# Sadaf Farooqi

## List of Publications by Year in Descending Order



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|---|---|---|---|
| 181 papers | 23,128 citations | 64 h-index | 151 g-index |
| 194 ext. papers | 26,997 ext. citations | 15.6 avg, IF | 6.55 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 181 | Central obesity is selectively associated with cerebral gray matter atrophy in 15,634 subjects in the UK Biobank.. *International Journal of Obesity*, **2022**, | 5.5 | 1 |
| 180 | Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors.. *Biological Psychiatry*, **2022**, | 7.9 | 3 |
| 179 | Visualization of sympathetic neural innervation in human white adipose tissue.. *Open Biology*, **2022**, 12, 210345 | 7 | 0 |
| 178 | Genes and Obesity **2022**, 47-57 | | |
| 177 | The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**, | 8.1 | 4 |
| 176 | Predicting novel candidate human obesity genes and their site of action by systematic functional screening in Drosophila. *PLoS Biology*, **2021**, 19, e3001255 | 9.7 | 0 |
| 175 | Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. *Clinical Endocrinology*, **2021**, | 3.4 | 1 |
| 174 | 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 | 11 | 0 |
| 173 | Obesity-Associated Mutations and the Melanocortin Pathway. *New England Journal of Medicine*, **2021**, 385, 1581-1592 | 59.2 | 2 |
| 172 | TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. *Human Mutation*, **2021**, 42, 445-459 | 4.7 | 4 |
| 171 | Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. *Cell Reports*, **2021**, 34, 108862 | 10.6 | 12 |
| 170 | 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 | 56.2 | 1 |
| 169 | Putting a brake on hunger. *Science*, **2021**, 372, 792-793 | 33.3 | |
| 168 | Cornelia de Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. *Nature Communications*, **2021**, 12, 3127 | 17.4 | 6 |
| 167 | Neural correlates of fat preference in frontotemporal dementia: translating insights from the obesity literature. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1318-1329 | 5.3 | 1 |
| 166 | Evaluating variants classified as pathogenic in ClinVar in the DDD Study. *Genetics in Medicine*, **2021**, 23, 571-575 | 8.1 | 6 |
| 165 | Monogenic human obesity syndromes. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2021**, 181, 301-310 | 3 | 1 |

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| 164 | Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. *Genome Research*, **2021**, 31, 1994-2007 | 9.7 | 2 |
| 163 | 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 | 3.7 | |
| 162 | Murine neuronatin deficiency is associated with a hypervariable food intake and bimodal obesity. *Scientific Reports*, **2021**, 11, 17571 | 4.9 | 1 |
| 161 | The contribution of X-linked coding variation to severe developmental disorders. *Nature Communications*, **2021**, 12, 627 | 17.4 | 5 |
| 160 | KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. *Brain*, **2020**, 143, 3242-3261 | 11.2 | 19 |
| 159 | Genomically Aided Diagnosis of Severe Developmental Disorders. *Annual Review of Genomics and Human Genetics*, **2020**, 21, 327-349 | 9.7 | 0 |
| 158 | Leptin-Mediated Changes in the Human Metabolome. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105, | 5.6 | 9 |
| 157 | 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 | 24.6 | 16 |
| 156 | Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. *Scientific Reports*, **2020**, 10, 9028 | 4.9 | 11 |
| 155 | Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. *Human Genetics*, **2020**, 139, 575-592 | 6.3 | 11 |
| 154 | GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. *Genetics in Medicine*, **2020**, 22, 878-888 | 8.1 | 9 |
| 153 | Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNA-protein interaction. *Genetics in Medicine*, **2020**, 22, 598-609 | 8.1 | 19 |
| 152 | Evidence for 28 genetic disorders discovered by combining healthcare and research data. *Nature*, **2020**, 586, 757-762 | 50.4 | 103 |
| 151 | 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 | 18.1 | 76 |
| 150 | Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. *PLoS Genetics*, **2020**, 16, e1008916 | 6 | 7 |
| 149 | Hypothalamic Reproductive Endocrine Pulse Generator Activity Independent of Neurokinin B and Dynorphin Signaling. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 4304-4318 | 5.6 | 17 |
| 148 | Genetic architecture of human thinness compared to severe obesity. *PLoS Genetics*, **2019**, 15, e1007603 | 6 | 51 |
| 147 | Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611 | 4.5 | 7 |

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| 146 | Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. *Genome Research*, **2019**, 29, 1047-1056 | 9.7 | 18 |
| 145 | Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. *Nature Communications*, **2019**, 10, 2373 | 17.4 | 22 |
| 144 | Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. *Cell*, **2019**, 177, 597-607.e9 | 56.2 | 113 |
| 143 | polyadenylation signal variants cause syndromic microphthalmia. *Journal of Medical Genetics*, **2019**, 56, 444-452 | 5.8 | 15 |
| 142 | ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. *PLoS Genetics*, **2019**, 15, e1007605 | 6 | 10 |
| 141 | Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. *Nature Communications*, **2019**, 10, 1718 | 17.4 | 20 |
| 140 | Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. *American Journal of Human Genetics*, **2019**, 105, 933-946 | 11 | 4 |
| 139 | Contribution of retrotransposition to developmental disorders. *Nature Communications*, **2019**, 10, 4630 | 17.4 | 25 |
| 138 | Genetic Obesity Syndromes **2019**, 729-736 | | 0 |
| 137 | Neural networks associated with body composition in frontotemporal dementia. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1707-1717 | 5.3 | 7 |
| 136 | Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. *Diabetes*, **2019**, 68, 2049-2062 | 0.9 | 6 |
| 135 | Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. *Cell*, **2019**, 176, 729-742.e18 | 56.2 | 38 |
| 134 | GDF15 Provides an Endocrine Signal of Nutritional Stress in Mice and Humans. *Cell Metabolism*, **2019**, 29, 707-718.e8 | 24.6 | 153 |
| 133 | The genetic architecture of aniridia and Gillespie syndrome. *Human Genetics*, **2019**, 138, 881-898 | 6.3 | 26 |
| 132 | NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. *Pediatrics*, **2018**, 141, S485-S490 | 7.4 | 14 |
| 131 | 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 | 40 | 49 |
| 130 | Paediatric genomics: diagnosing rare disease in children. *Nature Reviews Genetics*, **2018**, 19, 253-268 | 30.1 | 201 |
| 129 | BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Lange-like syndrome. *Nature Genetics*, **2018**, 50, 329-332 | 36.3 | 52 |

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|---|---|---|---|
| 128 | A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. *Cell Reports*, **2018**, 22, 3401-3408 | 10.6 | 51 |
| 127 | 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 | 2.6 | 8 |
| 126 | Cardiac Genetic Predisposition in Sudden Infant Death Syndrome. *Journal of the American College of Cardiology*, **2018**, 71, 1217-1227 | 15.1 | 43 |
| 125 | MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. *Nature Medicine*, **2018**, 24, 551-555 | 50.5 | 139 |
| 124 | Lipid Metabolism and Survival Across the Frontotemporal Dementia-Amyotrophic Lateral Sclerosis Spectrum: Relationships to Eating Behavior and Cognition. *Journal of Alzheimer's Disease*, **2018**, 61, 773-783 | 4.3 | 35 |
| 123 | Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41 | 36.3 | 186 |
| 122 | Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. *FASEB Journal*, **2018**, 32, 1830-1840 | 0.9 | 10 |
| 121 | Quantifying the contribution of recessive coding variation to developmental disorders. *Science*, **2018**, 362, 1161-1164 | 33.3 | 83 |
| 120 | Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. *Nature*, **2018**, 562, 268-271 | 50.4 | 149 |
| 119 | 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 | 6 | 13 |
| 118 | Quantitative mass spectrometry for human melanocortin peptides in vitro and in vivo suggests prominent roles for MSH and desacetyl MSH in energy homeostasis. *Molecular Metabolism*, **2018**, 17, 82-97 | 8.8 | 13 |
| 117 | The orphan G protein-coupled receptor GPR139 is activated by the peptides: Adrenocorticotropic hormone (ACTH), and melanocyte stimulating hormone (MSH, and MSH), and the conserved core motif HFRW. *Neurochemistry International*, **2017**, 102, 105-113 | 4.4 | 29 |
| 116 | SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. *Nature Genetics*, **2017**, 49, 238-248 | 36.3 | 88 |
| 115 | Clinical and molecular consequences of disease-associated de novo mutations in SATB2. *Genetics in Medicine*, **2017**, 19, 900-908 | 8.1 | 30 |
| 114 | Genotype-phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1566-1574 | 2.5 | 16 |
| 113 | A recurrent de novo mutation in ACTG1 causes isolated ocular coloboma. *Human Mutation*, **2017**, 38, 942-946 | 4.7 | 16 |
| 112 | Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. *European Journal of Medical Genetics*, **2017**, 60, 130-135 | 2.6 | 25 |
| 111 | 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 | 5.3 | 24 |

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| 110 | 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 | 11 | 36 |
| 109 | Resequencing at scale in neurodevelopmental disorders. *Nature Genetics*, **2017**, 49, 488-489 | 36.3 | 2 |
| 108 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685 | 11 | 214 |
| 107 | A Metabolomic Signature of Acute Caloric Restriction. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 4486-4495 | 5.6 | 28 |
| 106 | Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature **2017**, 58, 594-603 | | 12 |
| 105 | Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. *Molecular Metabolism*, **2017**, 6, 1321-1329 | 8.8 | 121 |
| 104 | Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. *Molecular Metabolism*, **2017**, 6, 1419-1428 | 8.8 | 7 |
| 103 | A New Drug Target for Type 2 Diabetes. *Cell*, **2017**, 170, 12-14 | 56.2 | 20 |
| 102 | Oxytocin administration suppresses hypothalamic activation in response to visual food cues. *Scientific Reports*, **2017**, 7, 4266 | 4.9 | 23 |
| 101 | Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. *Scientific Reports*, **2017**, 7, 4394 | 4.9 | 31 |
| 100 | The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45 | 36.3 | 172 |
| 99 | 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 | 4.8 | 30 |
| 98 | Energy expenditure in frontotemporal dementia: a behavioural and imaging study. *Brain*, **2017**, 140, 171-183 | 11.2 | 28 |
| 97 | Energy expenditure in frontotemporal dementia: a behavioural and imaging study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, e1.18-e1 | 5.5 | |
| 96 | Returning genome sequences to research participants: Policy and practice. *Wellcome Open Research*, **2017**, 2, 15 | 4.8 | 10 |
| 95 | 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 | 4.5 | 19 |
| 94 | Status dystonicus in two patients with SOX2-anophthalmia syndrome and nonsense mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3048-3050 | 2.5 | 6 |
| 93 | Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. *Nature Communications*, **2016**, 7, 13055 | 17.4 | 31 |

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| 92 | The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. *Sleep*, **2016**, 39, 1691-700 | 1.1 | 13 |
| 91 | Assessment of Eating Behavior Disturbance and Associated Neural Networks in Frontotemporal Dementia. *JAMA Neurology*, **2016**, 73, 282-90 | 17.2 | 56 |
| 90 | Amyotrophic lateral sclerosis and frontotemporal dementia: distinct and overlapping changes in eating behaviour and metabolism. *Lancet Neurology, The*, **2016**, 15, 332-42 | 24.1 | 88 |
| 89 | A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. *Human Molecular Genetics*, **2016**, 25, 1382-91 | 5.6 | 30 |
| 88 | Genetic Analysis of ‘PAX6-Negative’ Individuals with Aniridia or Gillespie Syndrome. *PLoS ONE*, **2016**, 11, e0153757 | 3.7 | 34 |
| 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 | 11 | 53 |
| 86 | Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. *Nature Genetics*, **2016**, 48, 1060-5 | 36.3 | 200 |
| 85 | A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. *European Journal of Medical Genetics*, **2015**, 58, 387-91 | 2.6 | 17 |
| 84 | The hunger genes: pathways to obesity. *Cell*, **2015**, 161, 119-132 | 56.2 | 216 |
| 83 | Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. *Nature Genetics*, **2015**, 47, 1363-9 | 36.3 | 91 |
| 82 | Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. *Nature Communications*, **2015**, 6, 6904 | 17.4 | 20 |
| 81 | De novo, heterozygous, loss-of-function mutations in SYNGAP1 cause a syndromic form of intellectual disability. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2231-7 | 2.5 | 59 |
| 80 | 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, 3227-30 | 5.6 | 44 |
| 79 | The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90 | 50.4 | 776 |
| 78 | Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. *Open Biology*, **2015**, 5, 150047 | 7 | 26 |
| 77 | Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. *Lancet, The*, **2015**, 385, 1305-14 | 40 | 451 |
| 76 | Clinical utility gene card for: Cornelia de Lange syndrome. *European Journal of Human Genetics*, **2015**, 23, | 5.3 | 28 |
| 75 | A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. *European Journal of Medical Genetics*, **2014**, 57, 587-95 | 2.6 | 14 |

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| 74 | 20 years of leptin: human disorders of leptin action. *Journal of Endocrinology*, **2014**, 223, T63-70 | 4.7 | 163 |
| 73 | 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 | 11 | 74 |
| 72 | The genetic architecture of microphthalmia, anophthalmia and coloboma. *European Journal of Medical Genetics*, **2014**, 57, 369-80 | 2.6 | 168 |
| 71 | A novel mutation in the leptin gene (W121X) in an Egyptian family. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 474-476 | 1.8 | 5 |
| 70 | Diagnostically relevant facial gestalt information from ordinary photos. *ELife*, **2014**, 3, e02020 | 8.9 | 81 |
| 69 | 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, 659-68 | 5.8 | 111 |
| 68 | Leptin mediates the increase in blood pressure associated with obesity. *Cell*, **2014**, 159, 1404-16 | 56.2 | 232 |
| 67 | Defining the neural basis of appetite and obesity: from genes to behaviour. *Clinical Medicine*, **2014**, 14, 286-9 | 1.9 | 27 |
| 66 | FRA2A is a CGG repeat expansion associated with silencing of AFF3. *PLoS Genetics*, **2014**, 10, e1004242 | 6 | 26 |
| 65 | Characterization of human variants in obesity-related SIM1 protein identifies a hot-spot for dimerization with the partner protein ARNT2. *Biochemical Journal*, **2014**, 461, 403-12 | 3.8 | 7 |
| 64 | Wired for obesity?. *Diabetes*, **2014**, 63, 4016-7 | 0.9 | 2 |
| 63 | Disruption of SATB2 or its long-range cis-regulation by SOX9 causes a syndromic form of Pierre Robin sequence. *Human Molecular Genetics*, **2014**, 23, 2569-79 | 5.6 | 42 |
| 62 | Genetic strategies to understand physiological pathways regulating body weight. *Mammalian Genome*, **2014**, 25, 377-83 | 3.2 | 7 |
| 61 | Expansion of ocular phenotypic features associated with mutations in ADAMTS18. *JAMA Ophthalmology*, **2014**, 132, 996-1001 | 3.9 | 8 |
| 60 | EJE Prize 2012: Obesity: from genes to behaviour. *European Journal of Endocrinology*, **2014**, 171, R191-5 | 6.5 | 3 |
| 59 | Neural and behavioral effects of a novel mu opioid receptor antagonist in binge-eating obese people. *Biological Psychiatry*, **2013**, 73, 887-94 | 7.9 | 75 |
| 58 | KSR2 mutations are associated with obesity, insulin resistance, and impaired cellular fuel oxidation. *Cell*, **2013**, 155, 765-77 | 56.2 | 113 |
| 57 | Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. *Nature Genetics*, **2013**, 45, 513-7 | 36.3 | 231 |

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| 56 | Postprandial total ghrelin suppression is modulated by melanocortin signaling in humans. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E288-92 | 5.6 | 10 |
| 55 | A trans-acting protein effect causes severe eye malformation in the Mp mouse. *PLoS Genetics*, **2013**, 9, e1003998 | 6 | 7 |
| 54 | Loss of function of the melanocortin 2 receptor accessory protein 2 is associated with mammalian obesity. *Science*, **2013**, 341, 275-8 | 33.3 | 179 |
| 53 | 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 | 2.3 | 69 |
| 52 | Rare variants in single-minded 1 (SIM1) are associated with severe obesity. *Journal of Clinical Investigation*, **2013**, 123, 3042-50 | 15.9 | 107 |
| 51 | Food addiction: is there a baby in the bathwater?. *Nature Reviews Neuroscience*, **2012**, 13, 514-514 | 13.5 | 88 |
| 50 | A genome-wide association meta-analysis identifies new childhood obesity loci. *Nature Genetics*, **2012**, 44, 526-31 | 36.3 | 292 |
| 49 | Human SH2B1 mutations are associated with maladaptive behaviors and obesity. *Journal of Clinical Investigation*, **2012**, 122, 4732-6 | 15.9 | 103 |
| 48 | FTO and obesity: the missing link. *Cell Metabolism*, **2011**, 13, 7-8 | 24.6 | 7 |
| 47 | Genetic, molecular and physiological insights into human obesity. *European Journal of Clinical Investigation*, **2011**, 41, 451-5 | 4.6 | 36 |
| 46 | Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48 | 36.3 | 2267 |
| 45 | 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, 14346-55 | 6.6 | 61 |
| 44 | Large, rare chromosomal deletions associated with severe early-onset obesity. *Nature*, **2010**, 463, 666-70 | 50.4 | 417 |
| 43 | Leptin: a pivotal regulator of human energy homeostasis. *American Journal of Clinical Nutrition*, **2009**, 89, 980S-984S | 7 | 226 |
| 42 | 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 | 7 | 171 |
| 41 | Obesity genes-it's all about the parents!. *Cell Metabolism*, **2009**, 9, 487-8 | 24.6 | 9 |
| 40 | Modulation of blood pressure by central melanocortinergic pathways. *New England Journal of Medicine*, **2009**, 360, 44-52 | 59.2 | 358 |
| 39 | Mutations in ligands and receptors of the leptin-melanocortin pathway that lead to obesity. *Nature Clinical Practice Endocrinology and Metabolism*, **2008**, 4, 569-77 | | 184 |

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|---|---|---|---|
| 38 | Monogenic human obesity. *Frontiers of Hormone Research*, **2008**, 36, 1-11 | 3.5 | 73 |
| 37 | Human obesity: a heritable neurobehavioral disorder that is highly sensitive to environmental conditions. *Diabetes*, **2008**, 57, 2905-10 | 0.9 | 129 |
| 36 | Prevalence of melanocortin-4 receptor deficiency in Europeans and their age-dependent penetrance in multigenerational pedigrees. *Diabetes*, **2008**, 57, 2511-8 | 0.9 | 198 |
| 35 | Leptin regulates peripheral lipid metabolism primarily through central effects on food intake. *Endocrinology*, **2008**, 149, 5432-9 | 4.8 | 71 |
| 34 | Melanocortin Receptors as Targets in the Treatment of Obesity. *Current Topics in Medicinal Chemistry*, **2007**, 7, 1098-1110 | 3 | 30 |
| 33 | 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, 3369-73 | 5.6 | 166 |
| 32 | Clinical and molecular genetic spectrum of congenital deficiency of the leptin receptor. *New England Journal of Medicine*, **2007**, 356, 237-47 | 59.2 | 503 |
| 31 | Insights from the genetics of severe childhood obesity. *Hormone Research in Paediatrics*, **2007**, 68 Suppl 5, 5-7 | 3.3 | 18 |
| 30 | Leptin regulates striatal regions and human eating behavior. *Science*, **2007**, 317, 1355 | 33.3 | 452 |
| 29 | The severely obese patient--a genetic work-up. *Nature Clinical Practice Endocrinology and Metabolism*, **2006**, 2, 172-7; quiz following 177 | | 20 |
| 28 | Genetics of obesity in humans. *Endocrine Reviews*, **2006**, 27, 710-18 | 27.2 | 376 |
| 27 | Heterozygosity for a POMC-null mutation and increased obesity risk in humans. *Diabetes*, **2006**, 55, 2549-53 | 6.3 | 176 |
| 26 | Treating obesity: does antagonism of NPY fit the bill?. *Cell Metabolism*, **2006**, 4, 260-2 | 24.6 | 5 |
| 25 | Genetic aspects of severe childhood obesity. *Pediatric Endocrinology Reviews*, **2006**, 3 Suppl 4, 528-36 | 1.1 | 10 |
| 24 | Developmental eye disorders. *Current Opinion in Genetics and Development*, **2005**, 15, 348-53 | 4.9 | 80 |
| 23 | Genetic and hereditary aspects of childhood obesity. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2005**, 19, 359-74 | 6.5 | 57 |
| 22 | Monogenic obesity in humans. *Annual Review of Medicine*, **2005**, 56, 443-58 | 17.4 | 308 |
| 21 | Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. *Trends in Genetics*, **2005**, 21, 249-53 | 8.5 | 65 |

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|---|---|---|---|
| 20 | Candidate genes for obesity--how might they interact with environment and diet?. *Advances in Experimental Medicine and Biology*, **2005**, 569, 33-4 | 3.6 | 2 |
| 19 | Clinical spectrum of obesity and mutations in the melanocortin 4 receptor gene. *New England Journal of Medicine*, **2003**, 348, 1085-95 | 59.2 | 1227 |
| 18 | Identification of SATB2 as the cleft palate gene on 2q32-q33. *Human Molecular Genetics*, **2003**, 12, 2491-501 | 5.6 | 209 |
| 17 | Transcriptome analysis of human autosomal trisomy. *Human Molecular Genetics*, **2002**, 11, 3249-56 | 5.6 | 127 |
| 16 | Leptin and the onset of puberty: insights from rodent and human genetics. *Seminars in Reproductive Medicine*, **2002**, 20, 139-44 | 1.4 | 95 |
| 15 | 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 | 15.9 | 845 |
| 14 | Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. *Journal of Clinical Investigation*, **2002**, 110, 1093-103 | 15.9 | 441 |
| 13 | Partial leptin deficiency and human adiposity. *Nature*, **2001**, 414, 34-5 | 50.4 | 305 |
| 12 | Genetics of body-weight regulation. *Nature*, **2000**, 404, 644-51 | 50.4 | 593 |
| 11 | Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. *Journal of Clinical Investigation*, **2000**, 106, 271-9 | 15.9 | 589 |
| 10 | The therapeutic value of somatostatin and its analogues. *Pituitary*, **1999**, 2, 79-88 | 4.3 | 7 |
| 9 | Carey-Fineman-Ziter (CFZ) syndrome: report on affected sibs. *American Journal of Medical Genetics Part A*, **1999**, 82, 110-3 | | 10 |
| 8 | A locus for isolated cleft palate, located on human chromosome 2q32. *American Journal of Human Genetics*, **1999**, 65, 387-96 | 11 | 60 |
| 7 | Effects of recombinant leptin therapy in a child with congenital leptin deficiency. *New England Journal of Medicine*, **1999**, 341, 879-84 | 59.2 | 1485 |
| 6 | A frameshift mutation in MC4R associated with dominantly inherited human obesity. *Nature Genetics*, **1998**, 20, 111-2 | 36.3 | 893 |
| 5 | ob gene mutations and human obesity. *Proceedings of the Nutrition Society*, **1998**, 57, 471-5 | 2.9 | 35 |
| 4 | Congenital leptin deficiency is associated with severe early-onset obesity in humans. *Nature*, **1997**, 387, 903-8 | 50.4 | 2309 |
| 3 | Ambulatory blood pressure monitoring in acute stroke. The West Birmingham Stroke Project. *Stroke*, **1997**, 28, 31-5 | 6.7 | 39 |