Sadaf Farooqi

List of Publications by Year
in descending order

180 papers	30,196 citations	12322 69 h-index	4988 167 g-index
195 all docs	195 docs citations	195 times ranked	33444 citing authors

#	Article	IF	Citations
1	Congenital leptin deficiency is associated with severe early-onset obesity in humans. Nature, 1997, 387, 903-908.	13.7	2,664
2	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	9.4	2,634
3	Effects of Recombinant Leptin Therapy in a Child with Congenital Leptin Deficiency. New England Journal of Medicine, 1999, 341, 879-884.	13.9	1,760
4	Clinical Spectrum of Obesity and Mutations in the Melanocortin 4 Receptor Gene. New England Journal of Medicine, 2003, 348, 1085-1095.	13.9	1,475
5	A frameshift mutation in MC4R associated with dominantly inherited human obesity. Nature Genetics, 1998, 20, 111-112.	9.4	1,026
6	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	13.7	1,014
7	Beneficial effects of leptin on obesity, T cell hyporesponsiveness, and neuroendocrine/metabolic dysfunction of human congenital leptin deficiency. Journal of Clinical Investigation, 2002, 110, 1093-1103.	3.9	953
8	Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. Journal of Clinical Investigation, 2000, 106, 271-279.	3.9	696
9	Genetics of body-weight regulation. Nature, 2000, 404, 644-651.	13.7	682
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.	3.9	670
11	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	6.3	651
12	Clinical and Molecular Genetic Spectrum of Congenital Deficiency of the Leptin Receptor. New England Journal of Medicine, 2007, 356, 237-247.	13.9	610
13	Leptin Regulates Striatal Regions and Human Eating Behavior. Science, 2007, 317, 1355-1355.	6.0	541
14	Large, rare chromosomal deletions associated with severe early-onset obesity. Nature, 2010, 463, 666-670.	13.7	487
15	Genetics of Obesity in Humans. Endocrine Reviews, 2006, 27, 710-718.	8.9	452
16	Modulation of Blood Pressure by Central Melanocortinergic Pathways. New England Journal of Medicine, 2009, 360, 44-52.	13.9	412
17	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	7.7	369
18	Monogenic Obesity in Humans. Annual Review of Medicine, 2005, 56, 443-458.	5.0	367

#	Article	IF	Citations
19	Partial leptin deficiency and human adiposity. Nature, 2001, 414, 34-35.	13.7	356
20	A genome-wide association meta-analysis identifies new childhood obesity loci. Nature Genetics, 2012, 44, 526-531.	9.4	352
21	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	9.4	351
22	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
23	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
24	The Hunger Genes: Pathways to Obesity. Cell, 2015, 161, 119-132.	13.5	293
25	Leptin Mediates the Increase in Blood Pressure Associated with Obesity. Cell, 2014, 159, 1404-1416.	13.5	288
26	GDF15 Provides an Endocrine Signal of Nutritional Stress in Mice and Humans. Cell Metabolism, 2019, 29, 707-718.e8.	7.2	286
27	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	9.4	286
28	Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. Nature Genetics, 2013, 45, 513-517.	9.4	278
29	Leptin: a pivotal regulator of human energy homeostasis. American Journal of Clinical Nutrition, 2009, 89, 980S-984S.	2.2	261
30	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
31	Identification of SATB2 as the cleft palate gene on 2q32-q33. Human Molecular Genetics, 2003, 12, 2491-2501.	1.4	248
32	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	13.7	246
33	Efficacy and safety of setmelanotide, an MC4R agonist, in individuals with severe obesity due to LEPR or POMC deficiency: single-arm, open-label, multicentre, phase 3 trials. Lancet Diabetes and Endocrinology,the, 2020, 8, 960-970.	5.5	235
34	Prevalence of Melanocortin-4 Receptor Deficiency in Europeans and Their Age-Dependent Penetrance in Multigenerational Pedigrees. Diabetes, 2008, 57, 2511-2518.	0.3	229
35	Mutations in ligands and receptors of the leptinâ€“melanocortin pathway that lead to obesity. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 569-577.	2.9	225
36	Loss of Function of the Melanocortin 2 Receptor Accessory Protein 2 Is Associated with Mammalian Obesity. Science, 2013, 341, 275-278.	6.0	225

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37	MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. Nature Medicine, 2018, 24, 551-555.	15.2	219
38	20 YEARS OF LEPTIN: Human disorders of leptin action. Journal of Endocrinology, 2014, 223, T63-T70.	1.2	218
39	The genetic architecture of microphthalmia, anophthalmia and coloboma. European Journal of Medical Genetics, 2014, 57, 369-380.	0.7	213
40	Heterozygosity for aPOMC-Null Mutation and Increased Obesity Risk in Humans. Diabetes, 2006, 55, 2549-2553.	0.3	205
41	Oral glutamine increases circulating glucagon-like peptide 1, glucagon, and insulin concentrations in lean, obese, and type 2 diabetic subjects. American Journal of Clinical Nutrition, 2009, 89, 106-113.	2.2	201
42	Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. Molecular Metabolism, 2017, 6, 1321-1329.	3.0	200
43	Hyperphagia and Early-Onset Obesity due to a Novel Homozygous Missense Mutation in Prohormone Convertase 1/3. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3369-3373.	1.8	196
44	Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. Cell, 2019, 177, 597-607.e9.	13.5	192
45	Human Obesity: A Heritable Neurobehavioral Disorder That Is Highly Sensitive to Environmental Conditions. Diabetes, 2008, 57, 2905-2910.	0.3	160
46	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	6.0	158
47	KSR2 Mutations Are Associated with Obesity, Insulin Resistance, and Impaired Cellular Fuel Oxidation. Cell, 2013, 155, 765-777.	13.5	154
48	Transcriptome analysis of human autosomal trisomy. Human Molecular Genetics, 2002, 11, 3249-3256.	1.4	150
49	Human SH2B1 mutations are associated with maladaptive behaviors and obesity. Journal of Clinical Investigation, 2012, 122, 4732-4736.	3.9	147
50	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
51	Rare variants in single-minded 1 (SIM1) are associated with severe obesity. Journal of Clinical Investigation, 2013, 123, 3042-3050.	3.9	135
52	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	9.4	133
53	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
54	Diagnostically relevant facial gestalt information from ordinary photos. ELife, 2014, 3, e02020.	2.8	129

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55	Amyotrophic lateral sclerosis and frontotemporal dementia: distinct and overlapping changes in eating behaviour and metabolism. Lancet Neurology, The, 2016, 15, 332-342.	4.9	120
56	Leptin and the Onset of Puberty: Insights from Rodent and Human Genetics. Seminars in Reproductive Medicine, 2002, 20, 139-144.	0.5	117
57	Food addiction: is there a baby in the bathwater?. Nature Reviews Neuroscience, 2012, 13, 514-514.	4.9	102
58	Genetic architecture of human thinness compared to severe obesity. PLoS Genetics, 2019, 15, e1007603.	1.5	98
59	De novo, heterozygous, lossâ€ofâ€function mutations in SYNGAP1 cause a syndromic form of intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 2231-2237.	0.7	96
60	BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Langeâ€“like syndrome. Nature Genetics, 2018, 50, 329-332.	9.4	96
61	Developmental eye disorders. Current Opinion in Genetics and Development, 2005, 15, 348-353.	1.5	95
62	Genetic and hereditary aspects of childhood obesity. Best Practice and Research in Clinical Endocrinology and Metabolism, 2005, 19, 359-374.	2.2	94
63	Heterozygous Loss-of-Function Mutations in YAP1 Cause Both Isolated and Syndromic Optic Fissure Closure Defects. American Journal of Human Genetics, 2014, 94, 295-302.	2.6	93
64	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	5.8	86
65	Monogenic Human Obesity. , 2008, 36, 1-11.		84
66	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992.	2.6	81
67	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. Cell Reports, 2018, 22, 3401-3408.	2.9	81
68	Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. Cell, 2019, 176, 729-742.e18.	13.5	80
69	Neural and Behavioral Effects of a Novel Mu Opioid Receptor Antagonist in Binge-Eating Obese People. Biological Psychiatry, 2013, 73, 887-894.	0.7	79
70	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. Molecular Genetics & Genomic Medicine, 2013, 1, 15-31.	0.6	79
71	Leptin Regulates Peripheral Lipid Metabolism Primarily through Central Effects on Food Intake. Endocrinology, 2008, 149, 5432-5439.	1.4	78
72	Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. Trends in Genetics, 2005, 21, 249-253.	2.9	77

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73	Assessment of Eating Behavior Disturbance and Associated Neural Networks in Frontotemporal Dementia. JAMA Neurology, 2016, 73, 282.	4.5	74
74	Severe Early-Onset Obesity Due to Bioinactive Leptin Caused by a p.N103K Mutation in the Leptin Gene. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 3227-3230.	1.8	71
75	Distinct Modulatory Effects of Satiety and Sibutramine on Brain Responses to Food Images in Humans: A Double Dissociation across Hypothalamus, Amygdala, and Ventral Striatum. Journal of Neuroscience, 2010, 30, 14346-14355.	1.7	69
76	Cardiac Genetic Predisposition in SuddenÂInfant Death Syndrome. Journal of the American College of Cardiology, 2018, 71, 1217-1227.	1.2	66
77	A Locus for Isolated Cleft Palate, Located on Human Chromosome 2q32. American Journal of Human Genetics, 1999, 65, 387-396.	2.6	63
78	Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. Lancet, The, 2018, 391, 1483-1492.	6.3	63
79	A New Drug Target for Type 2 Diabetes. Cell, 2017, 170, 12-14.	13.5	62
80	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	3.7	57
81	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	2.6	56
82	Ambulatory Blood Pressure Monitoring in Acute Stroke. Stroke, 1997, 28, 31-35.	1.0	56
83	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	1.1	56
84	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	1.1	54
85	A Metabolomic Signature of Acute Caloric Restriction. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4486-4495.	1.8	52
86	Disruption of SATB2 or its long-range cis-regulation by SOX9 causes a syndromic form of Pierre Robin sequence. Human Molecular Genetics, 2014, 23, 2569-2579.	1.4	51
87	The genetic architecture of aniridia and Gillespie syndrome. Human Genetics, 2019, 138, 881-898.	1.8	51
88	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. Scientific Reports, 2017, 7, 4394.	1.6	50
89	Melanocortin Receptors as Targets in the Treatment of Obesity. Current Topics in Medicinal Chemistry, 2007, 7, 1098-1110.	1.0	47
90	Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. European Journal of Medical Genetics, 2017, 60, 130-135.	0.7	47

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91	Lipid Metabolism and Survival Across the Frontotemporal Dementia-Amyotrophic Lateral Sclerosis Spectrum: Relationships to Eating Behavior and Cognition. Journal of Alzheimer's Disease, 2017, 61, 773-783.	1.2	47
92	Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. Open Biology, 2015, 5, 150047.	1.5	46
93	Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. Nature Communications, 2016, 7, 13055.	5.8	46
94	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	1.1	46
95	Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. Nature Communications, 2019, 10, 1718.	5.8	45
96	ob gene mutations and human obesity. Proceedings of the Nutrition Society, 1998, 57, 471-475.	0.4	44
97	Obesity-Associated GNAS Mutations and the Melanocortin Pathway. New England Journal of Medicine, 2021, 385, 1581-1592.	13.9	44
98	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Brain, 2017, 140, 171-183.	3.7	43
99	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	5.8	43
100	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609.	1.1	43
101	Genetic, molecular and physiological insights into human obesity. European Journal of Clinical Investigation, 2011, 41, 451-455.	1.7	42
102	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	1.4	42
103	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	1.5	41
104	The RNA-binding landscape of RBM10 and its role in alternative splicing regulation in models of mouse early development. RNA Biology, 2017, 14, 45-57.	1.5	41
105	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391.	1.4	40
106	Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. Scientific Reports, 2020, 10, 9028.	1.6	40
107	Defining the neural basis of appetite and obesity: from genes to behaviour. Clinical Medicine, 2014, 14, 286-289.	0.8	39
108	Exome Sequencing Identifies Genes and Gene Sets Contributing to Severe Childhood Obesity, Linking PHIP Variants to Repressed POMC Transcription. Cell Metabolism, 2020, 31, 1107-1119.e12.	7.2	38

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109	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	1.4	37
110	Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. Cell Reports, 2021, 34, 108862.	2.9	37
111	The orphan G protein-coupled receptor GPR139 is activated by the peptides: Adrenocorticotropic hormone (ACTH), Î±-, and Î²-melanocyte stimulating hormone (Î±-MSH, and Î²-MSH), and the conserved core motif HFRW. Neurochemistry International, 2017, 102, 105-113.	1.9	36
112	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	2.4	34
113	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	5.8	33
114	Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. Nature Communications, 2015, 6, 6904.	5.8	31
115	Oxytocin administration suppresses hypothalamic activation in response to visual food cues. Scientific Reports, 2017, 7, 4266.	1.6	28
116	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	1.5	28
117	Failure of sucrose replacement with the non-nutritive sweetener erythritol to alter GLP-1 or PYY release or test meal size in lean or obese people. Appetite, 2016, 107, 596-603.	1.8	26
118	Hypothalamic Reproductive Endocrine Pulse Generator Activity Independent of Neurokinin B and Dynorphin Signaling. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4304-4318.	1.8	26
119	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	1.1	26
120	A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. European Journal of Medical Genetics, 2015, 58, 387-391.	0.7	25
121	Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature. , 2017, 58, 594.		25
122	ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605.	1.5	25
123	The severely obese patientâ€”a genetic work-up. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 172-177.	2.9	24
124	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	24
125	Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15.	0.9	24
126	ProxECAT: Proxy External Controls Association Test. A new case-control gene region association test using allele frequencies from public controls. PLoS Genetics, 2018, 14, e1007591.	1.5	23

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127	Insights from the Genetics of Severe Childhood Obesity. Hormone Research in Paediatrics, 2007, 68, 5-7.	0.8	22
128	Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. PLoS Genetics, 2020, 16, e1008916.	1.5	22
129	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	1.1	22
130	A recurrent de novo mutation inACTG1causes isolated ocular coloboma. Human Mutation, 2017, 38, 942-946.	1.1	21
131	NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. Pediatrics, 2018, 141, S485-S490.	1.0	21
132	Quantitative mass spectrometry for human melanocortin peptides inÂvitro and inÂvivo suggests prominent roles for Î²-MSH and desacetyl Î±-MSH in energy homeostasis. Molecular Metabolism, 2018, 17, 82-97.	3.0	21
133	Genotypeâ€“phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. American Journal of Medical Genetics, Part A, 2017, 173, 1566-1574.	0.7	20
134	Leptin-Mediated Changes in the Human Metabolome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2541-2552.	1.8	20
135	The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. Sleep, 2016, 39, 1691-1700.	0.6	19
136	Neural deletion of Sh2b1 results in brain growth retardation and reactive aggression. FASEB Journal, 2018, 32, 1830-1840.	0.2	19
137	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	0.7	18
138	Monogenic human obesity syndromes. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 181, 301-310.	1.0	18
139	CorneliaÂde Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. Nature Communications, 2021, 12, 3127.	5.8	18
140	Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. Diabetes, 2019, 68, 2049-2062.	0.3	16
141	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	1.1	16
142	Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996.	1.4	15
143	Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. Molecular Metabolism, 2017, 6, 1419-1428.	3.0	15
144	Potential role of gender specific effect of leptin receptor deficiency in an extended consanguineous family with severe early-onset obesity. European Journal of Medical Genetics, 2018, 61, 465-467.	0.7	15

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145	Monogenic Obesity Syndromes Provide Insights Into the Hypothalamic Regulation of Appetite and Associated Behaviors. Biological Psychiatry, 2022, 91, 856-859.	0.7	15
146	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
147	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194.	2.6	12
148	Central obesity is selectively associated with cerebral gray matter atrophy in 15,634 subjects in the UK Biobank. International Journal of Obesity, 2022, 46, 1059-1067.	1.6	12
149	Carey-Fineman-Ziter (CFZ) syndrome: Report on affected sibs. , 1999, 82, 110-113.		11
150	A Trans-Acting Protein Effect Causes Severe Eye Malformation in the Mp Mouse. PLoS Genetics, 2013, 9, e1003998.	1.5	11
151	The therapeutic value of somatostatin and its analogues. Pituitary, 1999, 2, 79-88.	1.6	10
152	Obesity Genesâ€"It's All About the Parents!. Cell Metabolism, 2009, 9, 487-488.	7.2	10
153	FTO and Obesity: The Missing Link. Cell Metabolism, 2011, 13, 7-8.	7.2	10
154	Postprandial Total Ghrelin Suppression Is Modulated by Melanocortin Signaling in Humans. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E288-E292.	1.8	10
155	Characterization of human variants in obesity-related SIM1 protein identifies a hot-spot for dimerization with the partner protein ARNT2. Biochemical Journal, 2014, 461, 403-412.	1.7	10
156	Neural networks associated with body composition in frontotemporal dementia. Annals of Clinical and Translational Neurology, 2019, 6, 1707-1717.	1.7	10
157	Genetic aspects of severe childhood obesity. Pediatric Endocrinology Reviews, 2006, 3 Suppl 4, 528-36.	1.2	10
158	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	2.6	8
159	Treating obesity: Does antagonism of NPY fit the bill?. Cell Metabolism, 2006, 4, 260-262.	7.2	7
160	Genetic strategies to understand physiological pathways regulating body weight. Mammalian Genome, 2014, 25, 377-383.	1.0	7
161	A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476.	0.4	7
162	Status dystonicus in two patients with SOX2â€anophthalmia syndrome and nonsense mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3048-3050.	0.7	7

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163	Predicting novel candidate human obesity genes and their site of action by systematic functional screening in Drosophila. PLoS Biology, 2021, 19, e3001255.	2.6	7
164	Obesity due to melanocortin 4 receptor (MC4R) deficiency is associated with delayed gastric emptying. Clinical Endocrinology, 2022, 96, 270-275.	1.2	6
165	EJE PRIZE 2012: Obesity: from genes to behaviour. European Journal of Endocrinology, 2014, 171, R191-R195.	1.9	5
166	Murine neuronatin deficiency is associated with a hypervariable food intake and bimodal obesity. Scientific Reports, 2021, 11, 17571.	1.6	5
167	Obesity Due to Steroid Receptor Coactivator-1 Deficiency Is Associated With Endocrine and Metabolic Abnormalities. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2532-e2544.	1.8	5
168	Neural correlates of fat preference in frontotemporal dementia: translating insights from the obesity literature. Annals of Clinical and Translational Neurology, 2021, 8, 1318-1329.	1.7	4
169	Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. Genome Research, 2021, 31, 1994-2007.	2.4	4
170	Resequencing at scale in neurodevelopmental disorders. Nature Genetics, 2017, 49, 488-489.	9.4	3
171	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	2.5	3
172	Reply to Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. Cell, 2021, 184, 1652-1653.	13.5	3
173	Identification and functional modelling of plausibly causative cis-regulatory variants in a highly-selected cohort with X-linked intellectual disability. PLoS ONE, 2021, 16, e0256181.	1.1	3
174	Triple H Syndrome: A Novel Autoimmune Endocrinopathy Characterized by Dysfunction of the Hippocampus, Hair Follicle, and Hypothalamic-Pituitary-Adrenal Axis. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 2644-2648.	1.8	3
175	Wired for Obesity?. Diabetes, 2014, 63, 4016-4017.	0.3	2
176	Candidate Genes for Obesity â€" How Might They Interact with Environment and Diet ?. , 2005, 569, 33-34.		2
177	Visualization of sympathetic neural innervation in human white adipose tissue. Open Biology, 2022, 12, 210345.	1.5	2
178	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, e1.18-e1.	0.9	0
179	Putting a brake on hunger. Science, 2021, 372, 792-793.	6.0	0
180	Abstract 18543: Whole Exome Sequencing in Sudden Infant Death Syndrome Identifies a High Proportion of Putative Pathogenic and Functionally Significant Rare Variants Related to Inherited Cardiac Conditions. Circulation, 2014, 130, .	1.6	0