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	Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. Clinical Endocrinology, 2022, 96, 270-275.	2.4	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.	3.6	5
3	Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors. Biological Psychiatry, 2022, 91, 856-859.	1.3	15
4	Monogenic human obesity syndromes. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 181, 301-310.	1.8	18
5	Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. Cell Reports, 2021, 34, 108862.	6.4	37
6	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
7	Loss-of-function mutations in the melanocortin 4 receptor in a UK birth cohort. Nature Medicine, 2021, 27, 1088-1096.	30.7	49
8	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
9	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
10	Leptin-Mediated Changes in the Human Metabolome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2541-2552.	3.6	20
11	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
12	Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. Scientific Reports, 2020, 10, 9028.	3.3	40
13	Genetic architecture of human thinness compared to severe obesity. PLoS Genetics, 2019, 15, e1007603.	3.5	98
14	Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. Cell, 2019, 177, 597-607.e9.	28.9	192
15	Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. Nature Communications, 2019, 10, 1718.	12.8	45
16	Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. Diabetes, 2019, 68, 2049-2062.	0.6	16
17	Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. Cell, 2019, 176, 729-742.e18.	28.9	80
18	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

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19	Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. FASEB Journal, 2018, 32, 1830-1840.	0.5	19
20	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
21	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
22	MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. Nature Medicine, 2018, 24, 551-555.	30.7	219
23	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
24	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
25	A Metabolomic Signature of Acute Caloric Restriction. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4486-4495.	3.6	52
26	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
27	Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. Molecular Metabolism, 2017, 6, 1321-1329.	6.5	200
28	Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. Molecular Metabolism, 2017, 6, 1419-1428.	6.5	15
29	Oxytocin administration suppresses hypothalamic activation in response to visual food cues. Scientific Reports, 2017, 7, 4266.	3.3	28
30	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394.	3.3	50
31	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Brain, 2017, 140, 171-183.	7.6	43
32	Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. Nature Communications, 2016, 7, 13055.	12.8	46
33	The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. Sleep, 2016, 39, 1691-1700.	1.1	19
34	Genetic Syndromes Associated with Obesity. , 2016, , 491-497.e2.		1
35	The Hunger Genes: Pathways to Obesity. Cell, 2015, 161, 119-132.	28.9	293
36	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

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37	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
38	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
39	Leptin Mediates the Increase in Blood Pressure Associated with Obesity. Cell, 2014, 159, 1404-1416.	28.9	288
40	Defining the neural basis of appetite and obesity: from genes to behaviour. Clinical Medicine, 2014, 14, 286-289.	1.9	39
41	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
42	Functional Characterization of Obesity-Associated Variants Involving the Î± and Î² Isoforms of Human SH2B1. Endocrinology, 2014, 155, 3219-3226.	2.8	39
43	Wired for Obesity?. Diabetes, 2014, 63, 4016-4017.	0.6	2
44	EJE PRIZE 2012: Obesity: from genes to behaviour. European Journal of Endocrinology, 2014, 171, R191-R195.	3.7	5
45	20 YEARS OF LEPTIN: Human disorders of leptin action. Journal of Endocrinology, 2014, 223, T63-T70.	2.6	218
46	A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476.	1.1	7
47	Genetic Obesity Syndromes. , 2014, , 23-32.		5
48	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
49	KSR2 Mutations Are Associated with Obesity, Insulin Resistance, and Impaired Cellular Fuel Oxidation. Cell, 2013, 155, 765-777.	28.9	154
50	Severe obesity and diabetes insipidus in a patient with PCSK1 deficiency. Molecular Genetics and Metabolism, 2013, 110, 191-194.	1.1	62
51	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
52	Postprandial Total Ghrelin Suppression Is Modulated by Melanocortin Signaling in Humans. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E288-E292.	3.6	10
53	Loss of Function of the Melanocortin 2 Receptor Accessory Protein 2 Is Associated with Mammalian Obesity. Science, 2013, 341, 275-278.	12.6	225
54	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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55	Rare variants in single-minded 1 (SIM1) are associated with severe obesity. Journal of Clinical Investigation, 2013, 123, 3042-3050.	8.2	135
56	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
57	Food addiction: is there a baby in the bathwater?. Nature Reviews Neuroscience, 2012, 13, 514-514.	10.2	102
58	A genome-wide association meta-analysis identifies new childhood obesity loci. Nature Genetics, 2012, 44, 526-531.	21.4	352
59	Obesity and the brain: how convincing is the addiction model?. Nature Reviews Neuroscience, 2012, 13, 279-286.	10.2	409
60	Human SH2B1 mutations are associated with maladaptive behaviors and obesity. Journal of Clinical Investigation, 2012, 122, 4732-4736.	8.2	147
61	FTO and Obesity: The Missing Link. Cell Metabolism, 2011, 13, 7-8.	16.2	10
62	Genetic approaches to understanding human obesity. Journal of Clinical Investigation, 2011, 121, 2080-2086.	8.2	161
63	Genetic, molecular and physiological insights into human obesity. European Journal of Clinical Investigation, 2011, 41, 451-455.	3.4	42
64	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
65	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
66	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
67	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
68	Prevalence of Loss-of-Function FTO Mutations in Lean and Obese Individuals. Diabetes, 2010, 59, 311-318.	0.6	93
69	CNS Leptin Action Modulates Immune Response and Survival in Sepsis. Journal of Neuroscience, 2010, 30, 6036-6047.	3.6	86
70	Antidiabetic Effects of IGFBP2, a Leptin-Regulated Gene. Cell Metabolism, 2010, 11, 11-22.	16.2	251
71	Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670.	27.8	487
72	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

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73	Leptin: a pivotal regulator of human energy homeostasis. American Journal of Clinical Nutrition, 2009, 89, 980S-984S.	4.7	261
74	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
75	Functional Characterization and Structural Modeling of Obesity Associated Mutations in the Melanocortin 4 Receptor. Endocrinology, 2009, 150, 114-125.	2.8	75
76	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
77	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
78	Modulation of Blood Pressure by Central Melanocortinergic Pathways. New England Journal of Medicine, 2009, 360, 44-52.	27.0	412
79	Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nature Genetics, 2008, 40, 768-775.	21.4	1,179
80	Obesity Associated Genetic Variation inFTOIs Associated with Diminished Satiety. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3640-3643.	3.6	443
81	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
82	Monogenic Human Obesity. , 2008, 36, 1-11.		84
83	Human Obesity: A Heritable Neurobehavioral Disorder That Is Highly Sensitive to Environmental Conditions. Diabetes, 2008, 57, 2905-2910.	0.6	160
84	Prevalence of Melanocortin-4 Receptor Deficiency in Europeans and Their Age-Dependent Penetrance in Multigenerational Pedigrees. Diabetes, 2008, 57, 2511-2518.	0.6	229
85	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
86	Leptin Regulates Peripheral Lipid Metabolism Primarily through Central Effects on Food Intake. Endocrinology, 2008, 149, 5432-5439.	2.8	78
87	Functional Characterization of Naturally Occurring Pathogenic Mutations in the Human Leptin Receptor. Endocrinology, 2008, 149, 6043-6052.	2.8	40
88	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Presenting in Childhood. Pediatrics, 2007, 120, e179-e188.	2.1	175
89	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
90	Clinical and Molecular Genetic Spectrum of Congenital Deficiency of the Leptin Receptor. New England Journal of Medicine, 2007, 356, 237-247.	27.0	610

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91	The Hormonal Control of Food Intake. Cell, 2007, 129, 251-262.	28.9	508
92	The Obesity-AssociatedFTOGene Encodes a 2-Oxoglutarate-Dependent Nucleic Acid Demethylase. Science, 2007, 318, 1469-1472.	12.6	1,305
93	Leptin Regulates Striatal Regions and Human Eating Behavior. Science, 2007, 317, 1355-1355.	12.6	541
94	Is leptin an important physiological regulator of CRP?. Nature Medicine, 2007, 13, 16-17.	30.7	10
95	The central melanocortin system directly controls peripheral lipid metabolism. Journal of Clinical Investigation, 2007, 117, 3475-3488.	8.2	341
96	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
97	Genetics of obesity. Philosophical Transactions of the Royal Society B: Biological Sciences, 2006, 361, 1095-1105.	4.0	118
98	A POMC variant implicates Î²-melanocyte-stimulating hormone in the control of human energy balance. Cell Metabolism, 2006, 3, 135-140.	16.2	207
99	A Quantitative Trait Locus on Chromosome 18q for Physical Activity and Dietary Intake in Hispanic Children. Obesity, 2006, 14, 1596-1604.	3.0	113
100	The severely obese patientâ€"a genetic work-up. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 172-177.	2.8	24
101	Genetics of Obesity in Humans. Endocrine Reviews, 2006, 27, 710-718.	20.1	452
102	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
103	Heterozygosity for a POMC-Null Mutation and Increased Obesity Risk in Humans. Diabetes, 2006, 55, 2549-2553.	0.6	205
104	Genetic aspects of severe childhood obesity. Pediatric Endocrinology Reviews, 2006, 3 Suppl 4, 528-36.	1.2	10
105	Melanocortin receptors and energy homeostasis. Current Opinion in Endocrinology, Diabetes and Obesity, 2005, 12, 205-210.	0.6	2
106	Evaluation of Praderâ€Willi Syndrome Gene MAGEL2 in Severe Childhoodâ€Onset Obesity. Obesity, 2005, 13, 1841-1842.	4.0	5
107	Genetic and hereditary aspects of childhood obesity. Best Practice and Research in Clinical Endocrinology and Metabolism, 2005, 19, 359-374.	4.7	94
108	Monogenic Obesity in Humans. Annual Review of Medicine, 2005, 56, 443-458.	12.2	367

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109	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
110	Proopiomelanocortin and Energy Balance: Insights from Human and Murine Genetics. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 2557-2562.	3.6	197
111	Melanocortin receptors weigh in. Nature Medicine, 2004, 10, 351-352.	30.7	35
112	A de novo mutation affecting human TrkB associated with severe obesity and developmental delay. Nature Neuroscience, 2004, 7, 1187-1189.	14.8	499
113	Melaninâ€Concentrating Hormone Receptor Mutations and Human Obesity: Functional Analysis. Obesity, 2004, 12, 743-749.	4.0	50
114	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
115	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
116	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
117	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
118	Minireview: Human Obesityâ€”Lessons from Monogenic Disorders. Endocrinology, 2003, 144, 3757-3764.	2.8	194
119	Leptin and the Onset of Puberty. , 2003, , 287-295.		1
120	Small-intestinal dysfunction accompanies the complex endocrinopathy of human proprotein convertase 1 deficiency. Journal of Clinical Investigation, 2003, 112, 1550-1560.	8.2	140
121	Small-intestinal dysfunction accompanies the complex endocrinopathy of human proprotein convertase 1 deficiency. Journal of Clinical Investigation, 2003, 112, 1550-1560.	8.2	276
122	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
123	Leptin and the Onset of Puberty: Insights from Rodent and Human Genetics. Seminars in Reproductive Medicine, 2002, 20, 139-144.	1.1	117
124	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
125	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
126	Partial leptin deficiency and human adiposity. Nature, 2001, 414, 34-35.	27.8	356

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127	Genetics of body-weight regulation. Nature, 2000, 404, 644-651.	27.8	682
128	Recent advances: Recent advances in the genetics of severe childhood obesity. Archives of Disease in Childhood, 2000, 83, 31-34.	1.9	113
129	Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. Journal of Clinical Investigation, 2000, 106, 271-279.	8.2	696
130	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
131	A frameshift mutation in MC4R associated with dominantly inherited human obesity. Nature Genetics, 1998, 20, 111-112.	21.4	1,026
132	Congenital leptin deficiency is associated with severe early-onset obesity in humans. Nature, 1997, 387, 903-908.	27.8	2,664
133	Genetics of chronic disease: obesity. , 0, , 328-343.		0