Namik Kaya

List of Publications by Year
in descending order

84 papers	2,278 citations	257450 24 h-index	243625 44 g-index
85 all docs	85 docs citations	85 times ranked	4562 citing authors

#	Article	IF	Citations
1	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
2	A Novel GEMIN4 Variant in a Consanguineous Family Leads to Neurodevelopmental Impairment with Severe Microcephaly, Spastic Quadriplegia, Epilepsy, and Cataracts. Genes, 2022, 13, 92.	2.4	6
3	Clinical, genetic, and functional characterization of the glycine receptor Î²-subunit A455P variant in a family affected by hyperekplexia syndrome. Journal of Biological Chemistry, 2022, , 102018.	3.4	0
4	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
5	Hematological findings associated with tubulinâ€folding cofactors Dâ€related encephalopathy: Expanding the phenotype. Clinical Genetics, 2021, 99, 724-731.	2.0	0
6	<scp>SLC25A42</scp>â€associated mitochondrial encephalomyopathy: Report of additional founder cases and functional characterization of a novel deletion. JIMD Reports, 2021, 60, 75-87.	1.5	6
7	Expanding the mutational landscape and clinical phenotype of the YIF1B related brain disorder. Brain, 2021, 144, e85-e85.	7.6	2
8	Genetics of ataxia telangiectasia in a highly consanguineous population. Annals of Human Genetics, 2021, , .	0.8	4
9	Identification of Gene Signature as Diagnostic and Prognostic Blood Biomarker for Early Hepatocellular Carcinoma Using Integrated Cross-Species Transcriptomic and Network Analyses. Frontiers in Genetics, 2021, 12, 710049.	2.3	6
10	Influence of b2 adrenergic receptor polymorphism (rs1042713 and rs1042714) on anthropometric, hormonal and lipid profiles in polycystic ovarian syndrome. Journal of Medical Biochemistry, 2021, 40, 74-85.	1.7	0
11	Phenotypic Variability of MEGF10 Variants Causing Congenital Myopathy: Report of Two Unrelated Patients from a Highly Consanguineous Population. Genes, 2021, 12, 1783.	2.4	3
12	Exome Sequencing Reveals Novel TTN Variants in Saudi Patients with Congenital Titinopathies. Genetic Testing and Molecular Biomarkers, 2021, 25, 757-764.	0.7	4
13	Genetic and phenotypic characterization of NKX6â€2 â€related spastic ataxia and hypomyelination. European Journal of Neurology, 2020, 27, 334-342.	3.3	16
14	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
15	Pyrostigmine therapy in a patient with VAMP1-related congenital myasthenic syndrome. Neuromuscular Disorders, 2020, 30, 611-615.	0.6	6
16	Ancient founder mutation in RUBCN: a second unrelated family confirms Salih ataxia (SCAR15). BMC Neurology, 2020, 20, 207.	1.8	7
17	Rett Syndrome, a Neurodevelopmental Disorder, Whole-Transcriptome, and Mitochondrial Genome Multiomics Analyses Identify Novel Variations and Disease Pathways. OMICS A Journal of Integrative Biology, 2020, 24, 160-171.	2.0	18
18	Truncating mutations in YIF1B cause a progressive encephalopathy with various degrees of mixed movement disorder, microcephaly, and epilepsy. Acta Neuropathologica, 2020, 139, 791-794.	7.7	17

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19	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	2.3	13
20	Cell-free DNA levels of twins and sibling pairs indicate individuality and possible use as a personalized biomarker. PLoS ONE, 2019, 14, e0223470.	2.5	12
21	First report of two successive deletions on chromosome 15q13 cytogenetic bands in a boy and girl: additional data to 15q13.3 syndrome with a report of high IQ patient. Molecular Cytogenetics, 2019, 12, 21.	0.9	2
22	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	1.2	7
23	A new association between CDK5RAP2 microcephaly and congenital cataracts. Annals of Human Genetics, 2018, 82, 165-170.	0.8	9
24	Further delineation of the phenotypic spectrum ofÂ ISCA2 defect: A report of ten new cases. European Journal of Paediatric Neurology, 2018, 22, 46-55.	1.6	21
25	A Novel Homozygous Mutation in SPTBN2 Leads to Spinocerebellar Ataxia in a Consanguineous Family: Report of a New Infantile-Onset Case and Brief Review of the Literature. Cerebellum, 2018, 17, 276-285.	2.5	18
26	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. Molecular Cytogenetics, 2018, 11, 9.	0.9	4
27	Polymorphic Variations in VDR Gene in Saudi Women with and Without Polycystic Ovary Syndrome (PCOS) and Significant Influence of Seven Polymorphic Sites on Anthropometric And Hormonal Parameters. Journal of Medical Biochemistry, 2018, 37, 415-425.	1.7	8
28	Mutational analysis of PHEX, FGF23 and CLCN5 in patients with hypophosphataemic rickets. Clinical Endocrinology, 2017, 87, 103-112.	2.4	21
29	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
30	Hemophagocytic lymphohistiocytosis: A rare cause of recurrent encephalopathy. Intractable and Rare Diseases Research, 2016, 5, 227-230.	0.9	8
31	KCNA4deficiency leads to a syndrome of abnormal striatum, congenital cataract and intellectual disability. Journal of Medical Genetics, 2016, 53, 786-792.	3.2	24
32	A substitution mutation in cardiac ubiquitin ligase, FBXO32, is associated with an autosomal recessive form of dilated cardiomyopathy. BMC Medical Genetics, 2016, 17, 3.	2.1	19
33	Integrated Genomic and Network-Based Analyses of Complex Diseases and Human Disease Network. Journal of Genetics and Genomics, 2016, 43, 349-367.	3.9	21
34	Integrated Left Ventricular Global Transcriptome and Proteome Profiling in Human End-Stage Dilated Cardiomyopathy. PLoS ONE, 2016, 11, e0162669.	2.5	33
35	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161.	6.4	375
36	ISCA2mutation causes infantile neurodegenerative mitochondrial disorder. Journal of Medical Genetics, 2015, 52, 186-194.	3.2	90

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37	Pinar T. Ozand. Journal of Child Neurology, 2015, 30, 1090-1095.	1.4	0
38	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	2.4	19
39	JCN Calendar of Events. Journal of Child Neurology, 2015, 30, 1099-1099.	1.4	0
40	Genetics of autism spectrum disorder: an update on copy number variations leading to autism in the next generation sequencing era. Discovery Medicine, 2015, 19, 367-79.	0.5	4
41	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47
42	Novel homozygous DEAF1 variant suspected in causing white matter disease, intellectual disability, and microcephaly. American Journal of Medical Genetics, Part A, 2014, 164, 1565-1570.	1.2	21
43	Clinical, neuroimaging, and genetic features of L-2-hydroxyglutaric aciduria in Arab kindreds. Annals of Saudi Medicine, 2014, 34, 107-114.	1.1	14
44	Gene expression profiling of granulosa cells from PCOS patients following varying doses of human chorionic gonadotropin. Journal of Assisted Reproduction and Genetics, 2013, 30, 341-352.	2.5	13
45	Clinical and biochemical features associated with BCS1L mutation. Journal of Inherited Metabolic Disease, 2013, 36, 813-820.	3.6	25
46	Induction of cell proliferation in old rat liver can reset certain gene expression levels characteristic of old liver to those associated with young liver. Age, 2013, 35, 719-732.	3.0	8
47	Breast stromal fibroblasts from histologically normal surgical margins are proâ€carcinogenic. Journal of Pathology, 2013, 231, 457-465.	4.5	29
48	Mutations in NALCN Cause an Autosomal-Recessive Syndrome with Severe Hypotonia, Speech Impairment, and Cognitive Delay. American Journal of Human Genetics, 2013, 93, 721-726.	6.2	88
49	A novel mutation in a large family causes a unique phenotype of Mucolipidosis IV. Gene, 2013, 526, 464-466.	2.2	9
50	Identification of a novel IVD mutation in a consanguineous family with isovaleric acidemia. Gene, 2013, 513, 297-300.	2.2	5
51	Prevalence ofPIK3CAmutations and the SNP rs17849079 in Arab breast cancer patients. Cancer Biology and Therapy, 2013, 14, 888-896.	3.4	23
52	A novel syndrome of abnormal striatum and congenital cataract: evidence for linkage to chromosomes 11. Clinical Genetics, 2013, 84, 258-264.	2.0	6
53	Age-Specific Gene Expression Signatures for Breast Tumors and Cross-Species Conserved Potential Cancer Progression Markers in Young Women. PLoS ONE, 2013, 8, e63204.	2.5	116
54	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. Genetics in Medicine, 2012, 14, 515-519.	2.4	10

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55	Genome-Wide Expression Profiling of Patients with Primary Open Angle Glaucoma. , 2012, 53, 5899.		12
56	A novel Xâ€linked disorder with developmental delay and autistic features. Annals of Neurology, 2012, 71, 498-508.	5.3	33
57	Novel V97G ASAH1 mutation found in Farber disease patients: Unique appearance of the disease with an intermediate severity, and marked early involvement of central and peripheral nervous system. Brain and Development, 2012, 34, 400-404.	1.1	16
58	Novel mutation in GLRB in a large family with hereditary hyperekplexia. Clinical Genetics, 2012, 81, 479-484.	2.0	31
59	Abstract 52: Gene expression profiling of breast cancer associated fibroblasts and their adjacent counterparts. , 2012, , .		0
60	Genomic and transcriptomic analyses distinguish classic Rett and Rett-like syndrome and reveals shared altered pathways. Genomics, 2011, 97, 19-28.	2.9	28
61	p16INK4A Positively Regulates Cyclin D1 and E2F1 through Negative Control of AUF1. PLoS ONE, 2011, 6, e21111.	2.5	47
62	Novel intragenic deletion in OPHN1 in a family causing XLMR with cerebellar hypoplasia and distinctive facial appearance. Clinical Genetics, 2011, 79, 363-370.	2.0	24
63	A novel deletion of theMEN1gene in a large family of multiple endocrine neoplasia type 1 (MEN1) with aggressive phenotype. Clinical Endocrinology, 2011, 75, 791-800.	2.4	33
64	Chromosome 12q24.31-q24.33 deletion causes multiple dysmorphic features and developmental delay: First mosaic patient and overview of the phenotype related to 12q24qter defects. Molecular Cytogenetics, 2011, 4, 9.	0.9	10
65	ICF Syndrome in Saudi Arabia: Immunological, Cytogenetic and Molecular Analysis. Journal of Clinical Immunology, 2011, 31, 245-252.	3.8	11
66	GM2 gangliosidosis in Saudi Arabia: Multiple mutations and considerations for future carrier screening. American Journal of Medical Genetics, Part A, 2011, 155, 1281-1284.	1.2	19
67	A novel interstitial microdeletion of 7q22.1â€7q22.3 detected by array comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2011, 155, 3128-3131.	1.2	8
68	Phenotypical spectrum of cerebellar ataxia associated with a novel mutation in the CA8 gene, encoding carbonic anhydrase (CA) VIII. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 826-834.	1.7	55
69	Renal Failure Associated with APECED and Terminal 4q Deletion: Evidence of Autoimmune Nephropathy. Clinical and Developmental Immunology, 2010, 2010, 1-7.	3.3	15
70	Integrative and comparative genomics analysis of early hepatocellular carcinoma differentiated from liver regeneration in young and old. Molecular Cancer, 2010, 9, 146.	19.2	30
71	A novel mutation and first report of dilated cardiomyopathy in ALG6-CDG (CDG-Ic): a case report. Orphanet Journal of Rare Diseases, 2010, 5, 7.	2.7	33
72	Left ventricular global transcriptional profiling in human end-stage dilated cardiomyopathy. Genomics, 2009, 94, 20-31.	2.9	48

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73	A novel missense and a recurrent mutation in SLC2A10 gene of patients affected with arterial tortuosity syndrome. Atherosclerosis, 2009, 203, 466-471.	0.8	26
74	Mutations in C2orf37, Encoding a Nucleolar Protein, Cause Hypogonadism, Alopecia, Diabetes Mellitus, Mental Retardation, and Extrapyramidal Syndrome. American Journal of Human Genetics, 2008, 83, 684-691.	6.2	121
75	Comparison of two dependent within subject coefficients of variation to evaluate the reproducibility of measurement devices. BMC Medical Research Methodology, 2008, 8, 24.	3.1	32
76	Array comparative genomic hybridization (aCGH) reveals the largest novel deletion in PCCA found in a Saudi family with propionic acidemia. European Journal of Medical Genetics, 2008, 51, 558-565.	1.3	13
77	Identification of Gaucher disease mutations found in Saudi Arabia. Blood Cells, Molecules, and Diseases, 2008, 41, 200-201.	1.4	6
78	Genome-wide gene expression profiling and mutation analysis of Saudi patients with Canavan disease. Genetics in Medicine, 2008, 10, 675-684.	2.4	20
79	Computational Analysis of Transcriptional Profiling in Dysmorphic Syndrome. , 2007, , .		0
80	Expression, physiological action, and coexpression patterns of neuropeptide Y in rat taste-bud cells. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 11100-11105.	7.1	102
81	Expression and Physiological Actions of Cholecystokinin in Rat Taste Receptor Cells. Journal of Neuroscience, 2002, 22, 10018-10029.	3.6	105
82	Adrenergic signalling between rat taste receptor cells. Journal of Physiology, 2002, 543, 601-614.	2.9	60
83	Hereditary Disorders and Human Mutations of Iron-Sulfur Assembly Genes. , 0, , .		1
84	Involvement of mitochondrial dysfunction in pathogenesis of hemophagocytic lymphohistiocytosis. Journal of Biochemical and Clinical Genetics, 0, , 81-84.	0.1	1