(5):413-23. doi: 10.1002/mgg3.152. Epub 2015 May 10.
[PMID: 26436107] [PMCID: 4585449] [DOI: 10.1002/mgg3.152]
66. Lenz TL, Deutsch AJ, Han B, Hu X, Okada Y, Eyre S, Knapp M, Zhernakova A, Huizinga TWJ, Abecasis G, Becker J, Boeckxstaens GE, Chen W, Franke A, Gladman DD, Gockel I, Gutierrez-Achury J, Martin J, Nair RP, Nöthen MM, Onengut-Gumuscu S, Rahman P, Rantapää-Dahlqvist S, Stuart PE, Tsoi LC, van Heel DA, Worthington J, Wouters MM, Klareskog L, Elder JT, Gregersen PK, Schumacher J, Rich SS, Wijmenga C, Sunyaev SR, de Bakker PIW, Raychaudhuri S. Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. Nat Genet. 2015 Sep;47(9):1085-90. doi: 10.1038/ng.3379. Epub 2015 Aug 10.
[PMID: 26258845] [PMCID: 4552599] [DOI: 10.1038/ng.3379]
67. Sahni N, Yi S, Taipale M, Fuxman Bass JI, Coulombe-Huntington J, Yang F, Peng J, Weile J, Karras GI, Wang Y, Kovács IA, Kamburov A, Krykbaeva I, Lam MH, Tucker G, Khurana V, Sharma A, Liu Y, Yachie N, Zhong Q, Shen Y, Palagi A, San-Miguel A, Fan C, Balcha D, Dricot A, Jordan DM, Walsh JM, Shah AA, Yang X, Stoyanova AK, Leighton A, Calderwood MA, Jacob Y, Cusick ME, Salehi-Ashtiani K, Whitesell LJ, Sunyaev S, Berger B, Barabási A, Charloteaux B, Hill DE, Hao T, Roth FP, Xia Y, Walhout AJM, Lindquist S, Vidal M. Widespread macromolecular interaction perturbations in human

genetic disorders. Cell. 2015 Apr 23;161(3):647-660. doi: 10.1016/j.cell.2015.04.013.
[PMID: 25910212] [PMCID: 4441215] [DOI: 10.1016/j.cell.2015.04.013]

68. Kundaje A, Meuleman W, Ernst J, Bilenky M, Yen A, Heravi-Moussavi A, Kheradpour P, Zhang Z, Wang J, Ziller MJ, Amin V, Whitaker JW, Schultz MD, Ward LD, Sarkar A, Quon G, Sandstrom RS, Eaton ML, Wu Y, Pfenning AR, Wang X, Claussnitzer M, Liu Y, Coarfa C, Harris RA, Shoresh N, Epstein CB, Gjoneska E, Leung D, Xie W, Hawkins RD, Lister R, Hong C, Gascard P, Mungall AJ, Moore R, Chuah E, Tam A, Canfield TK, Hansen RS, Kaul R, Sabo PJ, Bansal MS, Carles A, Dixon JR, Farh K, Feizi S, Karlic R, Kim A, Kulkarni A, Li D, Lowdon R, Elliott G, Mercer TR, Neph SJ, Onuchic V, Polak P, Rajagopal N, Ray P, Sallari RC, Siebenthall KT, Sinnott-Armstrong NA, Stevens M, Thurman RE, Wu J, Zhang B, Zhou X, Beaudet AE, Boyer LA, De Jager PL, Farnham PJ, Fisher SJ, Haussler D, Jones SJM, Li W, Marra MA, McManus MT, Sunyaev S, Thomson JA, Tlsty TD, Tsai L, Wang W, Waterland RA, Zhang MQ, Chadwick LH, Bernstein BE, Costello JF, Ecker JR, Hirst M, Meissner A, Milosavljevic A, Ren B, Stamatoyannopoulos JA, Wang T, Kellis M, Roadmap Epigenomics Consortium. Integrative analysis of 111 reference human epigenomes. Nature. 2015 Feb 19;518(7539):317-30. doi: 10.1038/nature14248.
[PMID: 25693563] [PMCID: 4530010] [DOI: 10.1038/nature14248]

69. Do R, Stitziel NO, Won H, Jørgensen AB, Duga S, Angelica Merlini P, Kiezun A, Farrall M, Goel A, Zuk O, Guella I, Asselta R, Lange LA, Peloso GM, Auer PL, Girelli D, Martinelli N, Farlow DN, DePristo MA, Roberts R, Stewart AFR, Saleheen D, Danesh J, Epstein SE, Sivapalaratnam S, Hovingh GK, Kastelein JJ, Samani NJ, Schunkert H, Erdmann J, Shah SH, Kraus WE, Davies R, Nikpay M, Johansen CT, Wang J, Hegele RA, Hechter E, Marz W, Kleber ME, Huang J, Johnson AD, Li M, Burke GL, Gross M, Liu Y, Assimes TL, Heiss G, Lange EM, Folsom AR, Taylor HA, Olivieri O, Hamsten A, Clarke R, Reilly DF, Yin W, Rivas MA, Donnelly P, Rossouw JE, Psaty BM, Herrington DM, Wilson JG, Rich SS, Bamshad MJ, Tracy RP, Cupples LA, Rader DJ, Reilly MP, Spertus JA, Cresci S, Hartiala J, Tang WHW, Hazen SL, Allayee H, Reiner AP, Carlson CS, Kooperberg C, Jackson RD, Boerwinkle E, Lander ES, Schwartz SM, Siscovick DS, McPherson R, Tybjaerg-Hansen A, Abecasis GR, Watkins H, Nickerson DA, Ardissino D, Sunyaev SR, O'Donnell CJ, Altshuler D, Gabriel S, Kathiresan S, NHLBI Exome Sequencing Project. Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature. 2015 Feb 5;518(7537):102-6. doi: 10.1038/nature13917. Epub 2014 Dec 10.
[PMID: 25487149] [PMCID: 4319990] [DOI: 10.1038/nature13917]

70. Do R, Balick D, Li H, Adzhubei I, Sunyaev S, Reich D. No evidence that selection has been less effective at removing deleterious mutations in Europeans than in Africans. Nat Genet. 2015 Feb;47(2):126-31. doi: 10.1038/ng.3186. Epub 2015 Jan 12.
[PMID: 25581429] [PMCID: 4310772] [DOI: 10.1038/ng.3186]

71. MacArthur DG, Manolio TA, Dimmock DP, Rehm HL, Shendure J, Abecasis GR, Adams DR, Altman RB, Antonarakis SE, Ashley EA, Barrett JC, Biesecker LG, Conrad DF, Cooper GM, Cox NJ, Daly MJ, Gerstein MB, Goldstein DB, Hirschhorn JN, Leal SM, Pennacchio LA, Stamatoyannopoulos JA, Sunyaev SR, Valle D, Voight BF, Winckler W, Gunter C. Guidelines for investigating causality of sequence variants in human disease. Nature. 2014 Apr 24;508(7497):469-76. doi: 10.1038/nature13127.
[PMID: 24759409] [PMCID: 4180223] [DOI: 10.1038/nature13127]

72. Stanley CM, Sunyaev SR, Greenblatt MS, Oetting WS. Clinically relevant variants - identifying, collecting, interpreting, and disseminating: the 2013 annual scientific meeting of the Human Genome Variation Society. Hum Mutat. 2014 Apr;35(4):505-10. doi: 10.1002/humu.22516.
[PMID: 24470180] [DOI: 10.1002/humu.22516]

73. Sunagawa S, Schloissnig S, Arumugam M, Forslund K, Mitreva M, Tap J, Zhu A, Waller A, Mende DR, Kultima JR, Martin J, Kota K, Sunyaev SR, Typas A, Weinstock GM, Bork P. Individuality and temporal stability of the human gut microbiome. Cent Asian J Glob Health. 2014 Mar 27;2(Suppl):120. doi: 10.5195/cajgh.2013.120. eCollection 2013.
[PMID: 29805877] [PMCID: 5960903] [DOI: 10.5195/cajgh.2013.120]

74. Brownstein CA, Beggs AH, Homer N, Merriman B, Yu TW, Flannery KC, DeChene ET, Towne MC, Savage SK, Price EN, Holm IA, Luquette LJ, Lyon E, Majzoub J, Neupert P, McCallie DJ, Szolovits P, Willard HF, Mendelsohn NJ, Temme R, Finkel RS, Yum SW, Medne L, Sunyaev SR, Adzhubey I, Cassa CA, de Bakker PIW, Duzkale H, Dworzyński P, Fairbrother W, Francioli L, Funke BH, Giovanni MA, Handsaker RE, Lage K, Lebo MS, Lek M, Leshchiner I, MacArthur DG, McLaughlin HM, Murray MF, Pers TH, Polak PP, Raychaudhuri S, Rehm HL, Soemedi R, Stitziel NO, Vestecka S, Supper J, Gugenmus C, Klocke B, Hahn A, Schubach M, Menzel M, Biskup S, Freisinger P, Deng M, Braun M, Perner S, Smith RJH, Andorf JL, Huang J, Ryckman K, Sheffield VC, Stone EM, Bair T, Black-Ziegelbein EA, Braun TA, Darbro B, DeLuca AP, Kolbe DL, Scheetz TE, Shearer AE, Sompallae R, Wang K, Bassuk AG, Edens E, Mathews K, Moore SA, Shchelochkov OA, Trapane P, Bossler A, Campbell CA, Heusel JW, Kwitek A, Maga T, Panzer K, Wassink T, Van Daele D, Azaiez H, Booth K, Meyer N, Segal MM, Williams MS, Tromp G, White P, Corsmeier D, Fitzgerald-Butt S, Herman G, Lamb-Thrush D, McBride KL, Newsom D, Pierson CR, Rakowsky AT, Maver A, Lovrečić L, Palandačić A, Peterlin B, Torkamani A, Wedell A, Huss M, Alexeyenko A, Lindvall JM, Magnusson M, Nilsson D, Stranneheim H, Taylan F, Gilissen C, Hoischen A, van Bon B, Yntema H, Nelen M, Zhang W, Sager J, Zhang L, Blair K, Kural D, Cariaso M, Lennon GG, Javed A, Agrawal S, Ng PC, Sandhu KS, Krishna S, Veeramachaneni V, Isakov O, Halperin E, Friedman E, Shomron N, Glusman G, Roach JC, Caballero J, Cox HC, Mauldin D, Ament SA, Rowen L, Richards DR, San Lucas FA, Gonzalez-Garay ML, Caskey CT, Bai Y, Huang Y, Fang F, Zhang Y, Wang Z, Barrera J, Garcia-Lobo JM, González-Lamuño D, Llorca J, Rodriguez MC, Varela I, Reese MG, De La Vega FM, Kiruluta E, Cargill M, Hart RK, Sorenson JM, Lyon GJ, Stevenson DA, Bray BE, Moore BM, Eilbeck K, Yandell M, Zhao H, Hou L, Chen X, Yan X, Chen M, Li C, Yang C, Gunel M, Li P, Kong Y, Alexander AC, Albertyn ZI, Boycott KM, Bulman DE, Gordon PMK, Innes AM, Knoppers BM, Majewski J, Marshall CR, Parboosingh JS, Sawyer SL, Samuels ME, Schwartzentruber J, Kohane IS, Margulies DM. An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biol. 2014 Mar 25;15(3):R53. doi: 10.1186/gb-2014-15-3-r53.
[PMID: 24667040] [PMCID: 4073084] [DOI: 10.1186/gb-2014-15-3-r53]

75. Zuk O, Schaffner SF, Samocha K, Do R, Hechter E, Kathiresan S, Daly MJ, Neale BM, Sunyaev SR, Lander ES. Searching for missing heritability: designing rare variant association studies. Proc Natl Acad Sci U S A. 2014 Jan 28;111(4):E455-64. doi: 10.1073/pnas.1322563111. Epub 2014 Jan 17.
[PMID: 24443550] [PMCID: 3910587] [DOI: 10.1073/pnas.1322563111]

76. Polak P, Lawrence MS, Haugen E, Stoletzki N, Stojanov P, Thurman RE, Garraway LA, Mirkin S, Getz G, Stamatoyannopoulos JA, Sunyaev SR. Reduced local mutation density in regulatory DNA of cancer genomes is linked to DNA repair. Nat Biotechnol. 2014 Jan;32(1):71-5. doi: 10.1038/nbt.2778. Epub 2013 Dec 15.
[PMID: 24336318] [PMCID: 4116484] [DOI: 10.1038/nbt.2778]

77. Sunyaev SR, Roth FP. Systems biology and the analysis of genetic variation. Curr Opin Genet Dev. 2013 Dec;23(6):599-601. doi: 10.1016/j.gde.2013.11.010. Epub 2013 Nov 28.
[PMID: 24291212] [PMCID: 4116483] [DOI: 10.1016/j.gde.2013.11.010]

78. Agarwala V, Flannick J, Sunyaev S, Altshuler D, GoT2D Consortium. Evaluating empirical bounds on

complex disease genetic architecture. Nat Genet. 2013 Dec;45(12):1418-27. doi: 10.1038/ng.2804. Epub 2013 Oct 20.
[PMID: 24141362] [PMCID: 4158716] [DOI: 10.1038/ng.2804]

79. Lawrence MS, Stojanov P, Polak P, Kryukov GV, Cibulskis K, Sivachenko A, Carter SL, Stewart C, Mermel CH, Roberts SA, Kiezun A, Hammerman PS, McKenna A, Drier Y, Zou L, Ramos AH, Pugh TJ, Stransky N, Helman E, Kim J, Sougnez C, Ambrogio L, Nickerson E, Shefler E, Cortés ML, Auclair D, Saksena G, Voet D, Noble M, DiCara D, Lin P, Lichtenstein L, Heiman DI, Fennell T, Imielinski M, Hernandez B, Hodis E, Baca S, Dulak AM, Lohr J, Landau D, Wu CJ, Melendez-Zajgla J, Hidalgo-Miranda A, Koren A, McCarroll SA, Mora J, Crompton B, Onofrio R, Parkin M, Winckler W, Ardlie K, Gabriel SB, Roberts CWM, Biegel JA, Stegmaier K, Bass AJ, Garraway LA, Meyerson M, Golub TR, Gordenin DA, Sunyaev S, Lander ES, Getz G. Mutational heterogeneity in cancer and the search for new cancer-associated genes. Nature. 2013 Jul 11;499(7457):214-218. doi: 10.1038/nature12213. Epub 2013 Jun 16.
[PMID: 23770567] [PMCID: 3919509] [DOI: 10.1038/nature12213]
80. Goldstein DB, Allen A, Keebler J, Margulies EH, Petrou S, Petrovski S, Sunyaev S. Sequencing studies in human genetics: design and interpretation. Nat Rev Genet. 2013 Jul;14(7):460-70. doi: 10.1038/nrg3455. Epub 2013 Jun 11.
[PMID: 23752795] [PMCID: 4117319] [DOI: 10.1038/nrg3455]
81. Gokcumen O, Zhu Q, Mulder LCF, Iskow RC, Austermann C, Scharer CD, Raj T, Boss JM, Sunyaev S, Price A, Stranger B, Simon V, Lee C. Balancing selection on a regulatory region exhibiting ancient variation that predates human-neandertal divergence. PLoS Genet. 2013 Apr;9(4):e1003404. doi: 10.1371/journal.pgen.1003404. Epub 2013 Apr 11.
[PMID: 23593015] [PMCID: 3623772] [DOI: 10.1371/journal.pgen.1003404]
82. Coste B, Houge G, Murray MF, Stitziel N, Bandell M, Giovanni MA, Philippakis A, Hoischen A, Riemer G, Steen U, Steen VM, Mathur J, Cox J, Lebo M, Rehm H, Weiss ST, Wood JN, Maas RL, Sunyaev SR, Patapoutian A. Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. Proc Natl Acad Sci U S A. 2013 Mar 19;110(12):4667-72. doi: 10.1073/pnas.1221400110. Epub 2013 Mar 4.
[PMID: 23487782] [PMCID: 3607045] [DOI: 10.1073/pnas.1221400110]
83. McFarland CD, Korolev KS, Kryukov GV, Sunyaev SR, Mirny LA. Impact of deleterious passenger mutations on cancer progression. Proc Natl Acad Sci U S A. 2013 Feb 19;110(8):2910-5. doi: 10.1073/pnas.1213968110. Epub 2013 Feb 6.
[PMID: 23388632] [PMCID: 3581883] [DOI: 10.1073/pnas.1213968110]
84. Diogo D, Kurreeman F, Stahl EA, Liao KP, Gupta N, Greenberg JD, Rivas MA, Hickey B, Flannick J, Thomson B, Guiducci C, Ripke S, Adzhubey I, Barton A, Kremer JM, Alfredsson L, Sunyaev S, Martin J, Zhernakova A, Bowes J, Eyre S, Siminovitch KA, Gregersen PK, Worthington J, Klareskog L, Padyukov L, Raychaudhuri S, Plenge RM, Rheumatoid Arthritis Consortium International. Rare, low-frequency, and common variants in the protein-coding sequence of biological candidate genes from GWASs contribute to risk of rheumatoid arthritis. Am J Hum Genet. 2013 Jan 10;92(1):15-27. doi: 10.1016/j.ajhg.2012.11.012. Epub 2012 Dec 20.
[PMID: 23261300] [PMCID: 3542467] [DOI: 10.1016/j.ajhg.2012.11.012]
85. Schloissnig S, Arumugam M, Sunagawa S, Mitreva M, Tap J, Zhu A, Waller A, Mende DR, Kultima JR, Martin J, Kota K, Sunyaev SR, Weinstock GM, Bork P. Genomic variation landscape of the human gut microbiome. Nature. 2013 Jan 3;493(7430):45-50. doi: 10.1038/nature11711. Epub 2012 Dec 5.
[PMID: 23222524] [PMCID: 3536929] [DOI: 10.1038/nature11711]
86. Seim I, Fang X, Xiong Z, Lobanov AV, Huang Z, Ma S, Feng Y, Turanov AA, Zhu Y, Lenz TL,

Gerashchenko MV, Fan D, Hee Yim S, Yao X, Jordan D, Xiong Y, Ma Y, Lyapunov AN, Chen G, Kulakova OI, Sun Y, Lee S, Bronson RT, Moskalev AA, Sunyaev SR, Zhang G, Krogh A, Wang J, Gladyshev VN. Genome analysis reveals insights into physiology and longevity of the Brandt's bat Myotis brandtii. Nat Commun. 2013;4:2212. doi: 10.1038/ncomms3212.
[PMID: 23962925] [PMCID: 3753542] [DOI: 10.1038/ncomms3212]

87. Kiezun A, Pulit SL, Francioli LC, van Dijk F, Swertz M, Boomsma DI, van Duijn CM, Slagboom PE, van Ommen GJB, Wijmenga C, de Bakker PIW, Sunyaev SR, Genome of the Netherlands Consortium. Deleterious alleles in the human genome are on average younger than neutral alleles of the same frequency. PLoS Genet. 2013;9(2):e1003301. doi: 10.1371/journal.pgen.1003301. Epub 2013 Feb 28.
[PMID: 23468643] [PMCID: 3585140] [DOI: 10.1371/journal.pgen.1003301]
88. Adzhubei I, Jordan DM, Sunyaev SR. Predicting functional effect of human missense mutations using PolyPhen-2. Curr Protoc Hum Genet. 2013 Jan;Chapter 7:Unit7.20. doi: 10.1002/0471142905.hg0720s76.
[PMID: 23315928] [PMCID: 4480630] [DOI: 10.1002/0471142905.hg0720s76]
89. Thompson BA, Greenblatt MS, Vallee MP, Herkert JC, Tessereau C, Young EL, Adzhubey IA, Li B, Bell R, Feng B, Mooney SD, Radivojac P, Sunyaev SR, Frebourg T, Hofstra RMW, Sijmons RH, Boucher K, Thomas A, Goldgar DE, Spurdle AB, Tavtigian SV. Calibration of multiple in silico tools for predicting pathogenicity of mismatch repair gene missense substitutions. Hum Mutat. 2013 Jan;34(1):255-65. doi: 10.1002/humu.22214. Epub 2012 Oct 22.
[PMID: 22949387] [PMCID: 4318556] [DOI: 10.1002/humu.22214]
90. Koren A, Polak P, Nemesh J, Michaelson JJ, Sebat J, Sunyaev SR, McCarroll SA. Differential relationship of DNA replication timing to different forms of human mutation and variation. Am J Hum Genet. 2012 Dec 7;91(6):1033-40. doi: 10.1016/j.ajhg.2012.10.018. Epub 2012 Nov 21.
[PMID: 23176822] [PMCID: 3516607] [DOI: 10.1016/j.ajhg.2012.10.018]
91. Nusinow DP, Kiezun A, O'Connell DJ, Chick JM, Yue Y, Maas RL, Gygi SP, Sunyaev SR. Network-based inference from complex proteomic mixtures using SNIPE. Bioinformatics. 2012 Dec 1;28(23):3115-22. doi: 10.1093/bioinformatics/bts594. Epub 2012 Oct 11.
[PMID: 23060611] [PMCID: 3509492] [DOI: 10.1093/bioinformatics/bts594]
92. Sunyaev SR. Inferring causality and functional significance of human coding DNA variants. Hum Mol Genet. 2012 Oct 15;21(R1):R10-7. doi: 10.1093/hmg/dds385. Epub 2012 Sep 17.
[PMID: 22990389] [PMCID: 3459643] [DOI: 10.1093/hmg/dds385]
93. Maurano MT, Humbert R, Rynes E, Thurman RE, Haugen E, Wang H, Reynolds AP, Sandstrom R, Qu H, Brody J, Shafer A, Neri F, Lee K, Kutyavin T, Stehling-Sun S, Johnson AK, Canfield TK, Giste E, Diegel M, Bates D, Hansen RS, Neph S, Sabo PJ, Heimfeld S, Raubitschek A, Ziegler S, Cotsapas C, Sotoodehnia N, Glass I, Sunyaev SR, Kaul R, Stamatoyannopoulos JA. Systematic localization of common disease-associated variation in regulatory DNA. Science. 2012 Sep 7;337(6099):1190-5. doi: 10.1126/science.1222794. Epub 2012 Sep 5.
[PMID: 22955828] [PMCID: 3771521] [DOI: 10.1126/science.1222794]
94. Thurman RE, Rynes E, Humbert R, Vierstra J, Maurano MT, Haugen E, Sheffield NC, Stergachis AB, Wang H, Vernot B, Garg K, John S, Sandstrom R, Bates D, Boatman L, Canfield TK, Diegel M, Dunn D, Ebersol AK, Frum T, Giste E, Johnson AK, Johnson EM, Kutyavin T, Lajoie B, Lee B, Lee K, London D, Lotakis D, Neph S, Neri F, Nguyen ED, Qu H, Reynolds AP, Roach V, Safi A, Sanchez ME, Sanyal A, Shafer A, Simon JM, Song L, Vong S, Weaver M, Yan Y, Zhang Z, Zhang Z, Lenhard B, Tewari M, Dorschner MO, Hansen RS, Navas PA, Stamatoyannopoulos G, Iyer VR, Lieb JD, Sunyaev SR, Akey JM, Sabo PJ, Kaul R, Furey TS, Dekker J, Crawford GE, Stamatoyannopoulos JA. The accessible

chromatin landscape of the human genome. Nature. 2012 Sep 6;489(7414):75-82. doi: 10.1038/nature11232.
[PMID: 22955617] [PMCID: 3721348] [DOI: 10.1038/nature11232]

95. Leshchiner I, Alexa K, Kelsey P, Adzhubei I, Austin-Tse CA, Cooney JD, Anderson H, King MJ, Stottmann RW, Garnaas MK, Ha S, Drummond IA, Paw BH, North TE, Beier DR, Goessling W, Sunyaev SR. Mutation mapping and identification by whole-genome sequencing. Genome Res. 2012 Aug;22(8):1541-8. doi: 10.1101/gr.135541.111. Epub 2012 May 3.
[PMID: 22555591] [PMCID: 3409267] [DOI: 10.1101/gr.135541.111]
96. Tennessen JA, Bigham AW, O'Connor TD, Fu W, Kenny EE, Gravel S, McGee S, Do R, Liu X, Jun G, Kang HM, Jordan D, Leal SM, Gabriel S, Rieder MJ, Abecasis G, Altshuler D, Nickerson DA, Boerwinkle E, Sunyaev S, Bustamante CD, Bamshad MJ, Akey JM, NHLBI Exome Sequencing Project. Evolution and functional impact of rare coding variation from deep sequencing of human exomes. Science. 2012 Jul 6;337(6090):64-9. doi: 10.1126/science.1219240. Epub 2012 May 17.
[PMID: 22604720] [PMCID: 3708544] [DOI: 10.1126/science.1219240]
97. Liberles DA, Teichmann SA, Bahar I, Bastolla U, Bloom J, Bornberg-Bauer E, Colwell LJ, de Koning APJ, Dokholyan NV, Echave J, Elofsson A, Gerloff DL, Goldstein RA, Grahnen JA, Holder MT, Lakner C, Lartillot N, Lovell SC, Naylor G, Perica T, Pollock DD, Pupko T, Regan L, Roger A, Rubinstein N, Shakhnovich E, Sjölander K, Sunyaev S, Teufel AI, Thorne JL, Thornton JW, Weinreich DM, Whelan S. The interface of protein structure, protein biophysics, and molecular evolution. Protein Sci. 2012 Jun;21(6):769-85. doi: 10.1002/pro.2071. Epub 2012 Apr 23.
[PMID: 22528593] [PMCID: 3403413] [DOI: 10.1002/pro.2071]
98. Kiezun A, Garimella K, Do R, Stitziel NO, Neale BM, McLaren PJ, Gupta N, Sklar P, Sullivan PF, Moran JL, Hultman CM, Lichtenstein P, Magnusson P, Lehner T, Shugart YY, Price AL, de Bakker PIW, Purcell SM, Sunyaev SR. Exome sequencing and the genetic basis of complex traits. Nat Genet. 2012 May 29;44(6):623-30. doi: 10.1038/ng.2303.
[PMID: 22641211] [PMCID: 3727622] [DOI: 10.1038/ng.2303]
99. Pasaniuc B, Rohland N, McLaren PJ, Garimella K, Zaitlen N, Li H, Gupta N, Neale BM, Daly MJ, Sklar P, Sullivan PF, Bergen S, Moran JL, Hultman CM, Lichtenstein P, Magnusson P, Purcell SM, Haas DW, Liang L, Sunyaev S, Patterson N, de Bakker PIW, Reich D, Price AL. Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. Nat Genet. 2012 May 20;44(6):631-5. doi: 10.1038/ng.2283.
[PMID: 22610117] [PMCID: 3400344] [DOI: 10.1038/ng.2283]
100. Neale BM, Kou Y, Liu L, Ma'ayan A, Samocha KE, Sabo A, Lin C, Stevens C, Wang L, Makarov V, Polak P, Yoon S, Maguire J, Crawford EL, Campbell NG, Geller ET, Valladares O, Schafer C, Liu H, Zhao T, Cai G, Lihm J, Dannenfelser R, Jabado O, Peralta Z, Nagaswamy U, Muzny D, Reid JG, Newsham I, Wu Y, Lewis L, Han Y, Voight BF, Lim E, Rossin E, Kirby A, Flannick J, Fromer M, Shakir K, Fennell T, Garimella K, Banks E, Poplin R, Gabriel S, DePristo M, Wimbish JR, Boone BE, Levy SE, Betancur C, Sunyaev S, Boerwinkle E, Buxbaum JD, Cook EHJ, Devlin B, Gibbs RA, Roeder K, Schellenberg GD, Sutcliffe JS, Daly MJ. Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature. 2012 Apr 4;485(7397):242-5. doi: 10.1038/nature11011.
[PMID: 22495311] [PMCID: 3613847] [DOI: 10.1038/nature11011]
101. Du R, Litonjua AA, Tantisira KG, Lasky-Su J, Sunyaev SR, Klanderman BJ, Celedón JC, Avila L, Soto-Quiros ME, Weiss ST. Genome-wide association study reveals class I MHC-restricted T cell-associated molecule gene (CRTAM) variants interact with vitamin D levels to affect asthma exacerbations. J Allergy Clin Immunol. 2012 Feb;129(2):368-73, 373.e1-5. doi: 10.1016/j.jaci.2011.09.034. Epub 2011 Nov 1.

[PMID: 22051697] [PMCID: 3360942] [DOI: 10.1016/j.jaci.2011.09.034]

102. Zuk O, Hechter E, Sunyaev SR, Lander ES. The mystery of missing heritability: Genetic interactions create phantom heritability. Proc Natl Acad Sci U S A. 2012 Jan 24;109(4):1193-8. doi: 10.1073/pnas.1119675109. Epub 2012 Jan 5.
[PMID: 22223662] [PMCID: 3268279] [DOI: 10.1073/pnas.1119675109]

103. Kim EB, Fang X, Fushan AA, Huang Z, Lobanov AV, Han L, Marino SM, Sun X, Turanov AA, Yang P, Yim SH, Zhao X, Kasaikina MV, Stoletzki N, Peng C, Polak P, Xiong Z, Kiezun A, Zhu Y, Chen Y, Kryukov GV, Zhang Q, Peshkin L, Yang L, Bronson RT, Buffenstein R, Wang B, Han C, Li Q, Chen L, Zhao W, Sunyaev SR, Park TJ, Zhang G, Wang J, Gladyshev VN. Genome sequencing reveals insights into physiology and longevity of the naked mole rat. Nature. 2011 Oct 12;479(7372):223-7. doi: 10.1038/nature10533.
[PMID: 21993625] [PMCID: 3319411] [DOI: 10.1038/nature10533]

104. Stitziel NO, Kiezun A, Sunyaev S. Computational and statistical approaches to analyzing variants identified by exome sequencing. Genome Biol. 2011 Sep 14;12(9):227. doi: 10.1186/gb-2011-12-9-227.
[PMID: 21920052] [PMCID: 3308043] [DOI: 10.1186/gb-2011-12-9-227]

105. Gorelik L, Reid C, Testa M, Brickelmaier M, Bossolasco S, Pazzi A, Bestetti A, Carmillo P, Wilson E, McAuliffe M, Tonkin C, Carulli JP, Lugovskoy A, Lazzarin A, Sunyaev S, Simon K, Cinque P. Progressive multifocal leukoencephalopathy (PML) development is associated with mutations in JC virus capsid protein VP1 that change its receptor specificity. J Infect Dis. 2011 Jul 1;204(1):103-14. doi: 10.1093/infdis/jir198.
[PMID: 21628664] [PMCID: 3307153] [DOI: 10.1093/infdis/jir198]

106. Spirin V, Shpunt A, Seebacher J, Gentzel M, Shevchenko A, Gygi S, Sunyaev S. Assigning spectrum-specific P-values to protein identifications by mass spectrometry. Bioinformatics. 2011 Apr 15;27(8):1128-34. doi: 10.1093/bioinformatics/btr089. Epub 2011 Feb 23.
[PMID: 21349864] [PMCID: 3072553] [DOI: 10.1093/bioinformatics/btr089]

107. Jordan DM, Kiezun A, Baxter SM, Agarwala V, Green RC, Murray MF, Pugh T, Lebo MS, Rehm HL, Funke BH, Sunyaev SR. Development and validation of a computational method for assessment of missense variants in hypertrophic cardiomyopathy. Am J Hum Genet. 2011 Feb 11;88(2):183-92. doi: 10.1016/j.ajhg.2011.01.011.
[PMID: 21310275] [PMCID: 3035712] [DOI: 10.1016/j.ajhg.2011.01.011]

108. Price AL, Kryukov GV, de Bakker PIW, Purcell SM, Staples J, Wei L, Sunyaev SR. Pooled association tests for rare variants in exon-resequencing studies. Am J Hum Genet. 2010 Jun 11;86(6):832-8. doi: 10.1016/j.ajhg.2010.04.005. Epub 2010 May 13.
[PMID: 20471002] [PMCID: 3032073] [DOI: 10.1016/j.ajhg.2010.04.005]

109. Jordan DM, Ramensky VE, Sunyaev SR. Human allelic variation: perspective from protein function, structure, and evolution. Curr Opin Struct Biol. 2010 Jun;20(3):342-50. doi: 10.1016/j.sbi.2010.03.006.
[PMID: 20399638] [PMCID: 2921592] [DOI: 10.1016/j.sbi.2010.03.006]

110. Adzhubei IA, Schmidt S, Peshkin L, Ramensky VE, Gerasimova A, Bork P, Kondrashov AS, Sunyaev SR. A method and server for predicting damaging missense mutations. Nat Methods. 2010 Apr;7(4):248-9. doi: 10.1038/nmeth0410-248.
[PMID: 20354512] [PMCID: 2855889] [DOI: 10.1038/nmeth0410-248]

111. Molla M, Delcher A, Sunyaev S, Cantor C, Kasif S. Triplet repeat length bias and variation in the human transcriptome. Proc Natl Acad Sci U S A. 2009 Oct 6;106(40):17095-100. doi:

10.1073/pnas.0907112106. Epub 2009 Sep 17.
[PMID: 19805156] [PMCID: 2746125] [DOI: 10.1073/pnas.0907112106]

112. Li JB, Gao Y, Aach J, Zhang K, Kryukov GV, Xie B, Ahlford A, Yoon J, Rosenbaum AM, Zaranek AW, LeProust E, Sunyaev SR, Church GM. Multiplex padlock targeted sequencing reveals human hypermutable CpG variations. Genome Res. 2009 Sep;19(9):1606-15. doi: 10.1101/gr.092213.109. Epub 2009 Jun 12.
[PMID: 19525355] [PMCID: 2752131] [DOI: 10.1101/gr.092213.109]
113. Stamatoyannopoulos JA, Adzhubei I, Thurman RE, Kryukov GV, Mirkin SM, Sunyaev SR. Human mutation rate associated with DNA replication timing. Nat Genet. 2009 Apr;41(4):393-5. doi: 10.1038/ng.363. Epub 2009 Mar 15.
[PMID: 19287383] [PMCID: 2914101] [DOI: 10.1038/ng.363]
114. Kryukov GV, Shpunt A, Stamatoyannopoulos JA, Sunyaev SR. Power of deep, all-exon resequencing for discovery of human trait genes. Proc Natl Acad Sci U S A. 2009 Mar 10;106(10):3871-6. doi: 10.1073/pnas.0812824106. Epub 2009 Feb 6.
[PMID: 19202052] [PMCID: 2656172] [DOI: 10.1073/pnas.0812824106]
115. Sunyaev SR, Lugovskoy A, Simon K, Gorelik L. Adaptive mutations in the JC virus protein capsid are associated with progressive multifocal leukoencephalopathy (PML). PLoS Genet. 2009 Feb;5(2):e1000368. doi: 10.1371/journal.pgen.1000368. Epub 2009 Feb 6.
[PMID: 19197354] [PMCID: 2629573] [DOI: 10.1371/journal.pgen.1000368]
116. Schmidt S, Gerasimova A, Kondrashov FA, Adzhubei IA, Kondrashov AS, Sunyaev S. Erratum. PLoS Genet. 2008 Dec;4(12):10.1371/annotation/a81b1fab-890c-447b-a308-5bc8ca3eb21d. doi: 10.1371/annotation/a81b1fab-890c-447b-a308-5bc8ca3eb21d. Epub 2008 Dec 10.
[PMID: 19096535] [PMCID: 2605301] [DOI: 10.1371/annotation/a81b1fab-890c-447b-a308-5bc8ca3eb21d]
117. Schmidt S, Gerasimova A, Kondrashov FA, Adzhubei IA, Adzuhbei IA, Kondrashov AS, Sunyaev S. Hypermutable non-synonymous sites are under stronger negative selection. PLoS Genet. 2008 Nov;4(11):e1000281. doi: 10.1371/journal.pgen.1000281. Epub 2008 Nov 28.
[PMID: 19043566] [PMCID: 2583910] [DOI: 10.1371/journal.pgen.1000281]
118. Beckstead WA, Bjork BC, Stottmann RW, Sunyaev S, Beier DR. SNP2RFLP: a computational tool to facilitate genetic mapping using benchtop analysis of SNPs. Mamm Genome. 2008 Oct-Dec;19(10-12):687-90. doi: 10.1007/s00335-008-9149-2. Epub 2008 Oct 29.
[PMID: 18958524] [PMCID: 3001109] [DOI: 10.1007/s00335-008-9149-2]
119. Junqueira M, Spirin V, Balbuena TS, Thomas H, Adzhubei I, Sunyaev S, Shevchenko A. Protein identification pipeline for the homology-driven proteomics. J Proteomics. 2008 Aug 21;71(3):346-56. doi: 10.1016/j.jprot.2008.07.003. Epub 2008 Jul 12.
[PMID: 18639657] [PMCID: 2644553] [DOI: 10.1016/j.jprot.2008.07.003]
120. Junqueira M, Spirin V, Santana Balbuena T, Waridel P, Surendranath V, Kryukov G, Adzhubei I, Thomas H, Sunyaev S, Shevchenko A. Separating the wheat from the chaff: unbiased filtering of background tandem mass spectra improves protein identification. J Proteome Res. 2008 Aug;7(8):3382-95. doi: 10.1021/pr800140v. Epub 2008 Jun 18.
[PMID: 18558732] [PMCID: 2842913] [DOI: 10.1021/pr800140v]
121. Boyko AR, Williamson SH, Indap AR, Degenhardt JD, Hernandez RD, Lohmueller KE, Adams MD, Schmidt S, Sninsky JJ, Sunyaev SR, White TJ, Nielsen R, Clark AG, Bustamante CD. Assessing the evolutionary impact of amino acid mutations in the human genome. PLoS Genet. 2008 May 30;4(5):e1000083. doi: 10.1371/journal.pgen.1000083.

[PMID: 18516229] [PMCID: 2377339] [DOI: 10.1371/journal.pgen.1000083]

122. Lohmueller KE, Indap AR, Schmidt S, Boyko AR, Hernandez RD, Hubisz MJ, Sninsky JJ, White TJ, Sunyaev SR, Nielsen R, Clark AG, Bustamante CD. Proportionally more deleterious genetic variation in European than in African populations. Nature. 2008 Feb 21;451(7181):994-7. doi: 10.1038/nature06611.
[PMID: 18288194] [PMCID: 2923434] [DOI: 10.1038/nature06611]

123. Gorlov IP, Gorlova OY, Sunyaev SR, Spitz MR, Amos CI. Shifting paradigm of association studies: value of rare single-nucleotide polymorphisms. Am J Hum Genet. 2008 Jan;82(1):100-12. doi: 10.1016/j.ajhg.2007.09.006.
[PMID: 18179889] [PMCID: 2253956] [DOI: 10.1016/j.ajhg.2007.09.006]

124. Ballif BA, Carey GR, Sunyaev SR, Gygi SP. Large-scale identification and evolution indexing of tyrosine phosphorylation sites from murine brain. J Proteome Res. 2008 Jan;7(1):311-8. doi: 10.1021/pr0701254. Epub 2007 Nov 23.
[PMID: 18034455] [DOI: 10.1021/pr0701254]

125. Asthana S, Roytberg M, Stamatoyannopoulos J, Sunyaev S. Analysis of sequence conservation at nucleotide resolution. PLoS Comput Biol. 2007 Dec;3(12):e254. doi: 10.1371/journal.pcbi.0030254. Epub 2007 Nov 14.
[PMID: 18166073] [PMCID: 2230682] [DOI: 10.1371/journal.pcbi.0030254]

126. Spirin V, Schmidt S, Pertsemlidis A, Cooper RS, Cohen JC, Sunyaev SR. Common single-nucleotide polymorphisms act in concert to affect plasma levels of high-density lipoprotein cholesterol. Am J Hum Genet. 2007 Dec;81(6):1298-303. doi: 10.1086/522497.
[PMID: 17952847] [PMCID: 2276349] [DOI: 10.1086/522497]

127. Ilyinskii PO, Gabai VL, Sunyaev SR, Thoidis G, Shneider AM. Toxicity of influenza A virus matrix protein 2 for mammalian cells is associated with its intrinsic proton-channeling activity. Cell Cycle. 2007 Aug 15;6(16):2043-7. doi: 10.4161/cc.6.16.4564. Epub 2007 Jun 12.
[PMID: 17700063] [DOI: 10.4161/cc.6.16.4564]

128. Asthana S, Noble WS, Kryukov G, Grant CE, Sunyaev S, Stamatoyannopoulos JA. Widely distributed noncoding purifying selection in the human genome. Proc Natl Acad Sci U S A. 2007 Jul 24;104(30):12410-5. doi: 10.1073/pnas.0705140104. Epub 2007 Jul 17.
[PMID: 17640883] [PMCID: 1941483] [DOI: 10.1073/pnas.0705140104]

129. Waridel P, Frank A, Thomas H, Surendranath V, Sunyaev S, Pevzner P, Shevchenko A. Sequence similarity-driven proteomics in organisms with unknown genomes by LC-MS/MS and automated de novo sequencing. Proteomics. 2007 Jul;7(14):2318-29. doi: 10.1002/pmic.200700003.
[PMID: 17623296] [DOI: 10.1002/pmic.200700003]

130. Birney E, Stamatoyannopoulos JA, Dutta A, Guigó R, Gingeras TR, Margulies EH, Weng Z, Snyder M, Dermitzakis ET, Thurman RE, Kuehn MS, Taylor CM, Neph S, Koch CM, Asthana S, Malhotra A, Adzhubei I, Greenbaum JA, Andrews RM, Flicek P, Boyle PJ, Cao H, Carter NP, Clelland GK, Davis S, Day N, Dhami P, Dillon SC, Dorschner MO, Fiegler H, Giresi PG, Goldy J, Hawrylycz M, Haydock A, Humbert R, James KD, Johnson BE, Johnson EM, Frum TT, Rosenzweig ER, Karnani N, Lee K, Lefebvre GC, Navas PA, Neri F, Parker SCJ, Sabo PJ, Sandstrom R, Shafer A, Vetrie D, Weaver M, Wilcox S, Yu M, Collins FS, Dekker J, Lieb JD, Tullius TD, Crawford GE, Sunyaev S, Noble WS, Dunham I, Denoeud F, Reymond A, Kapranov P, Rozowsky J, Zheng D, Castelo R, Frankish A, Harrow J, Ghosh S, Sandelin A, Hofacker IL, Baertsch R, Keefe D, Dike S, Cheng J, Hirsch HA, Sekinger EA, Lagarde J, Abril JF, Shahab A, Flamm C, Fried C, Hackermüller J, Hertel J, Lindemeyer M, Missal K, Tanzer A, Washietl S, Korbel J, Emanuelsson O, Pedersen JS, Holroyd N, Taylor R,

Swarbreck D, Matthews N, Dickson MC, Thomas DJ, Weirauch MT, Gilbert J, Drenkow J, Bell I, Zhao X, Srinivasan KG, Sung W, Ooi HS, Chiu KP, Foissac S, Alioto T, Brent M, Pachter L, Tress ML, Valencia A, Choo SW, Choo CY, Ucla C, Manzano C, Wyss C, Cheung E, Clark TG, Brown JB, Ganesh M, Patel S, Tammana H, Chrast J, Henrichsen CN, Kai C, Kawai J, Nagalakshmi U, Wu J, Lian Z, Lian J, Newburger P, Zhang X, Bickel P, Mattick JS, Carninci P, Hayashizaki Y, Weissman S, Hubbard T, Myers RM, Rogers J, Stadler PF, Lowe TM, Wei C, Ruan Y, Struhl K, Gerstein M, Antonarakis SE, Fu Y, Green ED, Karaöz U, Siepel A, Taylor J, Liefer LA, Wetterstrand KA, Good PJ, Feingold EA, Guyer MS, Cooper GM, Asimenos G, Dewey CN, Hou M, Nikolaev S, Montoya-Burgos JI, Löytynoja A, Whelan S, Pardi F, Massingham T, Huang H, Zhang NR, Holmes I, Mullikin JC, Ureta-Vidal A, Paten B, Seringhaus M, Church D, Rosenbloom K, Kent WJ, Stone EA, Batzoglou S, Goldman N, Hardison RC, Haussler D, Miller W, Sidow A, Trinklein ND, Zhang ZD, Barrera L, Stuart R, King DC, Ameur A, Enroth S, Bieda MC, Kim J, Bhinge AA, Jiang N, Liu J, Yao F, Vega VB, Lee CWH, Ng P, Shahab A, Yang A, Moqtaderi Z, Zhu Z, Xu X, Squazzo S, Oberley MJ, Inman D, Singer MA, Richmond TA, Munn KJ, Rada-Iglesias A, Wallerman O, Komorowski J, Fowler JC, Couttet P, Bruce AW, Dovey OM, Ellis PD, Langford CF, Nix DA, Euskirchen G, Hartman S, Urban AE, Kraus P, Van Calcar S, Heintzman N, Kim TH, Wang K, Qu C, Hon G, Luna R, Glass CK, Rosenfeld MG, Aldred SF, Cooper SJ, Halees A, Lin JM, Shulha HP, Zhang X, Xu M, Haidar JNS, Yu Y, Ruan Y, Iyer VR, Green RD, Wadelius C, Farnham PJ, Ren B, Harte RA, Hinrichs AS, Trumbower H, Clawson H, Hillman-Jackson J, Zweig AS, Smith K, Thakkapallayil A, Barber G, Kuhn RM, Karolchik D, Armengol L, Bird CP, de Bakker PIW, Kern AD, Lopez-Bigas N, Martin JD, Stranger BE, Woodroffe A, Davydov E, Dimas A, Eyras E, Hallgrímsdóttir IB, Huppert J, Zody MC, Abecasis GR, Estivill X, Bouffard GG, Guan X, Hansen NF, Idol JR, Maduro VVB, Maskeri B, McDowell JC, Park M, Thomas PJ, Young AC, Blakesley RW, Muzny DM, Sodergren E, Wheeler DA, Worley KC, Jiang H, Weinstock GM, Gibbs RA, Graves T, Fulton R, Mardis ER, Wilson RK, Clamp M, Cuff J, Gnerre S, Jaffe DB, Chang JL, Lindblad-Toh K, Lander ES, Koriabine M, Nefedov M, Osoegawa K, Yoshinaga Y, Zhu B, de Jong PJ, Children's Hospital Oakland Research Institute. Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature. 2007 Jun 14;447(7146):799-816. doi: 10.1038/nature05874.
[PMID: 17571346] [PMCID: 2212820] [DOI: 10.1038/nature05874]

131. Ahituv N, Kavaslar N, Schackwitz W, Ustaszewska A, Martin J, Hebert S, Doelle H, Ersoy B, Kryukov G, Schmidt S, Yosef N, Ruppin E, Sharan R, Vaisse C, Sunyaev S, Dent R, Cohen J, McPherson R, Pennacchio LA. Medical sequencing at the extremes of human body mass. Am J Hum Genet. 2007 Apr;80(4):779-91. doi: 10.1086/513471. Epub 2007 Mar 5.
[PMID: 17357083] [PMCID: 1852707] [DOI: 10.1086/513471]

132. Kryukov GV, Pennacchio LA, Sunyaev SR. Most rare missense alleles are deleterious in humans: implications for complex disease and association studies. Am J Hum Genet. 2007 Apr;80(4):727-39. doi: 10.1086/513473. Epub 2007 Mar 8.
[PMID: 17357078] [PMCID: 1852724] [DOI: 10.1086/513473]

133. Liska AJ, Sunyaev S, Shilov IN, Schaeffer DA, Shevchenko A. Error-tolerant EST database searches by tandem mass spectrometry and multiTag software. Proteomics. 2005 Nov;5(16):4118-22. doi: 10.1002/pmic.200401262.
[PMID: 16121338] [DOI: 10.1002/pmic.200401262]

134. Keightley PD, Kryukov GV, Sunyaev S, Halligan DL, Gaffney DJ. Evolutionary constraints in conserved nongenic sequences of mammals. Genome Res. 2005 Oct;15(10):1373-8. doi: 10.1101/gr.3942005.
[PMID: 16204190] [PMCID: 1240079] [DOI: 10.1101/gr.3942005]

135. Kryukov GV, Schmidt S, Sunyaev S. Small fitness effect of mutations in highly conserved non-

coding regions. Hum Mol Genet. 2005 Aug 1;14(15):2221-9. doi: 10.1093/hmg/ddi226. Epub 2005 Jun 30.
[PMID: 15994173] [DOI: 10.1093/hmg/ddi226]

136. Jordan IK, Kondrashov FA, Adzhubei IA, Wolf YI, Koonin EV, Kondrashov AS, Sunyaev S. A universal trend of amino acid gain and loss in protein evolution. Nature. 2005 Feb 10;433(7026):633-8. doi: 10.1038/nature03306. Epub 2005 Jan 19.
[PMID: 15660107] [DOI: 10.1038/nature03306]

137. Sunyaev S. Biocomputing enters its adolescence. Genome Biol. 2005;6(6):325. doi: 10.1186/gb-2005-6-6-325. Epub 2005 May 31.
[PMID: 15960815] [PMCID: 1175967] [DOI: 10.1186/gb-2005-6-6-325]

138. Dimmic MW, Sunyaev S, Bustamante CD. Inferring SNP function using evolutionary, structural, and computational methods. Pac Symp Biocomput. 2005:382-4.
[PMID: 15759643]

139. Asthana S, Schmidt S, Sunyaev S. A limited role for balancing selection. Trends Genet. 2005 Jan;21(1):30-2. doi: 10.1016/j.tig.2004.11.001.
[PMID: 15680511] [DOI: 10.1016/j.tig.2004.11.001]

140. Liska AJ, Popov AV, Sunyaev S, Coughlin P, Habermann B, Shevchenko A, Bork P, Karsenti E, Shevchenko A. Homology-based functional proteomics by mass spectrometry: application to the Xenopus microtubule-associated proteome. Proteomics. 2004 Sep;4(9):2707-21. doi: 10.1002/pmic.200300813.
[PMID: 15352245] [DOI: 10.1002/pmic.200300813]

141. Ogurtsov AY, Sunyaev S, Kondrashov AS. Indel-based evolutionary distance and mouse-human divergence. Genome Res. 2004 Aug;14(8):1610-6. doi: 10.1101/gr.2450504.
[PMID: 15289479] [PMCID: 509270] [DOI: 10.1101/gr.2450504]

142. Bazykin GA, Kondrashov FA, Ogurtsov AY, Sunyaev S, Kondrashov AS. Positive selection at sites of multiple amino acid replacements since rat-mouse divergence. Nature. 2004 Jun 3;429(6991):558-62. doi: 10.1038/nature02601.
[PMID: 15175752] [DOI: 10.1038/nature02601]

143. Habermann B, Oegema J, Sunyaev S, Shevchenko A. The power and the limitations of cross-species protein identification by mass spectrometry-driven sequence similarity searches. Mol Cell Proteomics. 2004 Mar;3(3):238-49. doi: 10.1074/mcp.M300073-MCP200. Epub 2003 Dec 26.
[PMID: 14695901] [DOI: 10.1074/mcp.M300073-MCP200]

144. Sunyaev SR, Bogopolsky GA, Oleynikova NV, Vlasov PK, Finkelstein AV, Roytberg MA. From analysis of protein structural alignments toward a novel approach to align protein sequences. Proteins. 2004 Feb 15;54(3):569-82. doi: 10.1002/prot.10503.
[PMID: 14748004] [DOI: 10.1002/prot.10503]

145. Sunyaev S, Kondrashov FA, Bork P, Ramensky V. Impact of selection, mutation rate and genetic drift on human genetic variation. Hum Mol Genet. 2003 Dec 15;12(24):3325-30. doi: 10.1093/hmg/ddg359. Epub 2003 Oct 21.
[PMID: 14570704] [DOI: 10.1093/hmg/ddg359]

146. Schmidt S, Sunyaev S, Bork P, Dandekar T. Metabolites: a helping hand for pathway evolution?. Trends Biochem Sci. 2003 Jun;28(6):336-41. doi: 10.1016/S0968-0004(03)00114-2.
[PMID: 12826406] [DOI: 10.1016/S0968-0004(03)00114-2]

147. Sunyaev S, Liska AJ, Golod A, Shevchenko A, Shevchenko A. MultiTag: multiple error-tolerant sequence tag search for the sequence-similarity identification of proteins by mass spectrometry.

Anal Chem. 2003 Mar 15;75(6):1307-15. doi: 10.1021/ac026199a.
[PMID: 12659190] [DOI: 10.1021/ac026199a]

148. Stark A, Sunyaev S, Russell RB. A model for statistical significance of local similarities in structure. J Mol Biol. 2003 Mar 7;326(5):1307-16. doi: 10.1016/s0022-2836(03)00045-7.
[PMID: 12595245] [DOI: 10.1016/s0022-2836(03)00045-7]

149. Kriventseva EV, Koch I, Apweiler R, Vingron M, Bork P, Gelfand MS, Sunyaev S. Increase of functional diversity by alternative splicing. Trends Genet. 2003 Mar;19(3):124-8. doi: 10.1016/S0168-9525(03)00023-4.
[PMID: 12615003] [DOI: 10.1016/S0168-9525(03)00023-4]

150. Shevchenko A, Sunyaev S, Liska A, Bork P, Shevchenko A. Nanoelectrospray tandem mass spectrometry and sequence similarity searching for identification of proteins from organisms with unknown genomes. Methods Mol Biol. 2003;211:221-34. doi: 10.1385/1-59259-342-9:221.
[PMID: 12489434] [DOI: 10.1385/1-59259-342-9:221]

151. Kondrashov AS, Sunyaev S, Kondrashov FA. Dobzhansky-Muller incompatibilities in protein evolution. Proc Natl Acad Sci U S A. 2002 Nov 12;99(23):14878-83. doi: 10.1073/pnas.232565499. Epub 2002 Oct 28.
[PMID: 12403824] [PMCID: 137512] [DOI: 10.1073/pnas.232565499]

152. Ramensky V, Bork P, Sunyaev S. Human non-synonymous SNPs: server and survey. Nucleic Acids Res. 2002 Sep 1;30(17):3894-900. doi: 10.1093/nar/gkf493.
[PMID: 12202775] [PMCID: 137415] [DOI: 10.1093/nar/gkf493]

153. Shevchenko A, Sunyaev S, Loboda A, Shevchenko A, Bork P, Ens W, Standing KG. Charting the proteomes of organisms with unsequenced genomes by MALDI-quadrupole time-of-flight mass spectrometry and BLAST homology searching. Anal Chem. 2001 May 1;73(9):1917-26. doi: 10.1021/ac0013709.
[PMID: 11354471] [DOI: 10.1021/ac0013709]

154. Sunyaev S, Ramensky V, Koch I, Lathe W3, Kondrashov AS, Bork P. Prediction of deleterious human alleles. Hum Mol Genet. 2001 Mar 15;10(6):591-7. doi: 10.1093/hmg/10.6.591.
[PMID: 11230178] [DOI: 10.1093/hmg/10.6.591]

155. Sunyaev S, Lathe W3, Bork P. Integration of genome data and protein structures: prediction of protein folds, protein interactions and "molecular phenotypes" of single nucleotide polymorphisms. Curr Opin Struct Biol. 2001 Feb;11(1):125-30. doi: 10.1016/s0959-440x(00)00175-5.
[PMID: 11179902] [DOI: 10.1016/s0959-440x(00)00175-5]

156. Sunyaev SR, Lathe WC3, Ramensky VE, Bork P. SNP frequencies in human genes an excess of rare alleles and differing modes of selection. Trends Genet. 2000 Aug;16(8):335-7. doi: 10.1016/s0168-9525(00)02058-8.
[PMID: 10904261] [DOI: 10.1016/s0168-9525(00)02058-8]

157. Sunyaev S, Ramensky V, Bork P. Towards a structural basis of human non-synonymous single nucleotide polymorphisms. Trends Genet. 2000 May;16(5):198-200. doi: 10.1016/s0168-9525(00)01988-0.
[PMID: 10782110] [DOI: 10.1016/s0168-9525(00)01988-0]

158. Sunyaev S, Hanke J, Brett D, Aydin A, Zastrow I, Lathe W, Bork P, Reich J. Individual variation in protein-coding sequences of human genome. Adv Protein Chem. 2000;54:409-37. doi: 10.1016/s0065-3233(00)54012-1.
[PMID: 10829234] [DOI: 10.1016/s0065-3233(00)54012-1]

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