ournal	Title	Noncoding Annotation Tool	Reference
N	Common genetic variants influence human subcortical brain structures	N	Hibar et al.[1]
N	New genetic loci link adipose and insulin biology to body fat distribution	Y	Shungin et al.[2]
N	Genetic studies of body mass index yield new insights for obesity biology	Y	Locke et al.[3]
N	Sparsewhole-genome sequencing identifies two loci for major depressive disorder	N	Converge Consortium[4]
N	Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture	Y	Zheng et al.[5]
N	A novel locus of resistance to severe malaria in a region of ancient balancing selection	N	Malaria Genomic Epidemiology Network[6]
N	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism	N	Oldridge et al.[7]
N	Sex-dependent dominance at a single locus maintains variation in age at maturity in salmon	Y	Barson et al.[8]
NG	Identification of six new susceptibility loci for invasive epithelial ovarian cancer	N	Kuchenbaecker et al.[9]
NG	Discovery of six new susceptibility loci and analysis of pleiotropic effects in leprosy	N	Liu et al.[10]
NG	Common variants in *ACYP2* influence susceptibility to cisplatin-induced hearing loss	N	Xu et al.[11]
NG	A common variant mapping to *CACNA1A* is associated with susceptibility to exfoliation syndrome	Y	Aung et al.[12]
NG	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer	Y	Michailidou et al.[13]
NG	Susceptibility to tuberculosis is associated with variants in the *ASAP1* gene encoding a regulator of dendritic cell migration	N	Curtis et al.[14]
NG	Common variants at 10p12.31, 10q21.1 and 13q12.13 are associated with sporadic pituitary adenoma	N	Ye et al.[15]
NG	Genome-wide significant risk associations for mucinous ovarian carcinoma	N	The Ovarian Cancer Association Consortium[16]
NG	Loss-of-function variants in *ATM* confer risk of gastric cancer	Y	Helgason et al.[17]
NG	Common variation at 2p13.3, 3q29, 7p13 and 17q25.1 associated with susceptibility to pancreatic cancer	Y	Childs et al.[18]
NG	Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations	N	Liu et al.[19]

NG	Chimeric EWSR1-FLI1 regulates the Ewing sarcoma susceptibility gene *EGR2* via a GGAA microsatellite	N	Grünewald et al.[20]
NG	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma	Y	Law et al.[21]
NG	A coding variant in *RARG* confers susceptibility to anthracycline-induced cardiotoxicity in childhood cancer	N	Aminkeng et al.[22]
NG	Height-reducing variants and selection for short stature in Sardinia	N	Zoledziewska et al.[23]
NG	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers	Y	Sidore et al.[24]
NG	Trans-ancestry genome-wide association study identifies	Y	Kato et al.[25]
NG	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus	N	Bentham et al.[26]
NG	A genome-wide association study confirms *PNPLA3* and identifies *TM6SF2* and *MBOAT7* as risk loci for alcohol-related cirrhosis	N	Buch et al.[27]
NG	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci	N	Gaulton et al.[28]
NG	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants	N	Fritsche et al.[29]
NAR	Substantial DNA methylation differences between two major neuronal subtypes in human brain	N	Kozlenkov et al.[30]
NAR	Discover hidden splicing variations by mapping personal transcriptomes to personal genomes	N	Stein et al.[31]
AJHG	Genome-wide Comparative Analysis of Atopic Dermatitis and Psoriasis Gives Insight into Opposing Genetic Mechanisms	N	Baurecht et al.[32]
AJHG	UBE2L3 Polymorphism Amplifies NF-kB Activation and Promotes Plasma Cell Development, Linking Linear Ubiquitination to Multiple Autoimmune Diseases	N	Lewis et al.[33]
AJHG	Identification of Functional Variants for Cleft Lip with or without Cleft Palate in or near PAX7, FGFR2,and NOG by Targeted Sequencing of GWAS Loci	Y	Leslie et al.[34]
AJHG	Meta-analysis of 65,734 Individuals Identifies TSPAN15 and SLC44A2 as Two Susceptibility Loci for Venous Thromboembolism	Y	Germain et al.[35]
AJHG	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability	N	Jiang et al.[36]
AJHG	Genome-wide Analysis of Body Proportion Classifies Height-Associated Variants by Mechanism of Action and Implicates Genes Important for Skeletal Development	N	Chan et al.[37]
AJHG	Lupus Risk Variant Increases pSTAT1 Binding and Decreases ETS1 Expression	N	Lu et al.[38]
AJHG	A Functional SNP in BNC2 Is Associated with Adolescent Idiopathic Scoliosis	N	Ogura et al.[39]
AJHG	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression	Y	Darabi et al.[40]

AJHG	Long-Range Modulation of PAG1 Expression by 8q21 Allergy Risk Variants	Y	Vicente et al.[41]
AJHG	Amino Acid Variation in HLA Class II Proteins Is a Major Determinant of Humoral Response to Common Viruses	N	Hammer et al.[42]
AJHG	Multiple Hepatic Regulatory Variants Multiple Hepatic Regulatory Variants with High-Density Lipoprotein Cholesterol at the GALNT2 GWAS Locus Associated	N	Roman et al.[43]
AJHG	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture	Y	Stuart et al.[44]

1. Hibar, D. P. *et al.* Common genetic variants influence human subcortical brain structures. *Nature* **520,** 224–229 (2015).
2. Shungin, D. *et al.* New genetic loci link adipose and insulin biology to body fat distribution. *Nature* **518,** 187–196 (2015).
3. Locke, A. E. *et al.* Genetic studies of body mass index yield new insights for obesity biology. *Nature* **518,** 197–206 (2015).
4. CONVERGE consortium. Sparse whole-genome sequencing identifies two loci for major depressive disorder. *Nature* **523,** 588–591 (2015).
5. Zheng, H. F. *et al.* Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture. *Nature* **526,** 112–117 (2015).
6. Malaria Genomic Epidemiology Network *et al.* A novel locus of resistance to severe malaria in a region of ancient balancing selection. *Nature* **526,** 253–257 (2015).
7. Oldridge, D. A. *et al.* Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. *Nature* **528,** 418–421 (2015).
8. Barson, N. J. *et al.* Sex-dependent dominance at a single locus maintains variation in age at maturity in salmon. *Nature* **528,** 405–408 (2015).
9. Kuchenbaecker, K. B. *et al.* Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nat. Genet.* **47,** 164–171 (2015).
10. Liu, H. *et al.* Discovery of six new susceptibility loci and analysis of pleiotropic effects in leprosy. *Nat. Genet.* **47,** 267–271 (2015).
11. Xu, H. *et al.* Common variants in ACYP2 influence susceptibility to cisplatin-induced hearing loss. *Nat. Genet.* **47,** 263–266 (2015).
12. Aung, T. *et al.* A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. *Nat. Genet.* **47,** 387–392 (2015).
13. Michailidou, K. *et al.* Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nat. Genet.* **47,** 373–380 (2015).
14. Curtis, J. *et al.* Susceptibility to tuberculosis is associated with variants in the ASAP1 gene encoding a regulator of dendritic cell migration. *Nat. Genet.* **47,** 523–527 (2015).
15. Ye, Z. *et al.* Common variants at 10p12.31, 10q21.1 and 13q12.13 are associated with sporadic pituitary adenoma. *Nat. Genet.* **47,** 793–797 (2015).

16. Kelemen, L. E. *et al.* Genome-wide significant risk associations for mucinous ovarian carcinoma. *Nat. Genet.* **47,** 888–897 (2015).
17. Helgason, H. *et al.* Loss-of-function variants in ATM confer risk of gastric cancer. *Nat. Genet.* **47,** 906–910 (2015).
18. Childs, E. J. *et al.* Common variation at 2p13.3, 3q29, 7p13 and 17q25.1 associated with susceptibility to pancreatic cancer. *Nat. Genet.* **47,** 911–916 (2015).
19. Liu, J. Z. *et al.* Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. *Nat. Genet.* **47,** 979–986 (2015).
20. Grünewald, T. G. P. *et al.* Chimeric EWSR1-FLI1 regulates the Ewing sarcoma susceptibility gene EGR2 via a GGAA microsatellite. *Nat. Genet.* **47,** 1073–1078 (2015).
21. Law, M. H. *et al.* Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. *Nat. Genet.* **47,** 987–995 (2015).
22. Aminkeng, F. *et al.* A coding variant in RARG confers susceptibility to anthracycline-induced cardiotoxicity in childhood cancer. *Nat. Genet.* **47,** 1079–1084 (2015).
23. Zoledziewska, M. *et al.* Height-reducing variants and selection for short stature in Sardinia. *Nat. Genet.* **47,** 1352–1356 (2015).
24. Sidore, C. *et al.* Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. *Nat. Genet.* **47,** 1272–1281 (2015).
25. Kato, N. *et al.* Trans-ancestry genome-wide association study identifies 12 genetic loci influencing blood pressure and implicates a role for DNA methylation. *Nat. Genet.* **47,** 1282–1293 (2015).
26. Bentham, J. *et al.* Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus. *Nat. Genet.* **47,** 1457–1464 (2015).
27. Buch, S. *et al.* A genome-wide association study confirms PNPLA3 and identifies TM6SF2 and MBOAT7 as risk loci for alcohol-related cirrhosis. *Nat. Genet.* **47,** 1443–1448 (2015).
28. Gaulton, K. J. *et al.* Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. *Nat. Genet.* **47,** 1415–1425 (2015).
29. Fritsche, L. G. *et al.* A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nat. Genet.* **48,** 134–143 (2016).
30. Kozlenkov, A. *et al.* Substantial DNA methylation differences between two major neuronal subtypes in human brain. *Nucleic Acids Res.* **44,** 2593–2612 (2016).
31. Stein, S., Lu, Z.-X., Bahrami-Samani, E., Park, J. W. & Xing, Y. Discover hidden splicing variations by mapping personal transcriptomes to personal genomes. *Nucleic Acids Res.* **43,** 10612–10622 (2015).
32. Baurecht, H. *et al.* Genome-wide comparative analysis of atopic dermatitis and psoriasis gives insight into opposing genetic mechanisms. *Am. J. Hum. Genet.*

96, 104–120 (2015).
33. Lewis, M. J. *et al.* UBE2L3 polymorphism amplifies NF-κB activation and promotes plasma cell development, linking linear ubiquitination to multiple autoimmune diseases. *Am. J. Hum. Genet.* **96,** 221–234 (2015).
34. Leslie, E. J. *et al.* Identification of functional variants for cleft lip with or without cleft palate in or near PAX7, FGFR2, and NOG by targeted sequencing of GWAS loci. *Am. J. Hum. Genet.* **96,** 397–411 (2015).
35. Germain, M. *et al.* Meta-analysis of 65,734 individuals identifies TSPAN15 and SLC44A2 as two susceptibility loci for venous thromboembolism. *Am. J. Hum. Genet.* **96,** 532–542 (2015).
36. Jiang, Q. *et al.* Functional loss of semaphorin 3C and/or semaphorin 3D and their epistatic interaction with ret are critical to Hirschsprung disease liability. *Am. J. Hum. Genet.* **96,** 581–596 (2015).
37. Chan, Y. *et al.* Genome-wide Analysis of Body Proportion Classifies Height-Associated Variants by Mechanism of Action and Implicates Genes Important for Skeletal Development. *Am. J. Hum. Genet.* **96,** 695–708 (2015).
38. Lu, X. *et al.* Lupus Risk Variant Increases pSTAT1 Binding and Decreases ETS1 Expression. *Am. J. Hum. Genet.* **96,** 731–739 (2015).
39. Ogura, Y. *et al.* A Functional SNP in BNC2 Is Associated with Adolescent Idiopathic Scoliosis. *Am. J. Hum. Genet.* **97,** 337–342 (2015).
40. Darabi, H. *et al.* Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. *Am. J. Hum. Genet.* **97,** 22–34 (2015).
41. Vicente, C. T. *et al.* Long-Range Modulation of PAG1 Expression by 8q21 Allergy Risk Variants. *Am. J. Hum. Genet.* **97,** 329–336 (2015).
42. Hammer, C. *et al.* Amino Acid Variation in HLA Class II Proteins Is a Major Determinant of Humoral Response to Common Viruses. *Am. J. Hum. Genet.* **97,** 738–743 (2015).
43. Roman, T. S. *et al.* Multiple Hepatic Regulatory Variants at the GALNT2 GWAS Locus Associated with High-Density Lipoprotein Cholesterol. *Am. J. Hum. Genet.* **97,** 801–815 (2015).
44. Stuart, P. E. *et al.* Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. *Am. J. Hum. Genet.* **97,** 816–836 (2015).