

Sadaf Farooqi

List of Publications by Year in descending order

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Version: 2024-02-01

179
papers

30,196
citations

12303

69
h-index

5101

166
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. <i>Nature</i> , 1997, 387, 903-908.	13.7	2,664
2	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , 2010, 42, 937-948.	9.4	2,634
3	Effects of Recombinant Leptin Therapy in a Child with Congenital Leptin Deficiency. <i>New England Journal of Medicine</i> , 1999, 341, 879-884.	13.9	1,760
4	Clinical Spectrum of Obesity and Mutations in the Melanocortin 4 Receptor Gene. <i>New England Journal of Medicine</i> , 2003, 348, 1085-1095.	13.9	1,475
5	A frameshift mutation in MC4R associated with dominantly inherited human obesity. <i>Nature Genetics</i> , 1998, 20, 111-112.	9.4	1,026
6	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 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. <i>Journal of Clinical Investigation</i> , 2002, 110, 1093-1103.	3.9	953
8	Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. <i>Journal of Clinical Investigation</i> , 2000, 106, 271-279.	3.9	696
9	Genetics of body-weight regulation. <i>Nature</i> , 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. <i>Journal of Clinical Investigation</i> , 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. <i>Lancet, The</i> , 2015, 385, 1305-1314.	6.3	651
12	Clinical and Molecular Genetic Spectrum of Congenital Deficiency of the Leptin Receptor. <i>New England Journal of Medicine</i> , 2007, 356, 237-247.	13.9	610
13	Leptin Regulates Striatal Regions and Human Eating Behavior. <i>Science</i> , 2007, 317, 1355-1355.	6.0	541
14	Large, rare chromosomal deletions associated with severe early-onset obesity. <i>Nature</i> , 2010, 463, 666-670.	13.7	487
15	Genetics of Obesity in Humans. <i>Endocrine Reviews</i> , 2006, 27, 710-718.	8.9	452
16	Modulation of Blood Pressure by Central Melanocortineric Pathways. <i>New England Journal of Medicine</i> , 2009, 360, 44-52.	13.9	412
17	Paediatric genomics: diagnosing rare disease in children. <i>Nature Reviews Genetics</i> , 2018, 19, 253-268.	7.7	369
18	Monogenic Obesity in Humans. <i>Annual Review of Medicine</i> , 2005, 56, 443-458.	5.0	367

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19	Partial leptin deficiency and human adiposity. <i>Nature</i> , 2001, 414, 34-35.	13.7	356
20	A genome-wide association meta-analysis identifies new childhood obesity loci. <i>Nature Genetics</i> , 2012, 44, 526-531.	9.4	352
21	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. <i>Nature Genetics</i> , 2016, 48, 1060-1065.	9.4	351
22	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	13.7	343
23	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
24	The Hunger Genes: Pathways to Obesity. <i>Cell</i> , 2015, 161, 119-132.	13.5	293
25	Leptin Mediates the Increase in Blood Pressure Associated with Obesity. <i>Cell</i> , 2014, 159, 1404-1416.	13.5	288
26	GDF15 Provides an Endocrine Signal of Nutritional Stress in Mice and Humans. <i>Cell Metabolism</i> , 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. <i>Nature Genetics</i> , 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. <i>Nature Genetics</i> , 2013, 45, 513-517.	9.4	278
29	Leptin: a pivotal regulator of human energy homeostasis. <i>American Journal of Clinical Nutrition</i> , 2009, 89, 980S-984S.	2.2	261
30	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
31	Identification of SATB2 as the cleft palate gene on 2q32-q33. <i>Human Molecular Genetics</i> , 2003, 12, 2491-2501.	1.4	248
32	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. <i>Nature</i> , 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. <i>Lancet Diabetes and Endocrinology</i> , 2020, 8, 960-970.	5.5	235
34	Prevalence of Melanocortin-4 Receptor Deficiency in Europeans and Their Age-Dependent Penetrance in Multigenerational Pedigrees. <i>Diabetes</i> , 2008, 57, 2511-2518.	0.3	229
35	Mutations in ligands and receptors of the leptin-melanocortin pathway that lead to obesity. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , 2008, 4, 569-577.	2.9	225
36	Loss of Function of the Melanocortin 2 Receptor Accessory Protein 2 Is Associated with Mammalian Obesity. <i>Science</i> , 2013, 341, 275-278.	6.0	225

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37	MC4R agonism promotes durable weight loss in patients with leptin receptor deficiency. <i>Nature Medicine</i> , 2018, 24, 551-555.	15.2	219
38	20 YEARS OF LEPTIN: Human disorders of leptin action. <i>Journal of Endocrinology</i> , 2014, 223, T63-T70.	1.2	218
39	The genetic architecture of microphthalmia, anophthalmia and coloboma. <i>European Journal of Medical Genetics</i> , 2014, 57, 369-380.	0.7	213
40	Heterozygosity for aPOMC-Null Mutation and Increased Obesity Risk in Humans. <i>Diabetes</i> , 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. <i>American Journal of Clinical Nutrition</i> , 2009, 89, 106-113.	2.2	201
42	Evaluation of a melanocortin-4 receptor (MC4R) agonist (Setmelanotide) in MC4R deficiency. <i>Molecular Metabolism</i> , 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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 3369-3373.	1.8	196
44	Human Gain-of-Function MC4R Variants Show Signaling Bias and Protect against Obesity. <i>Cell</i> , 2019, 177, 597-607.e9.	13.5	192
45	Human Obesity: A Heritable Neurobehavioral Disorder That Is Highly Sensitive to Environmental Conditions. <i>Diabetes</i> , 2008, 57, 2905-2910.	0.3	160
46	Quantifying the contribution of recessive coding variation to developmental disorders. <i>Science</i> , 2018, 362, 1161-1164.	6.0	158
47	KSR2 Mutations Are Associated with Obesity, Insulin Resistance, and Impaired Cellular Fuel Oxidation. <i>Cell</i> , 2013, 155, 765-777.	13.5	154
48	Transcriptome analysis of human autosomal trisomy. <i>Human Molecular Genetics</i> , 2002, 11, 3249-3256.	1.4	150
49	Human SH2B1 mutations are associated with maladaptive behaviors and obesity. <i>Journal of Clinical Investigation</i> , 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. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	1.5	141
51	Rare variants in single-minded 1 (SIM1) are associated with severe obesity. <i>Journal of Clinical Investigation</i> , 2013, 123, 3042-3050.	3.9	135
52	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. <i>Nature Genetics</i> , 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. <i>Nature Genetics</i> , 2017, 49, 238-248.	9.4	131
54	Diagnostically relevant facial gestalt information from ordinary photos. <i>ELife</i> , 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. <i>Lancet Neurology</i> , The, 2016, 15, 332-342.	4.9	120
56	Leptin and the Onset of Puberty: Insights from Rodent and Human Genetics. <i>Seminars in Reproductive Medicine</i> , 2002, 20, 139-144.	0.5	117
57	Food addiction: is there a baby in the bathwater?. <i>Nature Reviews Neuroscience</i> , 2012, 13, 514-514.	4.9	102
58	Genetic architecture of human thinness compared to severe obesity. <i>PLoS Genetics</i> , 2019, 15, e1007603.	1.5	98
59	De novo, heterozygous, loss-of-function mutations in <i>SYNGAP1</i> cause a syndromic form of intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2231-2237.	0.7	96
60	BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Lange-like syndrome. <i>Nature Genetics</i> , 2018, 50, 329-332.	9.4	96
61	Developmental eye disorders. <i>Current Opinion in Genetics and Development</i> , 2005, 15, 348-353.	1.5	95
62	Genetic and hereditary aspects of childhood obesity. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2005, 19, 359-374.	2.2	94
63	Heterozygous Loss-of-Function Mutations in <i>YAP1</i> Cause Both Isolated and Syndromic Optic Fissure Closure Defects. <i>American Journal of Human Genetics</i> , 2014, 94, 295-302.	2.6	93
64	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. <i>Nature Communications</i> , 2019, 10, 2373.	5.8	86
65	Monogenic Human Obesity. , 2008, 36, 1-11.		84
66	A Restricted Repertoire of De Novo Mutations in <i>ITPR1</i> Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. <i>American Journal of Human Genetics</i> , 2016, 98, 981-992.	2.6	81
67	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. <i>Cell Reports</i> , 2018, 22, 3401-3408.	2.9	81
68	Human Semaphorin 3 Variants Link Melanocortin Circuit Development and Energy Balance. <i>Cell</i> , 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. <i>Biological Psychiatry</i> , 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. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 15-31.	0.6	79
71	Leptin Regulates Peripheral Lipid Metabolism Primarily through Central Effects on Food Intake. <i>Endocrinology</i> , 2008, 149, 5432-5439.	1.4	78
72	Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. <i>Trends in Genetics</i> , 2005, 21, 249-253.	2.9	77

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73	Assessment of Eating Behavior Disturbance and Associated Neural Networks in Frontotemporal Dementia. <i>JAMA Neurology</i> , 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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 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. <i>Journal of Neuroscience</i> , 2010, 30, 14346-14355.	1.7	69
76	Cardiac Genetic Predisposition in Sudden Infant Death Syndrome. <i>Journal of the American College of Cardiology</i> , 2018, 71, 1217-1227.	1.2	66
77	A Locus for Isolated Cleft Palate, Located on Human Chromosome 2q32. <i>American Journal of Human Genetics</i> , 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. <i>Lancet</i> , The, 2018, 391, 1483-1492.	6.3	63
79	A New Drug Target for Type 2 Diabetes. <i>Cell</i> , 2017, 170, 12-14.	13.5	62
80	<i>KMT2B</i>-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. <i>Brain</i> , 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. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56
82	Ambulatory Blood Pressure Monitoring in Acute Stroke. <i>Stroke</i> , 1997, 28, 31-35.	1.0	56
83	The Gene Curation Coalition: A global effort to harmonize gene disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	1.1	56
84	Genetic Analysis of "PAX6-Negative"™ Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016, 11, e0153757.	1.1	54
85	A Metabolomic Signature of Acute Caloric Restriction. <i>Journal of Clinical Endocrinology and Metabolism</i> , 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. <i>Human Molecular Genetics</i> , 2014, 23, 2569-2579.	1.4	51
87	The genetic architecture of aniridia and Gillespie syndrome. <i>Human Genetics</i> , 2019, 138, 881-898.	1.8	51
88	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. <i>Scientific Reports</i> , 2017, 7, 4394.	1.6	50
89	Melanocortin Receptors as Targets in the Treatment of Obesity. <i>Current Topics in Medicinal Chemistry</i> , 2007, 7, 1098-1110.	1.0	47
90	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.	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. <i>Journal of Alzheimer's Disease</i> , 2017, 61, 773-783.	1.2	47
92	Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. <i>Open Biology</i> , 2015, 5, 150047.	1.5	46
93	Divergent effects of central melanocortin signalling on fat and sucrose preference in humans. <i>Nature Communications</i> , 2016, 7, 13055.	5.8	46
94	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. <i>Genetics in Medicine</i> , 2017, 19, 900-908.	1.1	46
95	Steroid receptor coactivator-1 modulates the function of Pomc neurons and energy homeostasis. <i>Nature Communications</i> , 2019, 10, 1718.	5.8	45
96	ob gene mutations and human obesity. <i>Proceedings of the Nutrition Society</i> , 1998, 57, 471-475.	0.4	44
97	Obesity-Associated <i>GNAS</i> Mutations and the Melanocortin Pathway. <i>New England Journal of Medicine</i> , 2021, 385, 1581-1592.	13.9	44
98	Energy expenditure in frontotemporal dementia: a behavioural and imaging study. <i>Brain</i> , 2017, 140, 171-183.	3.7	43
99	Contribution of retrotransposition to developmental disorders. <i>Nature Communications</i> , 2019, 10, 4630.	5.8	43
100	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.	1.1	43
101	Genetic, molecular and physiological insights into human obesity. <i>European Journal of Clinical Investigation</i> , 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. <i>European Journal of Human Genetics</i> , 2017, 25, 552-559.	1.4	42
103	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. <i>PLoS Genetics</i> , 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. <i>RNA Biology</i> , 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. <i>Human Molecular Genetics</i> , 2016, 25, 1382-1391.	1.4	40
106	Human BDNF/TrkB variants impair hippocampal synaptogenesis and associate with neurobehavioural abnormalities. <i>Scientific Reports</i> , 2020, 10, 9028.	1.6	40
107	Defining the neural basis of appetite and obesity: from genes to behaviour. <i>Clinical Medicine</i> , 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. <i>Cell Metabolism</i> , 2020, 31, 1107-1119.e12.	7.2	38

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109	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	1.4	37
110	Human MC4R variants affect endocytosis, trafficking and dimerization revealing multiple cellular mechanisms involved in weight regulation. <i>Cell Reports</i> , 2021, 34, 108862.	2.9	37
111	The orphan G protein-coupled receptor GPR139 is activated by the peptides: Adrenocorticotrophic hormone (ACTH), I^{\pm} , and I^2 -melanocyte stimulating hormone (I^{\pm} -MSH, and I^2 -MSH), and the conserved core motif HFRW. <i>Neurochemistry International</i> , 2017, 102, 105-113.	1.9	36
112	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. <i>Genome Research</i> , 2019, 29, 1047-1056.	2.4	34
113	The contribution of X-linked coding variation to severe developmental disorders. <i>Nature Communications</i> , 2021, 12, 627.	5.8	33
114	Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. <i>Nature Communications</i> , 2015, 6, 6904.	5.8	31
115	Oxytocin administration suppresses hypothalamic activation in response to visual food cues. <i>Scientific Reports</i> , 2017, 7, 4266.	1.6	28
116	<i>NAA10</i> polyadenylation signal variants cause syndromic microphthalmia. <i>Journal of Medical Genetics</i> , 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. <i>Appetite</i> , 2016, 107, 596-603.	1.8	26
118	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.	1.8	26
119	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26
120	A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. <i>European Journal of Medical Genetics</i> , 2015, 58, 387-391.	0.7	25
121	Novel <i>PEX11B</i> 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. <i>PLoS Genetics</i> , 2019, 15, e1007605.	1.5	25
123	The severely obese patientâ€™s a genetic work-up. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , 2006, 2, 172-177.	2.9	24
124	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. <i>Human Genetics</i> , 2020, 139, 575-592.	1.8	24
125	Returning genome sequences to research participants: Policy and practice. <i>Wellcome Open Research</i> , 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. <i>PLoS Genetics</i> , 2018, 14, e1007591.	1.5	23

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127	Insights from the Genetics of Severe Childhood Obesity. <i>Hormone Research in Paediatrics</i> , 2007, 68, 5-7.	0.8	22
128	Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. <i>PLoS Genetics</i> , 2020, 16, e1008916.	1.5	22
129	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. <i>Genetics in Medicine</i> , 2020, 22, 878-888.	1.1	22
130	A recurrent de novo mutation in <i>ACTG1</i> causes isolated ocular coloboma. <i>Human Mutation</i> , 2017, 38, 942-946.	1.1	21
131	NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. <i>Pediatrics</i> , 2018, 141, S485-S490.	1.0	21
132	Quantitative mass spectrometry for human melanocortin peptides in vitro and in vivo suggests prominent roles for I ² -MSH and desacetyl I ¹ -MSH in energy homeostasis. <i>Molecular Metabolism</i> , 2018, 17, 82-97.	3.0	21
133	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.	0.7	20
134	Leptin-Mediated Changes in the Human Metabolome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2541-2552.	1.8	20
135	The Sleep/Wake Cycle is Directly Modulated by Changes in Energy Balance. <i>Sleep</i> , 2016, 39, 1691-1700.	0.6	19
136	Neural deletion of <i>Sh2b1</i> results in brain growth retardation and reactive aggression. <i>FASEB Journal</i> , 2018, 32, 1830-1840.	0.2	19
137	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, 587-595.	0.7	18
138	Monogenic human obesity syndromes. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2021, 181, 301-310.	1.0	18
139	Cornelia de Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. <i>Nature Communications</i> , 2021, 12, 3127.	5.8	18
140	Crucial Role of the SH2B1 PH Domain for the Control of Energy Balance. <i>Diabetes</i> , 2019, 68, 2049-2062.	0.3	16
141	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. <i>Genetics in Medicine</i> , 2021, 23, 571-575.	1.1	16
142	Expansion of Ocular Phenotypic Features Associated With Mutations in <i>ADAMTS18</i> . <i>JAMA Ophthalmology</i> , 2014, 132, 996.	1.4	15
143	Disruption of the homeodomain transcription factor orthopedia homeobox (Otp) is associated with obesity and anxiety. <i>Molecular Metabolism</i> , 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. <i>European Journal of Medical Genetics</i> , 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. <i>Biological Psychiatry</i> , 2022, 91, 856-859.	0.7	15
146	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
147	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.	2.6	12
148	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, 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. <i>PLoS Genetics</i> , 2013, 9, e1003998.	1.5	11
151	The therapeutic value of somatostatin and its analogues. <i>Pituitary</i> , 1999, 2, 79-88.	1.6	10
152	Obesity Genesâ€”It's All About the Parents!. <i>Cell Metabolism</i> , 2009, 9, 487-488.	7.2	10
153	FTO and Obesity: The Missing Link. <i>Cell Metabolism</i> , 2011, 13, 7-8.	7.2	10
154	Postprandial Total Ghrelin Suppression Is Modulated by Melanocortin Signaling in Humans. <i>Journal of Clinical Endocrinology and Metabolism</i> , 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. <i>Biochemical Journal</i> , 2014, 461, 403-412.	1.7	10
156	Neural networks associated with body composition in frontotemporal dementia. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1707-1717.	1.7	10
157	Genetic aspects of severe childhood obesity. <i>Pediatric Endocrinology Reviews</i> , 2006, 3 Suppl 4, 528-36.	1.2	10
158	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. <i>American Journal of Human Genetics</i> , 2019, 105, 933-946.	2.6	8
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