

Sadaf Farooqi

List of Publications by Citations

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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	Congenital leptin deficiency is associated with severe early-onset obesity in humans. <i>Nature</i> , 1997 , 387, 903-8	50.4	2309
180	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , 2010 , 42, 937-48	36.3	2267
179	Effects of recombinant leptin therapy in a child with congenital leptin deficiency. <i>New England Journal of Medicine</i> , 1999 , 341, 879-84	59.2	1485
178	Clinical spectrum of obesity and mutations in the melanocortin 4 receptor gene. <i>New England Journal of Medicine</i> , 2003 , 348, 1085-95	59.2	1227
177	A frameshift mutation in MC4R associated with dominantly inherited human obesity. <i>Nature Genetics</i> , 1998 , 20, 111-2	36.3	893
176	Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. <i>Journal of Clinical Investigation</i> , 2002 , 110, 1093-1103	15.9	845
175	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015 , 526, 82-90	50.4	776
174	Genetics of body-weight regulation. <i>Nature</i> , 2000 , 404, 644-51	50.4	593
173	Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. <i>Journal of Clinical Investigation</i> , 2000 , 106, 271-9	15.9	589
172	Clinical and molecular genetic spectrum of congenital deficiency of the leptin receptor. <i>New England Journal of Medicine</i> , 2007 , 356, 237-47	59.2	503
171	Leptin regulates striatal regions and human eating behavior. <i>Science</i> , 2007 , 317, 1355	33.3	452
170	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. <i>Lancet, The</i> , 2015 , 385, 1305-14	40	451
169	Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. <i>Journal of Clinical Investigation</i> , 2002 , 110, 1093-103	15.9	441
168	Large, rare chromosomal deletions associated with severe early-onset obesity. <i>Nature</i> , 2010 , 463, 666-70	50.4	417
167	Genetics of obesity in humans. <i>Endocrine Reviews</i> , 2006 , 27, 710-18	27.2	376
166	Modulation of blood pressure by central melanocortinergic pathways. <i>New England Journal of Medicine</i> , 2009 , 360, 44-52	59.2	358
165	Monogenic obesity in humans. <i>Annual Review of Medicine</i> , 2005 , 56, 443-58	17.4	308

164	Partial leptin deficiency and human adiposity. <i>Nature</i> , 2001 , 414, 34-5	50.4	305
163	A genome-wide association meta-analysis identifies new childhood obesity loci. <i>Nature Genetics</i> , 2012 , 44, 526-31	36.3	292
162	Leptin mediates the increase in blood pressure associated with obesity. <i>Cell</i> , 2014 , 159, 1404-16	56.2	232
161	Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. <i>Nature Genetics</i> , 2013 , 45, 513-7	36.3	231
160	Leptin: a pivotal regulator of human energy homeostasis. <i>American Journal of Clinical Nutrition</i> , 2009 , 89, 980S-984S	7	226
159	The hunger genes: pathways to obesity. <i>Cell</i> , 2015 , 161, 119-132	56.2	216
158	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017 , 101, 664-685	11	214
157	Identification of SATB2 as the cleft palate gene on 2q32-q33. <i>Human Molecular Genetics</i> , 2003 , 12, 2491-501	50.1	209
156	Paediatric genomics: diagnosing rare disease in children. <i>Nature Reviews Genetics</i> , 2018 , 19, 253-268	30.1	201
155	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. <i>Nature Genetics</i> , 2016 , 48, 1060-5	36.3	200
154	Prevalence of melanocortin-4 receptor deficiency in Europeans and their age-dependent penetrance in multigenerational pedigrees. <i>Diabetes</i> , 2008 , 57, 2511-8	0.9	198
153	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. <i>Nature Genetics</i> , 2018 , 50, 26-41	36.3	186
152	Mutations in ligands and receptors of the leptin-melanocortin pathway that lead to obesity. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , 2008 , 4, 569-77		184
151	Loss of function of the melanocortin 2 receptor accessory protein 2 is associated with mammalian obesity. <i>Science</i> , 2013 , 341, 275-8	33.3	179
150	Heterozygosity for a POMC-null mutation and increased obesity risk in humans. <i>Diabetes</i> , 2006 , 55, 2549-53	53	176
149	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017 , 49, 36-45	36.3	172
148	Oral glutamine increases circulating glucagon-like peptide 1, glucagon, and insulin concentrations in lean, obese, and type 2 diabetic subjects. <i>American Journal of Clinical Nutrition</i> , 2009 , 89, 106-113	7	171
147	The genetic architecture of microphthalmia, anophthalmia and coloboma. <i>European Journal of Medical Genetics</i> , 2014 , 57, 369-80	2.6	168

146	Hyperphagia and early-onset obesity due to a novel homozygous missense mutation in prohormone convertase 1/3. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007 , 92, 3369-73	5.6	166
145	20 years of leptin: human disorders of leptin action. <i>Journal of Endocrinology</i> , 2014 , 223, T63-70	4.7	163
144	GDF15 Provides an Endocrine Signal of Nutritional Stress in Mice and Humans. <i>Cell Metabolism</i> , 2019 , 29, 707-718.e8	24.6	153
143	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. <i>Nature</i> , 2018 , 562, 268-271	50.4	149
142	MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. <i>Nature Medicine</i> , 2018 , 24, 551-555	50.5	139
141	Human obesity: a heritable neurobehavioral disorder that is highly sensitive to environmental conditions. <i>Diabetes</i> , 2008 , 57, 2905-10	0.9	129
140	Transcriptome analysis of human autosomal trisomy. <i>Human Molecular Genetics</i> , 2002 , 11, 3249-56	5.6	127
139	Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. <i>Molecular Metabolism</i> , 2017 , 6, 1321-1329	8.8	121
138	Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. <i>Cell</i> , 2019 , 177, 597-607.e9	56.2	113
137	KSR2 mutations are associated with obesity, insulin resistance, and impaired cellular fuel oxidation. <i>Cell</i> , 2013 , 155, 765-77	56.2	113
136	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. <i>Journal of Medical Genetics</i> , 2014 , 51, 659-68	5.8	111
135	Rare variants in single-minded 1 (SIM1) are associated with severe obesity. <i>Journal of Clinical Investigation</i> , 2013 , 123, 3042-50	15.9	107
134	Human SH2B1 mutations are associated with maladaptive behaviors and obesity. <i>Journal of Clinical Investigation</i> , 2012 , 122, 4732-6	15.9	103
133	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020 , 586, 757-762	50.4	103
132	Leptin and the onset of puberty: insights from rodent and human genetics. <i>Seminars in Reproductive Medicine</i> , 2002 , 20, 139-44	1.4	95
131	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. <i>Nature Genetics</i> , 2015 , 47, 1363-9	36.3	91
130	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017 , 49, 238-248	36.3	88
129	Amyotrophic lateral sclerosis and frontotemporal dementia: distinct and overlapping changes in eating behaviour and metabolism. <i>Lancet Neurology</i> , 2016 , 15, 332-42	24.1	88

128	Food addiction: is there a baby in the bathwater?. <i>Nature Reviews Neuroscience</i> , 2012 , 13, 514-514	13.5	88
127	Quantifying the contribution of recessive coding variation to developmental disorders. <i>Science</i> , 2018 , 362, 1161-1164	33.3	83
126	Diagnostically relevant facial gestalt information from ordinary photos. <i>ELife</i> , 2014 , 3, e02020	8.9	81
125	Developmental eye disorders. <i>Current Opinion in Genetics and Development</i> , 2005 , 15, 348-53	4.9	80
124	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. <i>Lancet Diabetes and Endocrinology</i> , 2020 , 8, 960-970	18.1	76
123	Neural and behavioral effects of a novel mu opioid receptor antagonist in binge-eating obese people. <i>Biological Psychiatry</i> , 2013 , 73, 887-94	7.9	75
122	Heterozygous loss-of-function mutations in YAP1 cause both isolated and syndromic optic fissure closure defects. <i>American Journal of Human Genetics</i> , 2014 , 94, 295-302	11	74
121	Monogenic human obesity. <i>Frontiers of Hormone Research</i> , 2008 , 36, 1-11	3.5	73
120	Leptin regulates peripheral lipid metabolism primarily through central effects on food intake. <i>Endocrinology</i> , 2008 , 149, 5432-9	4.8	71
119	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. <i>Molecular Genetics & Genomic Medicine</i> , 2013 , 1, 15-31	2.3	69
118	Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. <i>Trends in Genetics</i> , 2005 , 21, 249-53	8.5	65
117	Distinct modulatory effects of satiety and sibutramine on brain responses to food images in humans: a double dissociation across hypothalamus, amygdala, and ventral striatum. <i>Journal of Neuroscience</i> , 2010 , 30, 14346-55	6.6	61
116	A locus for isolated cleft palate, located on human chromosome 2q32. <i>American Journal of Human Genetics</i> , 1999 , 65, 387-96	11	60
115	De novo, heterozygous, loss-of-function mutations in SYNGAP1 cause a syndromic form of intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2231-7	2.5	59
114	Genetic and hereditary aspects of childhood obesity. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2005 , 19, 359-74	6.5	57
113	Assessment of Eating Behavior Disturbance and Associated Neural Networks in Frontotemporal Dementia. <i>JAMA Neurology</i> , 2016 , 73, 282-90	17.2	56
112	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. <i>American Journal of Human Genetics</i> , 2016 , 98, 981-992	11	53
111	BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Lange-like syndrome. <i>Nature Genetics</i> , 2018 , 50, 329-332	36.3	52

110	Genetic architecture of human thinness compared to severe obesity. <i>PLoS Genetics</i> , 2019 , 15, e1007603	6	51
109	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. <i>Cell Reports</i> , 2018 , 22, 3401-3408	10.6	51
108	Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. <i>Lancet, The</i> , 2018 , 391, 1483-1492	40	49
107	Severe Early-Onset Obesity Due to Bioinactive Leptin Caused by a p.N103K Mutation in the Leptin Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, 3227-30	5.6	44
106	Cardiac Genetic Predisposition in Sudden Infant Death Syndrome. <i>Journal of the American College of Cardiology</i> , 2018 , 71, 1217-1227	15.1	43
105	Disruption of SATB2 or its long-range cis-regulation by SOX9 causes a syndromic form of Pierre Robin sequence. <i>Human Molecular Genetics</i> , 2014 , 23, 2569-79	5.6	42
104	Ambulatory blood pressure monitoring in acute stroke. The West Birmingham Stroke Project. <i>Stroke</i> , 1997 , 28, 31-5	6.7	39
103	Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. <i>Cell</i> , 2019 , 176, 729-742.e18	56.2	38
102	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017 , 100, 650-658	11	36
101	Genetic, molecular and physiological insights into human obesity. <i>European Journal of Clinical Investigation</i> , 2011 , 41, 451-5	4.6	36
100	Lipid Metabolism and Survival Across the Frontotemporal Dementia-Amyotrophic Lateral Sclerosis Spectrum: Relationships to Eating Behavior and Cognition. <i>Journal of Alzheimer's Disease</i> , 2018 , 61, 773-783	4.3	35
99	ob gene mutations and human obesity. <i>Proceedings of the Nutrition Society</i> , 1998 , 57, 471-5	2.9	35
98	Genetic Analysis of PAX6-Negative Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016 , 11, e0153757	3.7	34
97	Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. <i>Nature Communications</i> , 2016 , 7, 13055	17.4	31
96	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. <i>Scientific Reports</i> , 2017 , 7, 4394	4.9	31
95	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. <i>Genetics in Medicine</i> , 2017 , 19, 900-908	8.1	30
94	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. <i>Human Molecular Genetics</i> , 2016 , 25, 1382-91	5.6	30
93	The RNA-binding landscape of RBM10 and its role in alternative splicing regulation in models of mouse early development. <i>RNA Biology</i> , 2017 , 14, 45-57	4.8	30

92	Melanocortin Receptors as Targets in the Treatment of Obesity. <i>Current Topics in Medicinal Chemistry</i> , 2007 , 7, 1098-1110	3	30
91	The orphan G protein-coupled receptor GPR139 is activated by the peptides: Adrenocorticotrophic hormone (ACTH), β and β melanocyte stimulating hormone (β MSH, and β MSH), and the conserved core motif HFRW. <i>Neurochemistry International</i> , 2017 , 102, 105-113	4.4	29
90	A Metabolomic Signature of Acute Caloric Restriction. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 4486-4495	5.6	28
89	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. <i>Brain</i> , 2017 , 140, 1711-1723	11.3	28
88	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015 , 23,	5.3	28
87	Defining the neural basis of appetite and obesity: from genes to behaviour. <i>Clinical Medicine</i> , 2014 , 14, 286-9	1.9	27
86	Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. <i>Open Biology</i> , 2015 , 5, 1500477		26
85	FRA2A is a CGG repeat expansion associated with silencing of AFF3. <i>PLoS Genetics</i> , 2014 , 10, e1004242	6	26
84	The genetic architecture of aniridia and Gillespie syndrome. <i>Human Genetics</i> , 2019 , 138, 881-898	6.3	26
83	Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. <i>European Journal of Medical Genetics</i> , 2017 , 60, 130-135	2.6	25
82	Contribution of retrotransposition to developmental disorders. <i>Nature Communications</i> , 2019 , 10, 4630	17.4	25
81	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. <i>European Journal of Human Genetics</i> , 2017 , 25, 552-559	5.3	24
80	Oxytocin administration suppresses hypothalamic activation in response to visual food cues. <i>Scientific Reports</i> , 2017 , 7, 4266	4.9	23
79	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. <i>Nature Communications</i> , 2019 , 10, 2373	17.4	22
78	Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. <i>Nature Communications</i> , 2019 , 10, 1718	17.4	20
77	Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. <i>Nature Communications</i> , 2015 , 6, 6904	17.4	20
76	A New Drug Target for Type 2 Diabetes. <i>Cell</i> , 2017 , 170, 12-14	56.2	20
75	The severely obese patient--a genetic work-up. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , 2006 , 2, 172-7; quiz following 177		20

74	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. <i>Brain</i> , 2020 , 143, 3242-3261	11.2	19
73	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. <i>Appetite</i> , 2016 , 107, 596-603	4.5	19
72	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNA-protein interaction. <i>Genetics in Medicine</i> , 2020 , 22, 598-609	8.1	19
71	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. <i>Genome Research</i> , 2019 , 29, 1047-1056	9.7	18
70	Insights from the genetics of severe childhood obesity. <i>Hormone Research in Paediatrics</i> , 2007 , 68 Suppl 5, 5-7	3.3	18
69	Hypothalamic Reproductive Endocrine Pulse Generator Activity Independent of Neurokinin B and Dynorphin Signaling. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019 , 104, 4304-4318	5.6	17
68	A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. <i>European Journal of Medical Genetics</i> , 2015 , 58, 387-91	2.6	17
67	Genotype-phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1566-1574	2.5	16
66	A recurrent de novo mutation in ACTG1 causes isolated ocular coloboma. <i>Human Mutation</i> , 2017 , 38, 942-946	4.7	16
65	Exome Sequencing Identifies Genes and Gene Sets Contributing to Severe Childhood Obesity, Linking Phip Variants to Repressed POMC Transcription. <i>Cell Metabolism</i> , 2020 , 31, 1107-1119.e12	24.6	16
64	polyadenylation signal variants cause syndromic microphthalmia. <i>Journal of Medical Genetics</i> , 2019 , 56, 444-452	5.8	15
63	NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. <i>Pediatrics</i> , 2018 , 141, S485-S490	7.4	14
62	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. <i>European Journal of Medical Genetics</i> , 2014 , 57, 587-95	2.6	14
61	The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. <i>Sleep</i> , 2016 , 39, 1691-700	1.1	13
60	ProxECAT: Proxy External Controls Association Test. A new case-control gene region association test using allele frequencies from public controls. <i>PLoS Genetics</i> , 2018 , 14, e1007591	6	13
59	Quantitative mass spectrometry for human melanocortin peptides in vitro and in vivo suggests prominent roles for E ₁ MSH and desacetyl E ₁ MSH in energy homeostasis. <i>Molecular Metabolism</i> , 2018 , 17, 82-97	8.8	13
58	Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature 2017 , 58, 594-603		12
57	Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. <i>Cell Reports</i> , 2021 , 34, 108862	10.6	12

56	Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. <i>Scientific Reports</i> , 2020 , 10, 9028	4.9	11
55	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. <i>Human Genetics</i> , 2020 , 139, 575-592	6.3	11
54	ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. <i>PLoS Genetics</i> , 2019 , 15, e1007605	6	10
53	Postprandial total ghrelin suppression is modulated by melanocortin signaling in humans. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013 , 98, E288-92	5.6	10
52	Carey-Fineman-Ziter (CFZ) syndrome: report on affected sibs. <i>American Journal of Medical Genetics Part A</i> , 1999 , 82, 110-3		10
51	Returning genome sequences to research participants: Policy and practice. <i>Wellcome Open Research</i> , 2017 , 2, 15	4.8	10
50	Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. <i>FASEB Journal</i> , 2018 , 32, 1830-1840	0.9	10
49	Genetic aspects of severe childhood obesity. <i>Pediatric Endocrinology Reviews</i> , 2006 , 3 Suppl 4, 528-36	1.1	10
48	Leptin-Mediated Changes in the Human Metabolome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	9
47	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. <i>Genetics in Medicine</i> , 2020 , 22, 878-888	8.1	9
46	Obesity genes-it's all about the parents!. <i>Cell Metabolism</i> , 2009 , 9, 487-8	24.6	9
45	Potential role of gender specific effect of leptin receptor deficiency in an extended consanguineous family with severe early-onset obesity. <i>European Journal of Medical Genetics</i> , 2018 , 61, 465-467	2.6	8
44	Expansion of ocular phenotypic features associated with mutations in ADAMTS18. <i>JAMA Ophthalmology</i> , 2014 , 132, 996-1001	3.9	8
43	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019 , 10, 611	4.5	7
42	Neural networks associated with body composition in frontotemporal dementia. <i>Annals of Clinical and Translational Neurology</i> , 2019 , 6, 1707-1717	5.3	7
41	Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. <i>Molecular Metabolism</i> , 2017 , 6, 1419-1428	8.8	7
40	Characterization of human variants in obesity-related SIM1 protein identifies a hot-spot for dimerization with the partner protein ARNT2. <i>Biochemical Journal</i> , 2014 , 461, 403-12	3.8	7
39	Genetic strategies to understand physiological pathways regulating body weight. <i>Mammalian Genome</i> , 2014 , 25, 377-83	3.2	7

38	A trans-acting protein effect causes severe eye malformation in the Mp mouse. <i>PLoS Genetics</i> , 2013 , 9, e1003998	6	7
37	FTO and obesity: the missing link. <i>Cell Metabolism</i> , 2011 , 13, 7-8	24.6	7
36	The therapeutic value of somatostatin and its analogues. <i>Pituitary</i> , 1999 , 2, 79-88	4.3	7
35	Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. <i>PLoS Genetics</i> , 2020 , 16, e1008916	6	7
34	Status dystonicus in two patients with SOX2-anophthalmia syndrome and nonsense mutations. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 3048-3050	2.5	6
33	Cornelia de Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. <i>Nature Communications</i> , 2021 , 12, 3127	17.4	6
32	Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. <i>Diabetes</i> , 2019 , 68, 2049-2062	0.9	6
31	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. <i>Genetics in Medicine</i> , 2021 , 23, 571-575	8.1	6
30	A novel mutation in the leptin gene (W121X) in an Egyptian family. <i>Molecular Genetics and Metabolism Reports</i> , 2014 , 1, 474-476	1.8	5
29	Treating obesity: does antagonism of NPY fit the bill?. <i>Cell Metabolism</i> , 2006 , 4, 260-2	24.6	5
28	The contribution of X-linked coding variation to severe developmental disorders. <i>Nature Communications</i> , 2021 , 12, 627	17.4	5
27	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. <i>American Journal of Human Genetics</i> , 2019 , 105, 933-946	11	4
26	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021 , 42, 445-459	4.7	4
25	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. <i>Genetics in Medicine</i> , 2022 ,	8.1	4
24	EJE Prize 2012: Obesity: from genes to behaviour. <i>European Journal of Endocrinology</i> , 2014 , 171, R191-5	6.5	3
23	Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors.. <i>Biological Psychiatry</i> , 2022 ,	7.9	3
22	Resequencing at scale in neurodevelopmental disorders. <i>Nature Genetics</i> , 2017 , 49, 488-489	36.3	2
21	Wired for obesity?. <i>Diabetes</i> , 2014 , 63, 4016-7	0.9	2

20	Obesity-Associated Mutations and the Melanocortin Pathway. <i>New England Journal of Medicine</i> , 2021 , 385, 1581-1592	59.2	2
19	Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. <i>Genome Research</i> , 2021 , 31, 1994-2007	9.7	2
18	Candidate genes for obesity--how might they interact with environment and diet?. <i>Advances in Experimental Medicine and Biology</i> , 2005 , 569, 33-4	3.6	2
17	Central obesity is selectively associated with cerebral gray matter atrophy in 15,634 subjects in the UK Biobank.. <i>International Journal of Obesity</i> , 2022 ,	5.5	1
16	Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. <i>Clinical Endocrinology</i> , 2021 ,	3.4	1
15	Reply to Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. <i>Cell</i> , 2021 , 184, 1652-1653	56.2	1
14	Neural correlates of fat preference in frontotemporal dementia: translating insights from the obesity literature. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 1318-1329	5.3	1
13	Monogenic human obesity syndromes. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2021 , 181, 301-310	3	1
12	Murine neuronatin deficiency is associated with a hypervariable food intake and bimodal obesity. <i>Scientific Reports</i> , 2021 , 11, 17571	4.9	1
11	Cornelia de Lange Syndrome		1
10	Genomically Aided Diagnosis of Severe Developmental Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2020 , 21, 327-349	9.7	0
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8	Predicting novel candidate human obesity genes and their site of action by systematic functional screening in Drosophila. <i>PLoS Biology</i> , 2021 , 19, e3001255	9.7	0
7	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. <i>American Journal of Human Genetics</i> , 2021 , 108, 2186-2194 ¹¹		0
6	Visualization of sympathetic neural innervation in human white adipose tissue.. <i>Open Biology</i> , 2022 , 12, 210345	7	0
5	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017 , 88, e1.18-e1	5.5	
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