## Murat Günel

List of Publications by Year in descending order

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133 11,737 51 103
papers citations h-index g-index

137 137 20977
all docs docs citations times ranked citing authors

#	Article	IF	CITATIONS
1	Human Hypertension Caused by Mutations in WNK Kinases. Science, 2001, 293, 1107-1112.	12.6	1,344
2	Genomic Analysis of Non- <i>NF2</i> Meningiomas Reveals Mutations in <itraf7< i=""> , <i>KLF4</i> , <i>AKT1</i> , and <i>SMO</i> . Science, 2013, 339, 1077-1080.</itraf7<>	12.6	714
3	Neuroinvasion of SARS-CoV-2 in human and mouse brain. Journal of Experimental Medicine, 2021, 218, .	8.5	677
4	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
5	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	12.6	466
6	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	27.8	457
7	2-Hydroxyglutarate produced by neomorphic IDH mutations suppresses homologous recombination and induces PARP inhibitor sensitivity. Science Translational Medicine, 2017, 9, .	12.4	420
8	Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder. Cell, 2016, 167, 1481-1494.e18.	28.9	265
9	Recurrent somatic mutations in POLR2A define a distinct subset of meningiomas. Nature Genetics, 2016, 48, 1253-1259.	21.4	265
10	Genome-wide association study of intracranial aneurysm identifies three new risk loci. Nature Genetics, 2010, 42, 420-425.	21.4	262
11	Inflammation-dependent cerebrospinal fluid hypersecretion by the choroid plexus epithelium in posthemorrhagic hydrocephalus. Nature Medicine, 2017, 23, 997-1003.	30.7	256
12	Integrated genomic characterization of IDH1-mutant glioma malignant progression. Nature Genetics, 2016, 48, 59-66.	21.4	253
13	Susceptibility loci for intracranial aneurysm in European and Japanese populations. Nature Genetics, 2008, 40, 1472-1477.	21.4	247
14	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
15	The critical role of hemodynamics in the development of cerebral vascular disease. Journal of Neurosurgery, 2010, 112, 1240-1253.	1.6	197
16	PD-1 marks dysfunctional regulatory T cells in malignant gliomas. JCI Insight, 2016, 1, .	5.0	182
17	Inactivating mutations in MFSD2A, required for omega-3 fatty acid transport in brain, cause a lethal microcephaly syndrome. Nature Genetics, 2015, 47, 809-813.	21.4	180
18	AAV-mediated direct in vivo CRISPR screen identifies functional suppressors in glioblastoma. Nature Neuroscience, 2017, 20, 1329-1341.	14.8	179

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19	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
20	Integrated genomic analyses of de novo pathways underlying atypical meningiomas. Nature Communications, 2017, 8, 14433.	12.8	156
21	The Essential Role of Centrosomal NDE1 in Human Cerebral Cortex Neurogenesis. American Journal of Human Genetics, 2011, 88, 523-535.	6.2	146
22	Augmentor $\hat{l}_{\pm}$ and $\hat{l}_{-}^2$ (FAM150) are ligands of the receptor tyrosine kinases ALK and LTK: Hierarchy and specificity of ligandâ $\in$ "receptor interactions. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 15862-15867.	7.1	125
23	Exceptional aggressiveness of cerebral cavernous malformation disease associated with PDCD10 mutations. Genetics in Medicine, 2015, 17, 188-196.	2.4	116
24	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	8.1	112
25	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
26	Missense mutation in the ATPase, aminophospholipid transporter protein ATP8A2 is associated with cerebellar atrophy and quadrupedal locomotion. European Journal of Human Genetics, 2013, 21, 281-285.	2.8	110
27	<i>KRIT1</i> , a gene mutated in cerebral cavernous malformation, encodes a microtubule-associated protein. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 10677-10682.	7.1	108
28	Recessive LAMC3 mutations cause malformations of occipital cortical development. Nature Genetics, 2011, 43, 590-594.	21.4	102
29	Common variant near the endothelin receptor type A ( <i>EDNRA</i> ) gene is associated with intracranial aneurysm risk. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 19707-19712.	7.1	100
30	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	8.1	95
31	Somatic <i>POLE</i> mutations cause an ultramutated giant cell high-grade glioma subtype with better prognosis. Neuro-Oncology, 2015, 17, 1356-1364.	1.2	94
32	Loss of <i>cerebral cavernous malformation 3</i> ( <i>Ccm3</i> ) in neuroglia leads to CCM and vascular pathology. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 3737-3742.	7.1	92
33	Genetics Of Intracranial Aneurysms. Neurosurgery, 2007, 60, 213-226.	1.1	86
34	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	30.7	84
35	Associations of meningioma molecular subgroup and tumor recurrence. Neuro-Oncology, 2021, 23, 783-794.	1.2	83
36	Correlations between genomic subgroup and clinical features in a cohort of more than 3000 meningiomas. Journal of Neurosurgery, 2020, 133, 1345-1354.	1.6	83

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37	Mapping a Mendelian Form of Intracranial Aneurysm to 1p34.3-p36.13. American Journal of Human Genetics, 2005, 76, 172-179.	6.2	80
38	The distinct genetic pattern of ALS in Turkey and novel mutations. Neurobiology of Aging, 2015, 36, 1764.e9-1764.e18.	3.1	78
39	Seizure control for intracranial arteriovenous malformations is directly related to treatment modality: a meta-analysis. Journal of NeuroInterventional Surgery, 2014, 6, 684-690.	3.3	75
40	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	6.2	75
41	<i>ACOX2</i> deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 11289-11293.	7.1	75
42	Heparin is an activating ligand of the orphan receptor tyrosine kinase ALK. Science Signaling, 2015, 8, ra6.	3.6	72
43	Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. American Journal of Human Genetics, 2016, 99, 501-510.	6.2	70
44	Biallelic loss of human CTNNA2, encoding $\hat{l}\pm N$ -catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
45	NGLY1 mutation causes neuromotor impairment, intellectual disability, and neuropathy. European Journal of Medical Genetics, 2015, 58, 39-43.	1.3	69
46	Mutational analysis of 206 families with cavernous malformations. Journal of Neurosurgery, 2003, 99, 38-43.	1.6	66
47	Biallelic mutations in the $3\hat{a}\in^2$ exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	21.4	66
48	Homozygosity mapping and targeted genomic sequencing reveal the gene responsible for cerebellar hypoplasia and quadrupedal locomotion in a consanguineous kindred. Genome Research, 2011, 21, 1995-2003.	5.5	62
49	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. European Journal of Human Genetics, 2015, 23, 1482-1487.	2.8	62
50	Molecular Genetic Analysis of Two Large Kindreds With Intracranial Aneurysms Demonstrates Linkage to 11q24-25 and 14q23-31. Stroke, 2006, 37, 1021-1027.	2.0	58
51	Wholeâ€exome sequencing defines the mutational landscape of pheochromocytoma and identifies KMT 2 D as a recurrently mutated gene. Genes Chromosomes and Cancer, 2015, 54, 542-554.	2.8	57
52	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. European Journal of Human Genetics, 2015, 23, 165-172.	2.8	57
53	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	8.1	56
54	High Risk Population Isolate Reveals Low Frequency Variants Predisposing to Intracranial Aneurysms. PLoS Genetics, 2014, 10, e1004134.	3.5	55

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55	Autosomal recessive spastic tetraplegia caused by <i>AP4M1</i> and <i>AP4B1</i> gene mutation: Expansion of the facial and neuroimaging features. American Journal of Medical Genetics, Part A, 2014, 1677-1685.	1.2	55
56	Paediatric hepatocellular carcinoma due to somatic CTNNB1 and NFE2L2 mutations in the setting of inherited bi-allelic ABCB11 mutations. Journal of Hepatology, 2014, 61, 1178-1183.	3.7	48
57	Recurrent recessive mutation in deoxyguanosine kinase causes idiopathic noncirrhotic portal hypertension. Hepatology, 2016, 63, 1977-1986.	<b>7.</b> 3	46
58	Brain Malformations Associated With Knobloch Syndromeâ€"Review of Literature, Expanding Clinical Spectrum, and Identification of Novel Mutations. Pediatric Neurology, 2014, 51, 806-813.e8.	2.1	43
59	The genetic structure of the Turkish population reveals high levels of variation and admixture. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	42
60	A congenital disorder of deglycosylation: Biochemical characterization of <i>N</i> glycanase 1 deficiency in patient fibroblasts. Glycobiology, 2015, 25, 836-844.	2.5	40
61	B-Cell Depletion Reduces the Maturation of Cerebral Cavernous Malformations in Murine Models. Journal of NeuroImmune Pharmacology, 2016, 11, 369-377.	4.1	39
62	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
63	Disruptions in asymmetric centrosome inheritance and WDR62-Aurora kinase B interactions in primary microcephaly. Scientific Reports, 2017, 7, 43708.	3.3	37
64	Genetically Elevated <scp>LDL</scp> Associates with Lower Risk of Intracerebral Hemorrhage. Annals of Neurology, 2020, 88, 56-66.	5.3	35
65	Functional Synergy between Cholecystokinin Receptors CCKAR and CCKBR in Mammalian Brain Development. PLoS ONE, 2015, 10, e0124295.	2.5	34
66	Functional differences between PD-1+ and PD-1- CD4+ effector T cells in healthy donors and patients with glioblastoma multiforme. PLoS ONE, 2017, 12, e0181538.	2.5	34
67	Targeting the CSF1/CSF1R axis is a potential treatment strategy for malignant meningiomas. Neuro-Oncology, 2021, 23, 1922-1935.	1.2	33
68	<i>DIAPH1</i> Variants in Non–East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	9.0	33
69	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	4.1	32
70	Clinical characteristics and outcomes for 7,995 patients with SARS-CoV-2 infection. PLoS ONE, 2021, 16, e0243291.	2.5	31
71	<i>Ccm3</i> , a gene associated with cerebral cavernous malformations, is required for neuronal migration. Development (Cambridge), 2014, 141, 1404-1415.	2.5	30
72	Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly. American Journal of Human Genetics, 2016, 99, 1181-1189.	6.2	30

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73	FBXO7–R498X mutation: Phenotypic variability from chorea to early onset parkinsonism within a family. Parkinsonism and Related Disorders, 2014, 20, 1253-1256.	2.2	29
74	Use of telomerase promoter mutations to mark specific molecular subsets with reciprocal clinical behavior in IDH mutant and IDH wild-type diffuse gliomas. Journal of Neurosurgery, 2018, 128, 1102-1114.	1.6	26
75	A novel heterozygous deletion within the 3' region of the PAX6 gene causing isolated aniridia in a large family group. Journal of Clinical Neuroscience, 2009, 16, 1610-1614.	1.5	25
76	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive <i>c</i> erebellar, <i>o</i> cular, cranio <i>f</i> acial and <i>g</i> enital features (COFG) Tj ETQq0 0 0 rg	BT <b>\$Q</b> verlo	ck <b>2150</b> Tf 50 61
77	Combined HMG-COA reductase and prenylation inhibition in treatment of CCM. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 5503-5508.	7.1	24
78	PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans. Nature Medicine, 2021, 27, 2165-2175.	30.7	23
79	Longitudinal analysis of treatment-induced genomic alterations in gliomas. Genome Medicine, 2017, 9, 12.	8.2	20
80	ALPK3 gene mutation in a patient with congenital cardiomyopathy and dysmorphic features. Journal of Physical Education and Sports Management, 2017, 3, a001859.	1.2	20
81	Novel VLDLR microdeletion identified in two Turkish siblings with pachygyria and pontocerebellar atrophy. Neurogenetics, 2010, 11, 319-325.	1.4	19
82	Wholeâ€exome sequencing identified a patient with TMCO1 defect syndrome and expands the phenotic spectrum. Clinical Genetics, 2013, 84, 394-395.	2.0	19
83	Loss of <i>Protocadherinâ€12</i> <scp>L</scp> eads to <scp>D</scp> iencephalicâ€ <scp>M</scp> esencephalic <scp>J</scp> unction <scp>D</scp> ysplasia <scp>S</scp> yndrome. Annals of Neurology, 2018, 84, 638-647.	5.3	19
84	Results of a national cerebrovascular neurosurgery survey on the management of cerebral vasospasm/delayed cerebral ischemia. Journal of NeuroInterventional Surgery, 2015, 7, 408-411.	3.3	18
85	Clinical and genomic factors associated with seizures in meningiomas. Journal of Neurosurgery, 2021, 135, 835-844.	1.6	17
86	Familial occurrence of brain arteriovenous malformation: a novel ACVRL1 mutation detected by whole exome sequencing. Journal of Neurosurgery, 2016, 126, 1879-1883.	1.6	16
87	Genotype–phenotype investigation of 35 patients from 11 unrelated families with camptodactyly–arthropathy–coxa vara–pericarditis ( <scp>CACP</scp> ) syndrome. Molecular Genetics & Genomic Medicine, 2018, 6, 230-248.	1.2	15
88	Hypertension, Age, and Location Predict Rupture of Small Intracranial Aneurysms. Neurosurgery, 2005, 57, 676-683.	1.1	15
89	Apparently novel genetic syndrome of pachygyria, mental retardation, seizure, and arachnoid cysts. American Journal of Medical Genetics, Part A, 2007, 143A, 672-677.	1.2	14
90	Novel NTRK1 mutations cause hereditary sensory and autonomic neuropathy type IV: demonstration of a founder mutation in the Turkish population. Neurogenetics, 2008, 9, 119-125.	1.4	14

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91	A patient with a novel homozygous missense mutation in FTO and concomitant nonsense mutation in CETP. Journal of Human Genetics, 2016, 61, 395-403.	2.3	14
92	Recessive Inheritance of Congenital Hydrocephalus With Other Structural Brain Abnormalities Caused by Compound Heterozygous Mutations in ATP1A3. Frontiers in Cellular Neuroscience, 2019, 13, 425.	3.7	14
93	Counting strokes. Nature Genetics, 1996, 13, 384-385.	21.4	13
94	A quantitative model based on clinically relevant MRI features differentiates lower grade gliomas and glioblastoma. European Radiology, 2020, 30, 3073-3082.	4.5	13
95	Rapid identification of disease-causing mutations using copy number analysis within linkage intervals. Human Mutation, 2007, 28, 1236-1240.	2.5	12
96	Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus. World Neurosurgery, 2018, 119, 441-443.	1.3	12
97	The integrated multiomic diagnosis of sporadic meningiomas: a review of its clinical implications. Journal of Neuro-Oncology, 2022, 156, 205-214.	2.9	12
98	Type of bony involvement predicts genomic subgroup in sphenoid wing meningiomas. Journal of Neuro-Oncology, 2021, 154, 237-246.	2.9	11
99	De novo $\langle i \rangle$ MYH9 $\langle li \rangle$ mutation in congenital scalp hemangioma. Journal of Physical Education and Sports Management, 2018, 4, a002998.	1.2	9
100	Exome sequencing identifies SLIT2 variants in primary CNS lymphoma. British Journal of Haematology, 2021, 193, 375-379.	2.5	9
101	Spatially Resolved and Quantitative Analysis of the Immunological Landscape in Human Meningiomas. Journal of Neuropathology and Experimental Neurology, 2021, 80, 150-159.	1.7	9
102	The syndrome of pachygyria, mental retardation, and arachnoid cysts maps to 11p15. American Journal of Medical Genetics, Part A, 2009, 149A, 2569-2572.	1.2	8
103	A patient with Duchenne muscular dystrophy and autism demonstrates a hemizygous deletion affecting <i>Dystrophin</i> . American Journal of Medical Genetics, Part A, 2010, 152A, 1039-1042.	1.2	8
104	Digenic mutations of human OCRL paralogs in Dentâ $\in$ <sup>TM</sup> s disease type 2 associated with Chiari I malformation. Human Genome Variation, 2016, 3, 16042.	0.7	8
105	Novel compound heterozygous mutations in <i>GPT2</i> linked to microcephaly, and intellectual developmental disability with or without spastic paraplegia. American Journal of Medical Genetics, Part A, 2018, 176, 421-425.	1.2	8
106	9p24 triplication in syndromic hydrocephalus with diffuse villous hyperplasia of the choroid plexus. Journal of Physical Education and Sports Management, 2018, 4, a003145.	1.2	8
107	Exome analysis of the evolutionary path of hepatocellular adenoma-carcinoma transition, vascular invasion and brain dissemination. Journal of Hepatology, 2017, 67, 186-191.	3.7	7
108	Molecular genetics of meningiomas. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2020, 169, 101-119.	1.8	5

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109	Renal involvement in patients with mucolipidosis Illalpha/beta: Causal relation or coâ€occurrence?. American Journal of Medical Genetics, Part A, 2016, 170, 1187-1195.	1.2	4
110	Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Enomic Medicine, 2022, 10, e1944.	1.2	4
111	Biallelic BICD2 variant is a novel candidate for Cohen-like syndrome. Journal of Human Genetics, 2022, 67, 553-556.	2.3	3
112	Genomic profiling of sporadic multiple meningiomas. BMC Medical Genomics, 2022, 15, 112.	1.5	3
113	Extraction of Fronto-orbital Shower Hook through Transcranial Orbitotomy. Craniomaxillofacial Trauma & Reconstruction, 2014, 7, 147-148.	1.3	2
114	Personalized Medicine Through Advanced Genomics. , 2017, , 31-48.		1
115	Genomic alterations in Turcot syndrome: Insights from whole exome sequencing. Journal of the Neurological Sciences, 2020, 417, 117056.	0.6	1
116	A Quantitative Assessment of Pre-Operative MRI Reports in Glioma Patients: Report Metrics and IDH Prediction Ability. Frontiers in Oncology, 2020, 10, 600327.	2.8	1
117	Genetically Determined Lowâ€Density Lipoprotein Cholesterol and Risk of Subarachnoid Hemorrhage. Annals of Neurology, 2021, , .	<b>5.</b> 3	1
118	The Genomic Landscape of Meningiomas. , 2020, , 35-55.		1
119	NCOG-50. CLINICAL AND GENOMIC FACTORS ASSOCIATED WITH SEIZURES IN MENINGIOMAS. Neuro-Oncology, 2020, 22, ii140-ii140.	1.2	1
120	Response to Letter by Stahl and Felbor. Stroke, 2006, 37, 2215-2216.	2.0	0
121	GENO-15IDENTIFICATION AND GENOMIC ANALYSIS OF HYPER-MUTATED AND ULTRA-MUTATED GBMS. Neuro-Oncology, 2015, 17, v94.3-v94.	1.2	0
122	Genomic Landscape of Brain Tumors. , 2016, , 653-663.		0
123	GENE-56. MENINGIOMA GENOMIC SUBGROUP AS A PREDICTOR OF POST-OPERATIVE PATIENT OUTCOMES: IMPLICATIONS FOR TREATMENT AND FOLLOW-UP. Neuro-Oncology, 2019, 21, vi109-vi110.	1.2	0
124	A congenital disorder of deglycosylation: biochemical characterization of Nâ€glycanase 1 deficiency in patient fibroblasts (607.3). FASEB Journal, 2014, 28, 607.3.	0.5	0
125	MNGI-09. MENINGIOMA WITH MULTIPLE DRIVERS: GENOMIC LANDSCAPE AND CLINICAL CORRELATIONS. Neuro-Oncology, 2019, 21, vi141-vi141.	1.2	0
126	INNV-09. SURGICAL STRATEGIES FOR OLDER PATIENTS WITH GLIOBLASTOMA. Neuro-Oncology, 2021, 23, vi107-vi107.	1.2	0

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127	EPCO-29. GENOMIC PROFILING OF SPORADIC MULTIPLE MENINGIOMAS. Neuro-Oncology, 2021, 23, vi8-vi8.	1.2	0
128	NIMG-64. TYPE OF BONY INVOLVEMENT PREDICTS GENOMIC SUBGROUP IN SPHENOID WING MENINGIOMAS. Neuro-Oncology, 2021, 23, vi144-vi144.	1.2	0
129	Carotid endarterectomy prevention strategies and complications management. Neurosurgery Clinics of North America, 2000, $11$ , $351$ -64.	1.7	0
130	Clinical Implications of the Genomic Profiling of Sporadic Multiple Meningiomas. Journal of Neurological Surgery, Part B: Skull Base, 2022, 83, .	0.8	0
131	NF2 Mutant Sporadic Meningiomas Differ Based on Location Relative to the Tentorium. Journal of Neurological Surgery, Part B: Skull Base, 2022, 83, .	0.8	0
132	TRAF7 Mutated Subgroups Differ in Sphenoid Wing Meningiomas with Hyperostosis. Journal of Neurological Surgery, Part B: Skull Base, 2022, 83, .	0.8	0
133	The quest to unravel the complex genomics of intracranial aneurysms. , 2022, 1, 281-282.		0