

Zeynep Coban Akdemir

List of Publications by Year in descending order

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103
papers

4,360
citations

87888

38
h-index

138484

58
g-index

105
all docs

105
docs citations

105
times ranked

9055
citing authors

#	ARTICLE	IF	CITATIONS
1	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	8.1	258
2	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	8.2	184
3	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. <i>American Journal of Human Genetics</i> , 2018, 103, 171-187.	6.2	160
4	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	6.2	146
5	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. <i>Journal of Experimental Medicine</i> , 2019, 216, 2778-2799.	8.5	132
6	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 1126-1142.	6.2	128
7	Phenotypic expansion illuminates multilocus pathogenic variation. <i>Genetics in Medicine</i> , 2018, 20, 1528-1537.	2.4	104
8	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. <i>Nucleic Acids Research</i> , 2017, 45, gkw1237.	14.5	98
9	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 27-43.	6.2	88
10	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 2296-2299.	2.9	87
11	Molecular etiology of arthrogyrosis in multiple families of mostly Turkish origin. <i>Journal of Clinical Investigation</i> , 2016, 126, 762-778.	8.2	82
12	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. <i>BMC Medical Genomics</i> , 2016, 9, 42.	1.5	80
13	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	3.5	80
14	Biallelic mutations in IRF8 impair human NK cell maturation and function. <i>Journal of Clinical Investigation</i> , 2016, 127, 306-320.	8.2	76
15	Predicting human genes susceptible to genomic instability associated with Alu-mediated rearrangements. <i>Genome Research</i> , 2018, 28, 1228-1242.	5.5	74
16	The Genomics of Arthrogyrosis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. <i>American Journal of Human Genetics</i> , 2019, 105, 132-150.	6.2	74
17	Myc and SAGA rewire an alternative splicing network during early somatic cell reprogramming. <i>Genes and Development</i> , 2015, 29, 803-816.	5.9	73
18	Whole-Exome Sequencing in Familial Parkinson Disease. <i>JAMA Neurology</i> , 2016, 73, 68.	9.0	71

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19	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 562-570.	6.2	66
20	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. <i>Cell</i> , 2017, 168, 830-842.e7.	28.9	66
21	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	6.2	66
22	Phenotypic expansion in <i>DDX3X</i> a common cause of intellectual disability in females. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1277-1285.	3.7	66
23	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 904-913.	6.2	65
24	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. <i>Science</i> , 2020, 369, 202-207.	12.6	65
25	TRIM24 suppresses development of spontaneous hepatic lipid accumulation and hepatocellular carcinoma in mice. <i>Journal of Hepatology</i> , 2015, 62, 371-379.	3.7	63
26	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2017, 100, 117-127.	6.2	62
27	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	6.2	59
28	Whole-exome sequencing identifies novel homozygous mutation in <i>NPAS2</i> in family with nonobstructive azoospermia. <i>Fertility and Sterility</i> , 2015, 104, 286-291.	1.0	58
29	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	2.5	58
30	Bi-allelic Mutations in <i>PKD1L1</i> Are Associated with Laterality Defects in Humans. <i>American Journal of Human Genetics</i> , 2016, 99, 886-893.	6.2	57
31	The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. <i>Human Genetics</i> , 2018, 137, 553-567.	3.8	57
32	Phenotypic expansion of <i>TBX4</i> mutations to include acinar dysplasia of the lungs. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2440-2444.	1.2	56
33	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	6.2	56
34	Identification of novel candidate disease genes from de novo exonic copy number variants. <i>Genome Medicine</i> , 2017, 9, 83.	8.2	50
35	Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippel-Feil syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2795-2799.	1.2	47
36	Mechanisms for Complex Chromosomal Insertions. <i>PLoS Genetics</i> , 2016, 12, e1006446.	3.5	45

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37	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156.	6.2	44
38	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	2.8	44
39	Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286.	8.2	44
40	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
41	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
42	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	6.2	41
43	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
44	Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. Human Molecular Genetics, 2018, 27, 1913-1926.	2.9	39
45	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	6.2	38
46	Mutations in PI3K110 β cause impaired natural killer cell function partially rescued by rapamycin treatment. Journal of Allergy and Clinical Immunology, 2018, 142, 605-617.e7.	2.9	36
47	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	6.2	36
48	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
49	Phenotypic expansion of <i>POGZ</i> -related intellectual disability syndrome (White-Sutton) Tj ETQq1 1 0.784314 rgBT /Overlock	1.2	35
50	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
51	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
52	A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1294-1305.	2.8	28
53	<i>TBX6</i> missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	2.5	27
54	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	27

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55	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. <i>European Journal of Human Genetics</i> , 2020, 28, 1243-1264.	2.8	27
56	Dual molecular diagnosis contributes to atypical Prader-Willi phenotype in monozygotic twins. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2451-2455.	1.2	26
57	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. <i>Blood</i> , 2021, 137, 493-499.	1.4	26
58	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	3.8	24
59	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	6.2	24
60	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 365.	2.7	24
61	Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. <i>Frontiers in Immunology</i> , 2017, 8, 576.	4.8	23
62	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. <i>Genome Medicine</i> , 2019, 11, 25.	8.2	22
63	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	2.4	22
64	Exome sequencing reveals homozygous TRIM2 mutation in a patient with early onset CMT and bilateral vocal cord paralysis. <i>Human Genetics</i> , 2015, 134, 671-673.	3.8	21
65	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. <i>Genome Medicine</i> , 2016, 8, 105.	8.2	20
66	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	3.7	20
67	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in <i>FBN1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1023.	1.2	19
68	Bi-allelic <i>CCDC47</i> Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2018, 103, 794-807.	6.2	18
69	Novel Heterozygous Mutation in <i>NFKB2</i> Is Associated With Early Onset CVID and a Functional Defect in NK Cells Complicated by Disseminated CMV Infection and Severe Nephrotic Syndrome. <i>Frontiers in Pediatrics</i> , 2019, 7, 303.	1.9	18
70	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	1.2	17
71	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	7.6	17
72	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3593-3600.	1.2	16

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73	A novel homozygous <i>SLC13A5</i> whole-gene deletion generated by <i>Alu/Alu</i> -mediated rearrangement in an Iraqi family with epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1972-1980.	1.2	16
74	Biallelic and <i>De Novo</i> Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleopathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	1.2	15
75	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	3.7	15
76	<i>Wolff</i> "Parkinson" White syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1387-1399.	1.2	14
77	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 735-750.	1.2	14
78	First Case of CD40LG Deficiency in Ecuador, Diagnosed after Whole Exome Sequencing in a Patient with Severe Cutaneous Histoplasmosis. <i>Frontiers in Pediatrics</i> , 2017, 5, 17.	1.9	13
79	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 302-304.	1.1	13
80	Neurodevelopmental disorder in an Egyptian family with a biallelic <i>ALKBH8</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	1.2	13
81	Exome sequencing identifies a homozygous <i>C5orf42</i> variant in a Turkish kindred with oral-facial-digital syndrome type VI. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2132-2137.	1.2	12
82	Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. <i>Frontiers in Pediatrics</i> , 2021, 9, 673957.	1.9	12
83	A biallelic <i>ANTXR1</i> variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1015-1022.	1.2	11
84	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. <i>Neurology: Genetics</i> , 2020, 6, e498.	1.9	11
85	Disease-associated CTNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. <i>Journal of Clinical Investigation</i> , 2020, 130, 4411-4422.	8.2	11
86	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	2.5	11
87	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. <i>Parkinsonism and Related Disorders</i> , 2021, 82, 84-86.	2.2	10
88	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 2455-2460.	2.4	9
89	Biallelic in-frame deletion in <i>TRAPPC4</i> in a family with developmental delay and cerebellar atrophy. <i>Brain</i> , 2020, 143, e83-e83.	7.6	8
90	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. <i>Kidney International</i> , 2022, 101, 1039-1053.	5.2	8

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91	Phenotypic and mutational spectrum of <i>ROR2</i> -related Robinow syndrome. <i>Human Mutation</i> , 2022, 43, 900-918.	2.5	8
92	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1897-1909.	1.2	7
93	Biallelic Pathogenic Variants in <i>TNNT3</i> Associated With Congenital Myopathy. <i>Neurology: Genetics</i> , 2021, 7, e589.	1.9	6
94	Risk of sudden cardiac death in <i>EXOSC5</i> -related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2532-2540.	1.2	6
95	Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 758-766.	2.9	6
96	Novel <i>RETREG1</i> (<i>FAM134B</i>) founder allele is linked to <i>HSAN2B</i> and renal disease in a Turkish family. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2153-2161.	1.2	4
97	Biallelic Variants in the Ectonucleotidase <i>ENTPD1</i> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 92, 304-321.	5.3	2
98	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). <i>JBMR Plus</i> , 2020, 4, e10335.	2.7	1
99	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic <i>EMD</i> splice variant. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2052-2058.	3.7	1
100	Whole Exome Sequencing Identifies Potential Defects in Multiple Immunodeficiency-Associated Genes in Individual Patients and Families with Primary Immunodeficiency Diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, AB172.	2.9	0
101	Front Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, i.	2.5	0
102	Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. <i>Blood</i> , 2019, 134, 83-83.	1.4	0
103	Elucidating the clinical spectrum and molecular basis of <i>HYAL2</i> deficiency. <i>Genetics in Medicine</i> , 2022, 24, 631-644.	2.4	0