I Sadaf Farooqi

List of Publications by Year
in descending order

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		8755	14208
133	34,484	75	128
papers	citations	h-index	g-index

136	136	136	28872
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Congenital leptin deficiency is associated with severe early-onset obesity in humans. Nature, 1997, 387, 903-908.	27.8	2,664
2	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	21.4	2,634
3	Effects of Recombinant Leptin Therapy in a Child with Congenital Leptin Deficiency. New England Journal of Medicine, 1999, 341, 879-884.	27.0	1,760
4	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	21.4	1,572
5	Clinical Spectrum of Obesity and Mutations in the Melanocortin 4 Receptor Gene. New England Journal of Medicine, 2003, 348, 1085-1095.	27.0	1,475
6	The Obesity-AssociatedFTOGene Encodes a 2-Oxoglutarate-Dependent Nucleic Acid Demethylase. Science, 2007, 318, 1469-1472.	12.6	1,305
7	Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nature Genetics, 2008, 40, 768-775.	21.4	1,179
8	A frameshift mutation in MC4R associated with dominantly inherited human obesity. Nature Genetics, 1998, 20, 111-112.	21.4	1,026
9	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
10	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.	8.2	953
11	Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. Journal of Clinical Investigation, 2000, 106, 271-279.	8.2	696
12	Genetics of body-weight regulation. Nature, 2000, 404, 644-651.	27.8	682
13	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.	8.2	670
14	Clinical and Molecular Genetic Spectrum of Congenital Deficiency of the Leptin Receptor. New England Journal of Medicine, 2007, 356, 237-247.	27.0	610
15	Leptin Regulates Striatal Regions and Human Eating Behavior. Science, 2007, 317, 1355-1355.	12.6	541
16	The Hormonal Control of Food Intake. Cell, 2007, 129, 251-262.	28.9	508
17	A de novo mutation affecting human TrkB associated with severe obesity and developmental delay. Nature Neuroscience, 2004, 7, 1187-1189.	14.8	499
18	Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670.	27.8	487

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19	Genetics of Obesity in Humans. Endocrine Reviews, 2006, 27, 710-718.	20.1	452
20	Obesity Associated Genetic Variation inFTOIs Associated with Diminished Satiety. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3640-3643.	3.6	443
21	Hyperphagia, Severe Obesity, Impaired Cognitive Function, and Hyperactivity Associated With Functional Loss of One Copy of the Brain-Derived Neurotrophic Factor (BDNF) Gene. Diabetes, 2006, 55, 3366-3371.	0.6	421
22	Modulation of Blood Pressure by Central Melanocortinergic Pathways. New England Journal of Medicine, 2009, 360, 44-52.	27.0	412
23	Obesity and the brain: how convincing is the addiction model?. Nature Reviews Neuroscience, 2012, 13, 279-286.	10.2	409
24	Monogenic Obesity in Humans. Annual Review of Medicine, 2005, 56, 443-458.	12.2	367
25	Partial leptin deficiency and human adiposity. Nature, 2001, 414, 34-35.	27.8	356
26	Serum Ghrelin Levels Are Inversely Correlated with Body Mass Index, Age, and Insulin Concentrations in Normal Children and Are Markedly Increased in Prader-Willi Syndrome. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 174-178.	3.6	356
27	A genome-wide association meta-analysis identifies new childhood obesity loci. Nature Genetics, 2012, 44, 526-531.	21.4	352
28	The central melanocortin system directly controls peripheral lipid metabolism. Journal of Clinical Investigation, 2007, 117, 3475-3488.	8.2	341
29	Loss-of-Function Mutation in the Dioxygenase-Encoding FTO Gene Causes Severe Growth Retardation and Multiple Malformations. American Journal of Human Genetics, 2009, 85, 106-111.	6.2	340
30	The Hunger Genes: Pathways to Obesity. Cell, 2015, 161, 119-132.	28.9	293
31	Leptin Mediates the Increase in Blood Pressure Associated with Obesity. Cell, 2014, 159, 1404-1416.	28.9	288
32	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	21.4	286
33	Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. Nature Genetics, 2013, 45, 513-517.	21.4	278
34	Small-intestinal dysfunction accompanies the complex endocrinopathy of human proprotein convertase 1 deficiency. Journal of Clinical Investigation, 2003, 112, 1550-1560.	8.2	276
35	Leptin: a pivotal regulator of human energy homeostasis. American Journal of Clinical Nutrition, 2009, 89, 980S-984S.	4.7	261
36	A deletion of the HBII-85 class of small nucleolar RNAs (snoRNAs) is associated with hyperphagia, obesity and hypogonadism. Human Molecular Genetics, 2009, 18, 3257-3265.	2.9	253

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37	Antidiabetic Effects of IGFBP2, a Leptin-Regulated Gene. Cell Metabolism, 2010, 11, 11-22.	16.2	251
38	A missense mutation disrupting a dibasic prohormone processing site in pro-opiomelanocortin (POMC) increases susceptibility to early-onset obesity through a novel molecular mechanism. Human Molecular Genetics, 2002, 11, 1997-2004.	2.9	249
39	Congenital Leptin Deficiency Due to Homozygosity for the Î”133G Mutation: Report of Another Case and Evaluation of Response to Four Years of Leptin Therapy. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 4821-4826.	3.6	245
40	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.	11.4	235
41	Prevalence of Melanocortin-4 Receptor Deficiency in Europeans and Their Age-Dependent Penetrance in Multigenerational Pedigrees. Diabetes, 2008, 57, 2511-2518.	0.6	229
42	Mutations in ligands and receptors of the leptinâ€“melanocortin pathway that lead to obesity. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 569-577.	2.8	225
43	Loss of Function of the Melanocortin 2 Receptor Accessory Protein 2 Is Associated with Mammalian Obesity. Science, 2013, 341, 275-278.	12.6	225
44	MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. Nature Medicine, 2018, 24, 551-555.	30.7	219
45	20 YEARS OF LEPTIN: Human disorders of leptin action. Journal of Endocrinology, 2014, 223, T63-T70.	2.6	218
46	A POMC variant implicates Î²-melanocyte-stimulating hormone in the control of human energy balance. Cell Metabolism, 2006, 3, 135-140.	16.2	207
47	Heterozygosity for a POMC-Null Mutation and Increased Obesity Risk in Humans. Diabetes, 2006, 55, 2549-2553.	0.6	205
48	Mutations in the human melanocortin-4 receptor gene associated with severe familial obesity disrupts receptor function through multiple molecular mechanisms. Human Molecular Genetics, 2003, 12, 561-574.	2.9	201
49	Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. Molecular Metabolism, 2017, 6, 1321-1329.	6.5	200
50	Proopiomelanocortin and Energy Balance: Insights from Human and Murine Genetics. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 2557-2562.	3.6	197
51	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.	3.6	196
52	Minireview: Human Obesityâ€”Lessons from Monogenic Disorders. Endocrinology, 2003, 144, 3757-3764.	2.8	194
53	Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. Cell, 2019, 177, 597-607.e9.	28.9	192
54	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Presenting in Childhood. Pediatrics, 2007, 120, e179-e188.	2.1	175

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55	Genetic approaches to understanding human obesity. Journal of Clinical Investigation, 2011, 121, 2080-2086.	8.2	161
56	Human Obesity: A Heritable Neurobehavioral Disorder That Is Highly Sensitive to Environmental Conditions. Diabetes, 2008, 57, 2905-2910.	0.6	160
57	KSR2 Mutations Are Associated with Obesity, Insulin Resistance, and Impaired Cellular Fuel Oxidation. Cell, 2013, 155, 765-777.	28.9	154
58	Human SH2B1 mutations are associated with maladaptive behaviors and obesity. Journal of Clinical Investigation, 2012, 122, 4732-4736.	8.2	147
59	Melanocortin-4 Receptor Signaling Is Required for Weight Loss after Gastric Bypass Surgery. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1023-E1031.	3.6	143
60	Small-intestinal dysfunction accompanies the complex endocrinopathy of human proprotein convertase 1 deficiency. Journal of Clinical Investigation, 2003, 112, 1550-1560.	8.2	140
61	Rare variants in single-minded 1 (SIM1) are associated with severe obesity. Journal of Clinical Investigation, 2013, 123, 3042-3050.	8.2	135
62	Obesity due to Melanocortin 4 Receptor (MC4R) Deficiency Is Associated with Increased Linear Growth and Final Height, Fasting Hyperinsulinemia, and Incompletely Suppressed Growth Hormone Secretion. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E181-E188.	3.6	120
63	Genetics of obesity. Philosophical Transactions of the Royal Society B: Biological Sciences, 2006, 361, 1095-1105.	4.0	118
64	Leptin and the Onset of Puberty: Insights from Rodent and Human Genetics. Seminars in Reproductive Medicine, 2002, 20, 139-144.	1.1	117
65	Recent advances: Recent advances in the genetics of severe childhood obesity. Archives of Disease in Childhood, 2000, 83, 31-34.	1.9	113
66	A Quantitative Trait Locus on Chromosome 18q for Physical Activity and Dietary Intake in Hispanic Children. Obesity, 2006, 14, 1596-1604.	3.0	113
67	Food addiction: is there a baby in the bathwater?. Nature Reviews Neuroscience, 2012, 13, 514-514.	10.2	102
68	Genetic architecture of human thinness compared to severe obesity. PLoS Genetics, 2019, 15, e1007603.	3.5	98
69	Genetic and hereditary aspects of childhood obesity. Best Practice and Research in Clinical Endocrinology and Metabolism, 2005, 19, 359-374.	4.7	94
70	Prevalence of Loss-of-Function FTO Mutations in Lean and Obese Individuals. Diabetes, 2010, 59, 311-318.	0.6	93
71	Resistance to thyroid hormone is associated with raised energy expenditure, muscle mitochondrial uncoupling, and hyperphagia. Journal of Clinical Investigation, 2010, 120, 1345-1354.	8.2	90
72	Effects of the mu-opioid receptor antagonist GSK1521498 on hedonic and consummatory eating behaviour: a proof of mechanism study in binge-eating obese subjects. Molecular Psychiatry, 2013, 18, 1287-1293.	7.9	89

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73	CNS Leptin Action Modulates Immune Response and Survival in Sepsis. Journal of Neuroscience, 2010, 30, 6036-6047.	3.6	86
74	Monogenic Human Obesity. , 2008, 36, 1-11.		84
75	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. Cell Reports, 2018, 22, 3401-3408.	6.4	81
76	Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. Cell, 2019, 176, 729-742.e18.	28.9	80
77	Neural and Behavioral Effects of a Novel Mu Opioid Receptor Antagonist in Binge-Eating Obese People. Biological Psychiatry, 2013, 73, 887-894.	1.3	79
78	Leptin Regulates Peripheral Lipid Metabolism Primarily through Central Effects on Food Intake. Endocrinology, 2008, 149, 5432-5439.	2.8	78
79	Functional Characterization and Structural Modeling of Obesity Associated Mutations in the Melanocortin 4 Receptor. Endocrinology, 2009, 150, 114-125.	2.8	75
80	Pharmacological Chaperones Restore Function to MC4R Mutants Responsible for Severe Early-Onset Obesity. Journal of Pharmacology and Experimental Therapeutics, 2010, 335, 520-532.	2.5	74
81	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.	3.6	71
82	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.	3.6	69
83	Severe obesity and diabetes insipidus in a patient with PCSK1 deficiency. Molecular Genetics and Metabolism, 2013, 110, 191-194.	1.1	62
84	A Metabolomic Signature of Acute Caloric Restriction. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4486-4495.	3.6	52
85	Melaninâ€Concentrating Hormone Receptor Mutations and Human Obesity: Functional Analysis. Obesity, 2004, 12, 743-749.	4.0	50
86	Mutations in the Amino-Terminal Region of Proopiomelanocortin (POMC) in Patients with Early-Onset Obesity Impair POMC Sorting to the Regulated Secretory Pathway. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4494-4499.	3.6	50
87	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394.	3.3	50
88	Loss-of-function mutations in the melanocortin 4 receptor in a UK birth cohort. Nature Medicine, 2021, 27, 1088-1096.	30.7	49
89	Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. Nature Communications, 2016, 7, 13055.	12.8	46
90	Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. Nature Communications, 2019, 10, 1718.	12.8	45

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91	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Brain, 2017, 140, 171-183.	7.6	43
92	Genetic, molecular and physiological insights into human obesity. European Journal of Clinical Investigation, 2011, 41, 451-455.	3.4	42
93	Molecular Genetic Analysis of Normosmic Hypogonadotropic Hypogonadism in a Turkish Population: Identification and Detailed Functional Characterization of a Novel Mutation in the Gonadotropin-Releasing Hormone Receptor Gene. Neuroendocrinology, 2006, 84, 301-308.	2.5	41
94	Functional Characterization of Naturally Occurring Pathogenic Mutations in the Human Leptin Receptor. Endocrinology, 2008, 149, 6043-6052.	2.8	40
95	Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. Scientific Reports, 2020, 10, 9028.	3.3	40
96	Defining the neural basis of appetite and obesity: from genes to behaviour. Clinical Medicine, 2014, 14, 286-289.	1.9	39
97	Functional Characterization of Obesity-Associated Variants Involving the Î± and Î² Isoforms of Human SH2B1. Endocrinology, 2014, 155, 3219-3226.	2.8	39
98	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.	16.2	38
99	Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. Cell Reports, 2021, 34, 108862.	6.4	37
100	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.	3.8	36
101	Melanocortin receptors weigh in. Nature Medicine, 2004, 10, 351-352.	30.7	35
102	Oxytocin administration suppresses hypothalamic activation in response to visual food cues. Scientific Reports, 2017, 7, 4266.	3.3	28
103	The severely obese patientâ€"a genetic work-up. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 172-177.	2.8	24
104	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.	3.5	23
105	Mutational analysis of the serotonin receptor 5HT2c in severe early-onset human obesity. Canadian Journal of Physiology and Pharmacology, 2004, 82, 426-429.	1.4	22
106	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.	6.5	21
107	Leptin-Mediated Changes in the Human Metabolome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2541-2552.	3.6	20
108	The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. Sleep, 2016, 39, 1691-1700.	1.1	19

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109	Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. FASEB Journal, 2018, 32, 1830-1840.	0.5	19
110	Obesity-Associated Melanocortin-4 Receptor Mutations Are Associated With Changes in the Brain Response to Food Cues. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2101-E2106.	3.6	18
111	Monogenic human obesity syndromes. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 181, 301-310.	1.8	18
112	Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. Diabetes, 2019, 68, 2049-2062.	0.6	16
113	Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. Molecular Metabolism, 2017, 6, 1419-1428.	6.5	15
114	Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors. Biological Psychiatry, 2022, 91, 856-859.	1.3	15
115	Is leptin an important physiological regulator of CRP?. Nature Medicine, 2007, 13, 16-17.	30.7	10
116	FTO and Obesity: The Missing Link. Cell Metabolism, 2011, 13, 7-8.	16.2	10
117	Postprandial Total Ghrelin Suppression Is Modulated by Melanocortin Signaling in Humans. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E288-E292.	3.6	10
118	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.	3.7	10
119	Genetic aspects of severe childhood obesity. Pediatric Endocrinology Reviews, 2006, 3 Suppl 4, 528-36.	1.2	10
120	A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476.	1.1	7
121	Predicting novel candidate human obesity genes and their site of action by systematic functional screening in Drosophila. PLoS Biology, 2021, 19, e3001255.	5.6	7
122	Hypothalamic atrophy is related to body mass index and age at onset in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 1006-1007.	1.9	6
123	Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. Clinical Endocrinology, 2022, 96, 270-275.	2.4	6
124	Evaluation of Praderâ€Willi Syndrome Gene MAGEL2 in Severe Childhoodâ€Onset Obesity. Obesity, 2005, 13, 1841-1842.	4.0	5
125	EJE PRIZE 2012: Obesity: from genes to behaviour. European Journal of Endocrinology, 2014, 171, R191-R195.	3.7	5
126	Genetic Obesity Syndromes. , 2014, , 23-32.		5

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127	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.	3.6	5
128	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.	28.9	3
129	Melanocortin receptors and energy homeostasis. Current Opinion in Endocrinology, Diabetes and Obesity, 2005, 12, 205-210.	0.6	2
130	Wired for Obesity?. Diabetes, 2014, 63, 4016-4017.	0.6	2
131	Genetic Syndromes Associated with Obesity. , 2016, , 491-497.e2.		1
132	Leptin and the Onset of Puberty. , 2003, , 287-295.		1
133	Genetics of chronic disease: obesity. , 0, , 328-343.		0