Guney Bademci

List of Publications by Year
in descending order

65 papers	1,766 citations	304602 22 h-index	302012 39 g-index
69 all docs	69 docs citations	69 times ranked	3647 citing authors

#	Article	IF	Citations
1	Genome sequencing reveals novel noncoding variants in <scp>PLA2G6</scp> and <scp>LMNB1</scp> causing progressive neurologic disease. Molecular Genetics & Genomic Medicine, 2022, 10, e1892.	0.6	4
2	Biallelic <scp> KITLG </scp> variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. Journal of the European Academy of Dermatology and Venereology, 2022, , .	1.3	1
3	Mutations in MINAR2 encoding membrane integral NOTCH2-associated receptor 2 cause deafness in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	3
4	First reported adult patient with retinal dystrophy and leukodystrophy caused by a novel <scp>ACBD5</scp> variant: A case report and review of literature. American Journal of Medical Genetics, Part A, 2021, 185, 1236-1241.	0.7	10
5	Confirmation of COL4A6 variants in X-linked nonsyndromic hearing loss and its clinical implications. European Journal of Human Genetics, 2021, , .	1.4	5
6	A founder noncoding GALT variant interfering with splicing causes galactosemia. Journal of Inherited Metabolic Disease, 2020, 43, 1199-1204.	1.7	1
7	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
8	Analyses of del(GJB6â€Ð13S1830) and del(GJB6â€Ð13S1834) deletions in a large cohort with hearing loss: Caveats to interpretation of molecular test results in multiplex families. Molecular Genetics & Genomic Medicine, 2020, 8, e1171.	0.6	10
9	Spectrum of Genetic Variants Associated with Anterior Segment Dysgenesis in South Florida. Genes, 2020, 11, 350.	1.0	14
10	Long-range cis-regulatory elements controlling GDF6 expression are essential for ear development. Journal of Clinical Investigation, 2020, 130, 4213-4217.	3.9	9
11	Radixin modulates the function of outer hair cell stereocilia. Communications Biology, 2020, 3, 792.	2.0	5
12	Identification of Main Genetic Causes Responsible for Non-Syndromic Hearing Loss in a Peruvian Population. Genes, 2019, 10, 581.	1.0	5
13	Novel variant p.E269K confirms causative role of PLS1 mutations in autosomal dominant hearing loss. Clinical Genetics, 2019, 96, 575-578.	1.0	8
14	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	2.6	29
15	A truncating CLDN9 variant is associated with autosomal recessive nonsyndromic hearing loss. Human Genetics, 2019, 138, 1071-1075.	1.8	17
16	FOXF2is required for cochlear development in humans and mice. Human Molecular Genetics, 2019, 28, 1286-1297.	1.4	20
17	Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1347-1352.	3.3	15
18	A MECOM variant in an African American child with radioulnar synostosis and thrombocytopenia. Clinical Dysmorphology, 2018, 27, 9-11.	0.1	18

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19	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	1.0	46
20	Identification of candidate gene FAM183A and novel pathogenic variants in known genes: High genetic heterogeneity for autosomal recessive intellectual disability. PLoS ONE, 2018, 13, e0208324.	1.1	15
21	Ripor2 is involved in auditory hair cell stereociliary bundle structure and orientation. Journal of Molecular Medicine, 2018, 96, 1227-1238.	1.7	8
22	Monosomy chromosome 21 compensated by 21q22.11q22.3 duplication in a case with small size and minor anomalies. Molecular Cytogenetics, 2018, 11, 43.	0.4	2
23	MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss. Human Genetics, 2018, 137, 479-486.	1.8	19
24	Dominant deafnessâ€“onychodystrophy syndrome caused by an <scp>ATP</scp>6V1B2 mutation. Clinical Case Reports (discontinued), 2017, 5, 376-379.	0.2	27
25	Novel Causative Variants in DYRK1A, KARS, and KAT6A Associated with Intellectual Disability and Additional Phenotypic Features. Journal of Pediatric Genetics, 2017, 06, 077-083.	0.3	25
26	Novel EYA1 variants causing Branchio-oto-renal syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 98, 59-63.	0.4	16
27	Novel pathogenic variants underlie SLC26A4 -related hearing loss in a multiethnic cohort. International Journal of Pediatric Otorhinolaryngology, 2017, 101, 167-171.	0.4	11
28	Research of genetic bases of hereditary non-syndromic hearing loss. Turk Pediatri Arsivi, 2017, 52, 122-132.	0.9	7
29	A Mayan founder mutation is a common cause of deafness in Guatemala. Clinical Genetics, 2016, 89, 461-465.	1.0	17
30	ROR1 is essential for proper innervation of auditory hair cells and hearing in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 5993-5998.	3.3	42
31	Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss. Scientific Reports, 2016, 6, 31622.	1.6	44
32	Targeted Resequencing of Deafness Genes Reveals a FounderMYO15AVariant in Northeastern Brazil. Annals of Human Genetics, 2016, 80, 327-331.	0.3	17
33	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	1.8	102
34	A next-generation sequencing gene panel (MiamiOtoGenes) for comprehensive analysis of deafness genes. Hearing Research, 2016, 333, 179-184.	0.9	38
35	Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort. Genetics in Medicine, 2016, 18, 364-371.	1.1	124
36	Nijmegen-Breakage Syndrome; Two Siblings Presenting with Different Phenotypes. Asim, Allerji, Immunoloji, 2016, , .	0.2	0

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37	Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 128.	1.2	46
38	Novel mutations confirm that COL11A2 is responsible for autosomal recessive non-syndromic hearing loss DFNB53. Molecular Genetics and Genomics, 2015, 290, 1327-1334.	1.0	25
39	MORFAN Syndrome: An Infantile Hypoinsulinemic Hypoketotic Hypoglycemia Due to an AKT2 Mutation. Journal of Pediatrics, 2015, 167, 489-491.	0.9	13
40	Whole-exome sequencing and its impact in hereditary hearing loss. Genetical Research, 2015, 97, e4.	0.3	43
41	Novel domain-specific POU3F4 mutations are associated with X-linked deafness: examples from different populations. BMC Medical Genetics, 2015, 16, 9.	2.1	15
42	Comprehensive Analysis of Deafness Genes in Families with Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2015, 10, e0142154.	1.1	52
43	FAM65B is a membrane-associated protein of hair cell stereocilia required for hearing. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 9864-9868.	3.3	70
44	Common genes for non-syndromic deafness are uncommon in sub-Saharan Africa: A report from Nigeria. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 1870-1873.	0.4	26
45	Identification of Copy Number Variants Through Whole-Exome Sequencing in Autosomal Recessive Nonsyndromic Hearing Loss. Genetic Testing and Molecular Biomarkers, 2014, 18, 658-661.	0.3	30
46	Evidence for genotypeâ€“phenotype correlation for OTOF mutations. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 950-953.	0.4	15
47	C9ORF72 Intermediate Repeat Copies Are a Significant Risk Factor for Parkinson Disease. Annals of Human Genetics, 2013, 77, 351-363.	0.3	69
48	High-Resolution Survey in Familial Parkinson Disease Genes Reveals Multiple Independent Copy Number Variation Events in PARK2. Human Mutation, 2013, 34, 1071-1074.	1.1	13
49	Whole exome sequencing of rare variants in EIF4G1 and VPS35 in Parkinson disease. Neurology, 2013, 80, 982-989.	1.5	68
50	Tyrosine Hydroxylase Gene: Another Piece of the Genetic Puzzle of Parkinsonâ€™s Disease. CNS and Neurological Disorders - Drug Targets, 2012, 11, 469-481.	0.8	21
51	Exploring the relationship between the severity of oligozoospermia and the frequencies of sperm chromosome aneuploidies. Andrologia, 2012, 44, 416-422.	1.0	13
52	Identifying Consensus Disease Pathways in Parkinson's Disease Using an Integrative Systems Biology Approach. PLoS ONE, 2011, 6, e16917.	1.1	72
53	Comparison of Three Targeted Enrichment Strategies on the SOLiD Sequencing Platform. PLoS ONE, 2011, 6, e18595.	1.1	66
54	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodontia. American Journal of Human Genetics, 2011, 89, 289-294.	2.6	205

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55	Conventional and molecular cytogenetic analyses in Turkish patients with multiple myeloma. Turkish Journal of Haematology, 2011, 29, 135-42.	0.2	6
56	A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss. American Journal of Human Genetics, 2010, 86, 797-804.	2.6	56
57	A rare novel deletion of the tyrosine hydroxylase gene in Parkinson disease. Human Mutation, 2010, 31, E1767-E1771.	1.1	29
58	Can facet joint infiltrative analgesia reduce postoperative pain in degenerative lumbar disc surgery?. Neurocirugia, 2008, 19, 45-49.	0.2	3
59	Extremely delayed renal cell carcinoma metastasis mimicking convexity meningioma. Neurocirugia, 2008, 19, 562-4.	0.2	3
60	Persistent metopic suture can mimic the skull fractures in the emergency setting?. Neurocirugia, 2007, 18, 238-40.	0.2	5
61	Late dissemination of ependymoma: case report. Neurocirugia, 2007, 18, 333-6.	0.2	6
62	First illustrations of female â€œNeurosurgeonsâ€•in the fifteenth century by Serefeddin Sabuncuoglu. Neurocirugia, 2006, 17, 162-165.	0.2	17
63	810 COMPLEX REGIONAL PAIN SYNDROME DUE TO RADIATION INDUCED BRACHIAL PLEXOPATHY. European Journal of Pain, 2006, 10, S210c-S211.	1.4	0
64	A novel mutation in the ARS (component B) gene encoding SLURP-1 in a Turkish family with mal de Meleda. British Journal of Dermatology, 2006, 155, 467-469.	1.4	12
65	Non-Traumatic Elevation Techniques of the Hypoglossal Nerve during Carotid Endarterectomy: A Cadaveric Study. Minimally Invasive Neurosurgery, 2005, 48, 108-112.	0.9	12