# Sadaf Farooqi

List of Publications by Year
in descending order



| 179 | 30,196 | 14124 | 69 | 5873 | 166 |
|---|---|---|---|---|---|
| papers | citations | h-index | | g-index | |

| 195 | 195 | 195 | 36607 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. Clinical Endocrinology, 2022, 96, 270-275. | 1.2 | 6 |
| 2 | Obesity Due to Steroid Receptor Coactivator-1 Deficiency Is Associated With Endocrine and Metabolic Abnormalities. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2532-e2544. | 1.8 | 5 |
| 3 | Central obesity is selectively associated with cerebral gray matter atrophy in 15,634 subjects in the UK Biobank. International Journal of Obesity, 2022, 46, 1059-1067. | 1.6 | 12 |
| 4 | Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors. Biological Psychiatry, 2022, 91, 856-859. | 0.7 | 15 |
| 5 | Visualization of sympathetic neural innervation in human white adipose tissue. Open Biology, 2022, 12, 210345. | 1.5 | 2 |
| 6 | The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742. | 1.1 | 56 |
| 7 | Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575. | 1.1 | 16 |
| 8 | Monogenic human obesity syndromes. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 181, 301-310. | 1.0 | 18 |
| 9 | TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459. | 1.1 | 26 |
| 10 | Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. Cell Reports, 2021, 34, 108862. | 2.9 | 37 |
| 11 | Reply to Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. Cell, 2021, 184, 1652-1653. | 13.5 | 3 |
| 12 | Putting a brake on hunger. Science, 2021, 372, 792-793. | 6.0 | 0 |
| 13 | CorneliaÂde Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. Nature Communications, 2021, 12, 3127. | 5.8 | 18 |
| 14 | Neural correlates of fat preference in frontotemporal dementia: translating insights from the obesity literature. Annals of Clinical and Translational Neurology, 2021, 8, 1318-1329. | 1.7 | 4 |
| 15 | Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. Genome Research, 2021, 31, 1994-2007. | 2.4 | 4 |
| 16 | Identification and functional modelling of plausibly causative cis-regulatory variants in a highly-selected cohort with X-linked intellectual disability. PLoS ONE, 2021, 16, e0256181. | 1.1 | 3 |
| 17 | Murine neuronatin deficiency is associated with a hypervariable food intake and bimodal obesity. Scientific Reports, 2021, 11, 17571. | 1.6 | 5 |
| 18 | The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627. | 5.8 | 33 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194. | 2.6 | 12 |
| 20 | Obesity-Associated GNAS Mutations and the Melanocortin Pathway. New England Journal of Medicine, 2021, 385, 1581-1592. | 13.9 | 44 |
| 21 | Predicting novel candidate human obesity genes and their site of action by systematic functional screening in Drosophila. PLoS Biology, 2021, 19, e3001255. | 2.6 | 7 |
| 22 | Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609. | 1.1 | 43 |
| 23 | Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762. | 13.7 | 343 |
| 24 | Efficacy and safety of setmelanotide, an MC4R agonist, in individuals with severe obesity due to LEPR or POMC deficiency: single-arm, open-label, multicentre, phase 3 trials. Lancet Diabetes and Endocrinology,the, 2020, 8, 960-970. | 5.5 | 235 |
| 25 | Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. PLoS Genetics, 2020, 16, e1008916. | 1.5 | 22 |
| 26 | KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261. | 3.7 | 57 |
| 27 | Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349. | 2.5 | 3 |
| 28 | Leptin-Mediated Changes in the Human Metabolome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2541-2552. | 1.8 | 20 |
| 29 | Exome Sequencing Identifies Genes and Gene Sets Contributing to Severe Childhood Obesity, Linking PHIP Variants to Repressed POMC Transcription. Cell Metabolism, 2020, 31, 1107-1119.e12. | 7.2 | 38 |
| 30 | Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. Scientific Reports, 2020, 10, 9028. | 1.6 | 40 |
| 31 | Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592. | 1.8 | 24 |
| 32 | GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888. | 1.1 | 22 |
| 33 | Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946. | 2.6 | 8 |
| 34 | Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630. | 5.8 | 43 |
| 35 | Neural networks associated with body composition in frontotemporal dementia. Annals of Clinical and Translational Neurology, 2019, 6, 1707-1717. | 1.7 | 10 |
| 36 | Hypothalamic Reproductive Endocrine Pulse Generator Activity Independent of Neurokinin B and Dynorphin Signaling. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4304-4318. | 1.8 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Genetic architecture of human thinness compared to severe obesity. PLoS Genetics, 2019, 15, e1007603. | 1.5 | 98 |
| 38 | Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611. | 1.1 | 14 |
| 39 | Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056. | 2.4 | 34 |
| 40 | Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373. | 5.8 | 86 |
| 41 | Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. Cell, 2019, 177, 597-607.e9. | 13.5 | 192 |
| 42 | NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452. | 1.5 | 28 |
| 43 | ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605. | 1.5 | 25 |
| 44 | Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. Nature Communications, 2019, 10, 1718. | 5.8 | 45 |
| 45 | Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. Diabetes, 2019, 68, 2049-2062. | 0.3 | 16 |
| 46 | Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. Cell, 2019, 176, 729-742.e18. | 13.5 | 80 |
| 47 | GDF15 Provides an Endocrine Signal of Nutritional Stress in Mice and Humans. Cell Metabolism, 2019, 29, 707-718.e8. | 7.2 | 286 |
| 48 | The genetic architecture of aniridia and Gillespie syndrome. Human Genetics, 2019, 138, 881-898. | 1.8 | 51 |
| 49 | NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. Pediatrics, 2018, 141, S485-S490. | 1.0 | 21 |
| 50 | Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. Lancet, The, 2018, 391, 1483-1492. | 6.3 | 63 |
| 51 | Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268. | 7.7 | 369 |
| 52 | BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Langeâ€“like syndrome. Nature Genetics, 2018, 50, 329-332. | 9.4 | 96 |
| 53 | A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. Cell Reports, 2018, 22, 3401-3408. | 2.9 | 81 |
| 54 | Potential role of gender specific effect of leptin receptor deficiency in an extended consanguineous family with severe early-onset obesity. European Journal of Medical Genetics, 2018, 61, 465-467. | 0.7 | 15 |

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|---|---|---|---|
| 55 | Cardiac Genetic Predisposition in SuddenÂInfant Death Syndrome. Journal of the American College of Cardiology, 2018, 71, 1217-1227. | 1.2 | 66 |
| 56 | Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. FASEB Journal, 2018, 32, 1830-1840. | 0.2 | 19 |
| 57 | Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164. | 6.0 | 158 |
| 58 | Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271. | 13.7 | 246 |
| 59 | ProxECAT: Proxy External Controls Association Test. A new case-control gene region association test using allele frequencies from public controls. PLoS Genetics, 2018, 14, e1007591. | 1.5 | 23 |
| 60 | Quantitative mass spectrometry for human melanocortin peptides inÂvitro and inÂvivo suggests prominent roles for Î²-MSH and desacetyl Î±-MSH in energy homeostasis. Molecular Metabolism, 2018, 17, 82-97. | 3.0 | 21 |
| 61 | MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. Nature Medicine, 2018, 24, 551-555. | 15.2 | 219 |
| 62 | Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41. | 9.4 | 286 |
| 63 | The orphan G protein-coupled receptor GPR139 is activated by the peptides: Adrenocorticotropic hormone (ACTH), Î±-, and Î²-melanocyte stimulating hormone (Î±-MSH, and Î²-MSH), and the conserved core motif HFRW. Neurochemistry International, 2017, 102, 105-113. | 1.9 | 36 |
| 64 | SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248. | 9.4 | 131 |
| 65 | Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908. | 1.1 | 46 |
| 66 | Genotypeâ€“phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. American Journal of Medical Genetics, Part A, 2017, 173, 1566-1574. | 0.7 | 20 |
| 67 | A recurrent de novo mutation inACTG1causes isolated ocular coloboma. Human Mutation, 2017, 38, 942-946. | 1.1 | 21 |
| 68 | Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. European Journal of Medical Genetics, 2017, 60, 130-135. | 0.7 | 47 |
| 69 | PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559. | 1.4 | 42 |
| 70 | De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658. | 2.6 | 56 |
| 71 | Resequencing at scale in neurodevelopmental disorders. Nature Genetics, 2017, 49, 488-489. | 9.4 | 3 |
| 72 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685. | 2.6 | 337 |

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|---|---|---|---|
| 73 | A Metabolomic Signature of Acute Caloric Restriction. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4486-4495. | 1.8 | 52 |
| 74 | Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. Molecular Metabolism, 2017, 6, 1321-1329. | 3.0 | 200 |
| 75 | Disruption of the homeodomain transcription factor orthopedia homeobox ( Otp ) is associated with obesity and anxiety. Molecular Metabolism, 2017, 6, 1419-1428. | 3.0 | 15 |
| 76 | A New Drug Target for Type 2 Diabetes. Cell, 2017, 170, 12-14. | 13.5 | 62 |
| 77 | Oxytocin administration suppresses hypothalamic activation in response to visual food cues. Scientific Reports, 2017, 7, 4266. | 1.6 | 28 |
| 78 | Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394. | 1.6 | 50 |
| 79 | The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45. | 9.4 | 251 |
| 80 | The RNA-binding landscape of RBM10 and its role in alternative splicing regulation in models of mouse early development. RNA Biology, 2017, 14, 45-57. | 1.5 | 41 |
| 81 | Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Brain, 2017, 140, 171-183. | 3.7 | 43 |
| 82 | Lipid Metabolism and Survival Across the Frontotemporal Dementia-Amyotrophic Lateral Sclerosis Spectrum: Relationships to Eating Behavior and Cognition. Journal of Alzheimer's Disease, 2017, 61, 773-783. | 1.2 | 47 |
| 83 | Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, e1.18-e1. | 0.9 | 0 |
| 84 | Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature. , 2017, 58, 594. | | 25 |
| 85 | Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15. | 0.9 | 24 |
| 86 | Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757. | 1.1 | 54 |
| 87 | A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992. | 2.6 | 81 |
| 88 | Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065. | 9.4 | 351 |
| 89 | Failure of sucrose replacement with the non-nutritive sweetener erythritol to alter GLP-1 or PYY release or test meal size in lean or obese people. Appetite, 2016, 107, 596-603. | 1.8 | 26 |
| 90 | Status dystonicus in two patients with SOX2â€anophthalmia syndrome and nonsense mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3048-3050. | 0.7 | 7 |

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|---|---|---|---|
| 91 | Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. Nature Communications, 2016, 7, 13055. | 5.8 | 46 |
| 92 | The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. Sleep, 2016, 39, 1691-1700. | 0.6 | 19 |
| 93 | Assessment of Eating Behavior Disturbance and Associated Neural Networks in Frontotemporal Dementia. JAMA Neurology, 2016, 73, 282. | 4.5 | 74 |
| 94 | Amyotrophic lateral sclerosis and frontotemporal dementia: distinct and overlapping changes in eating behaviour and metabolism. Lancet Neurology, The, 2016, 15, 332-342. | 4.9 | 120 |
| 95 | A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391. | 1.4 | 40 |
| 96 | Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. Open Biology, 2015, 5, 150047. | 1.5 | 46 |
| 97 | Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314. | 6.3 | 651 |
| 98 | Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431. | 1.4 | 37 |
| 99 | A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. European Journal of Medical Genetics, 2015, 58, 387-391. | 0.7 | 25 |
| 100 | The Hunger Genes: Pathways to Obesity. Cell, 2015, 161, 119-132. | 13.5 | 293 |
| 101 | Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369. | 9.4 | 133 |
| 102 | Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. Nature Communications, 2015, 6, 6904. | 5.8 | 31 |
| 103 | De novo, heterozygous, lossâ€ofâ€function mutations in SYNGAP1 cause a syndromic form of intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 2231-2237. | 0.7 | 96 |
| 104 | Severe Early-Onset Obesity Due to Bioinactive Leptin Caused by a p.N103K Mutation in the Leptin Gene. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 3227-3230. | 1.8 | 71 |
| 105 | The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90. | 13.7 | 1,014 |
| 106 | Diagnostically relevant facial gestalt information from ordinary photos. ELife, 2014, 3, e02020. | 2.8 | 129 |
| 107 | Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668. | 1.5 | 141 |
| 108 | Leptin Mediates the Increase in Blood Pressure Associated with Obesity. Cell, 2014, 159, 1404-1416. | 13.5 | 288 |

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|---|---|---|---|
| 109 | Defining the neural basis of appetite and obesity: from genes to behaviour. Clinical Medicine, 2014, 14, 286-289. | 0.8 | 39 |
| 110 | FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242. | 1.5 | 41 |
| 111 | Characterization of human variants in obesity-related SIM1 protein identifies a hot-spot for dimerization with the partner protein ARNT2. Biochemical Journal, 2014, 461, 403-412. | 1.7 | 10 |
| 112 | Wired for Obesity?. Diabetes, 2014, 63, 4016-4017. | 0.3 | 2 |
| 113 | Disruption of SATB2 or its long-range cis-regulation by SOX9 causes a syndromic form of Pierre Robin sequence. Human Molecular Genetics, 2014, 23, 2569-2579. | 1.4 | 51 |
| 114 | Genetic strategies to understand physiological pathways regulating body weight. Mammalian Genome, 2014, 25, 377-383. | 1.0 | 7 |
| 115 | Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996. | 1.4 | 15 |
| 116 | EJE PRIZE 2012: Obesity: from genes to behaviour. European Journal of Endocrinology, 2014, 171, R191-R195. | 1.9 | 5 |
| 117 | A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595. | 0.7 | 18 |
| 118 | 20 YEARS OF LEPTIN: Human disorders of leptin action. Journal of Endocrinology, 2014, 223, T63-T70. | 1.2 | 218 |
| 119 | Heterozygous Loss-of-Function Mutations in YAP1 Cause Both Isolated and Syndromic Optic Fissure Closure Defects. American Journal of Human Genetics, 2014, 94, 295-302. | 2.6 | 93 |
| 120 | The genetic architecture of microphthalmia, anophthalmia and coloboma. European Journal of Medical Genetics, 2014, 57, 369-380. | 0.7 | 213 |
| 121 | A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476. | 0.4 | 7 |
| 122 | Abstract 18543: Whole Exome Sequencing in Sudden Infant Death Syndrome Identifies a High Proportion of Putative Pathogenic and Functionally Significant Rare Variants Related to Inherited Cardiac Conditions. Circulation, 2014, 130, . | 1.6 | 0 |
| 123 | Neural and Behavioral Effects of a Novel Mu Opioid Receptor Antagonist in Binge-Eating Obese People. Biological Psychiatry, 2013, 73, 887-894. | 0.7 | 79 |
| 124 | KSR2 Mutations Are Associated with Obesity, Insulin Resistance, and Impaired Cellular Fuel Oxidation. Cell, 2013, 155, 765-777. | 13.5 | 154 |
| 125 | Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. Nature Genetics, 2013, 45, 513-517. | 9.4 | 278 |
| 126 | Postprandial Total Ghrelin Suppression Is Modulated by Melanocortin Signaling in Humans. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E288-E292. | 1.8 | 10 |

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|---|---|---|---|
| 127 | A Trans-Acting Protein Effect Causes Severe Eye Malformation in the Mp Mouse. PLoS Genetics, 2013, 9, e1003998. | 1.5 | 11 |
| 128 | Loss of Function of the Melanocortin 2 Receptor Accessory Protein 2 Is Associated with Mammalian Obesity. Science, 2013, 341, 275-278. | 6.0 | 225 |
| 129 | Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. Molecular Genetics & Genomic Medicine, 2013, 1, 15-31. | 0.6 | 79 |
| 130 | Rare variants in single-minded 1 (SIM1) are associated with severe obesity. Journal of Clinical Investigation, 2013, 123, 3042-3050. | 3.9 | 135 |
| 131 | Food addiction: is there a baby in the bathwater?. Nature Reviews Neuroscience, 2012, 13, 514-514. | 4.9 | 102 |
| 132 | A genome-wide association meta-analysis identifies new childhood obesity loci. Nature Genetics, 2012, 44, 526-531. | 9.4 | 352 |
| 133 | Human SH2B1 mutations are associated with maladaptive behaviors and obesity. Journal of Clinical Investigation, 2012, 122, 4732-4736. | 3.9 | 147 |
| 134 | FTO and Obesity: The Missing Link. Cell Metabolism, 2011, 13, 7-8. | 7.2 | 10 |
| 135 | Genetic, molecular and physiological insights into human obesity. European Journal of Clinical Investigation, 2011, 41, 451-455. | 1.7 | 42 |
| 136 | Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948. | 9.4 | 2,634 |
| 137 | Distinct Modulatory Effects of Satiety and Sibutramine on Brain Responses to Food Images in Humans: A Double Dissociation across Hypothalamus, Amygdala, and Ventral Striatum. Journal of Neuroscience, 2010, 30, 14346-14355. | 1.7 | 69 |
| 138 | Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670. | 13.7 | 487 |
| 139 | Leptin: a pivotal regulator of human energy homeostasis. American Journal of Clinical Nutrition, 2009, 89, 980S-984S. | 2.2 | 261 |
| 140 | Oral glutamine increases circulating glucagon-like peptide 1, glucagon, and insulin concentrations in lean, obese, and type 2 diabetic subjects. American Journal of Clinical Nutrition, 2009, 89, 106-113. | 2.2 | 201 |
| 141 | Obesity Genesâ€"It's All About the Parents!. Cell Metabolism, 2009, 9, 487-488. | 7.2 | 10 |
| 142 | Modulation of Blood Pressure by Central Melanocortinergic Pathways. New England Journal of Medicine, 2009, 360, 44-52. | 13.9 | 412 |
| 143 | Mutations in ligands and receptors of the leptinâ€"melanocortin pathway that lead to obesity. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 569-577. | 2.9 | 225 |
| 144 | Monogenic Human Obesity. , 2008, 36, 1-11. | | 84 |

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|---|---|---|---|
| 145 | Human Obesity: A Heritable Neurobehavioral Disorder That Is Highly Sensitive to Environmental Conditions. Diabetes, 2008, 57, 2905-2910. | 0.3 | 160 |
| 146 | Prevalence of Melanocortin-4 Receptor Deficiency in Europeans and Their Age-Dependent Penetrance in Multigenerational Pedigrees. Diabetes, 2008, 57, 2511-2518. | 0.3 | 229 |
| 147 | Leptin Regulates Peripheral Lipid Metabolism Primarily through Central Effects on Food Intake. Endocrinology, 2008, 149, 5432-5439. | 1.4 | 78 |
| 148 | Melanocortin Receptors as Targets in the Treatment of Obesity. Current Topics in Medicinal Chemistry, 2007, 7, 1098-1110. | 1.0 | 47 |
| 149 | Hyperphagia and Early-Onset Obesity due to a Novel Homozygous Missense Mutation in Prohormone Convertase 1/3. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3369-3373. | 1.8 | 196 |
| 150 | Clinical and Molecular Genetic Spectrum of Congenital Deficiency of the Leptin Receptor. New England Journal of Medicine, 2007, 356, 237-247. | 13.9 | 610 |
| 151 | Insights from the Genetics of Severe Childhood Obesity. Hormone Research in Paediatrics, 2007, 68, 5-7. | 0.8 | 22 |
| 152 | Leptin Regulates Striatal Regions and Human Eating Behavior. Science, 2007, 317, 1355-1355. | 6.0 | 541 |
| 153 | Treating obesity: Does antagonism of NPY fit the bill?. Cell Metabolism, 2006, 4, 260-262. | 7.2 | 7 |
| 154 | The severely obese patientâ€"a genetic work-up. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 172-177. | 2.9 | 24 |
| 155 | Genetics of Obesity in Humans. Endocrine Reviews, 2006, 27, 710-718. | 8.9 | 452 |
| 156 | Heterozygosity for aPOMC-Null Mutation and Increased Obesity Risk in Humans. Diabetes, 2006, 55, 2549-2553. | 0.3 | 205 |
| 157 | Genetic aspects of severe childhood obesity. Pediatric Endocrinology Reviews, 2006, 3 Suppl 4, 528-36. | 1.2 | 10 |
| 158 | Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. Trends in Genetics, 2005, 21, 249-253. | 2.9 | 77 |
| 159 | Developmental eye disorders. Current Opinion in Genetics and Development, 2005, 15, 348-353. | 1.5 | 95 |
| 160 | Genetic and hereditary aspects of childhood obesity. Best Practice and Research in Clinical Endocrinology and Metabolism, 2005, 19, 359-374. | 2.2 | 94 |
| 161 | Monogenic Obesity in Humans. Annual Review of Medicine, 2005, 56, 443-458. | 5.0 | 367 |
| 162 | Candidate Genes for Obesity â€" How Might They Interact with Environment and Diet ?. , 2005, 569, 33-34. | | 2 |

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| 163 | Clinical Spectrum of Obesity and Mutations in the Melanocortin 4 Receptor Gene. New England Journal of Medicine, 2003, 348, 1085-1095. | 13.9 | 1,475 |
| 164 | Identification of SATB2 as the cleft palate gene on 2q32-q33. Human Molecular Genetics, 2003, 12, 2491-2501. | 1.4 | 248 |
| 165 | Transcriptome analysis of human autosomal trisomy. Human Molecular Genetics, 2002, 11, 3249-3256. | 1.4 | 150 |
| 166 | Leptin and the Onset of Puberty: Insights from Rodent and Human Genetics. Seminars in Reproductive Medicine, 2002, 20, 139-144. | 0.5 | 117 |
| 167 | Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. Journal of Clinical Investigation, 2002, 110, 1093-1103. | 3.9 | 953 |
| 168 | Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. Journal of Clinical Investigation, 2002, 110, 1093-1103. | 3.9 | 670 |
| 169 | Partial leptin deficiency and human adiposity. Nature, 2001, 414, 34-35. | 13.7 | 356 |
| 170 | Genetics of body-weight regulation. Nature, 2000, 404, 644-651. | 13.7 | 682 |
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