

Pelin Ozlem Simsek Kiper

List of Publications by Year in descending order

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Version: 2024-02-01

90
papers

1,638
citations

394421

19
h-index

330143

37
g-index

94
all docs

94
docs citations

94
times ranked

3545
citing authors

#	ARTICLE	IF	CITATIONS
1	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. <i>Human Molecular Genetics</i> , 2013, 22, 5121-5135.	2.9	190
2	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. <i>Human Mutation</i> , 2016, 37, 847-864.	2.5	134
3	Cortical-Bone Fragility â€” Insights from sFRP4 Deficiency in Pyleâ€™s Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2553-2562.	27.0	119
4	A mutation screen in patients with Kabuki syndrome. <i>Human Genetics</i> , 2011, 130, 715-724.	3.8	106
5	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Málformation. <i>American Journal of Human Genetics</i> , 2019, 104, 925-935.	6.2	92
6	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	2.4	80
7	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 3585-3599.	8.2	69
8	A Homozygous Deletion in <i>GRID2</i> Causes a Human Phenotype With Cerebellar Ataxia and Atrophy. <i>Journal of Child Neurology</i> , 2013, 28, 926-932.	1.4	67
9	Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. <i>Human Genetics</i> , 2015, 134, 553-568.	3.8	59
10	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	2.8	56
11	Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. <i>Genetics in Medicine</i> , 2016, 18, 882-891.	2.4	37
12	HERC1 mutations in idiopathic intellectual disability. <i>European Journal of Medical Genetics</i> , 2017, 60, 279-283.	1.3	37
13	Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 60.	2.7	35
14	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by <i>PAPSS2</i> Mutations. <i>Human Mutation</i> , 2013, 34, 1381-1386.	2.5	29
15	Functional analysis of a duplication (p.E63_D69dup) in the switch II region of HRAS: new aspects of the molecular pathogenesis underlying Costello syndrome. <i>Human Molecular Genetics</i> , 2013, 22, 1643-1653.	2.9	26
16	Etiological yield of SNP microarrays in idiopathic intellectual disability. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 327-337.	1.6	26
17	TMCO1 deficiency causes autosomal recessive cerebrotiothoracic dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 291-304.	1.2	25
18	Clinical and molecular analysis of RASopathies in a group of Turkish patients. <i>Clinical Genetics</i> , 2013, 83, 181-186.	2.0	24

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19	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
20	Positive effects of an angiotensin II type 1 receptor antagonist in Camurati-Engelmann disease: A single case observation. American Journal of Medical Genetics, Part A, 2014, 164, 2667-2671.	1.2	21
21	Striking hematological abnormalities in patients with microcephalic osteodysplastic primordial dwarfism type II (MOPD II): A potential role of pericentrin in hematopoiesis. Pediatric Blood and Cancer, 2014, 61, 302-305.	1.5	21
22	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. Human Mutation, 2021, 42, 711-730.	2.5	19
23	Novel homozygous mutations in the osteoprotegerin gene TNFRSF11B in two unrelated patients with juvenile Paget's disease. Bone, 2014, 68, 6-10.	2.9	18
24	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscaphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	2.8	18
25	HEATR3 variants impair nuclear import of uL18 (RPL5) and drive Diamond-Blackfan anemia. Blood, 2022, 139, 3111-3126.	1.4	15
26	Homozygosity for a novel truncating mutation confirms TBX15 deficiency as the cause of Cousin syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 3161-3165.	1.2	14
27	Further expanding the mutational spectrum and investigation of genotype-phenotype correlation in 3M syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1157-1172.	1.2	14
28	Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features. American Journal of Medical Genetics, Part A, 2021, 185, 1888-1896.	1.2	14
29	Homozygous indel mutation in CDH11 as the probable cause of Elshahy-Waters syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3143-3152.	1.2	13
30	Diagnostic yield of whole-exome sequencing in non-syndromic intellectual disability. Journal of Intellectual Disability Research, 2021, 65, 577-588.	2.0	11
31	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	6.2	11
32	Experience of a skeletal dysplasia registry in Turkey: A five-years retrospective analysis. American Journal of Medical Genetics, Part A, 2015, 167, 2065-2074.	1.2	10
33	Intrafamilial variability of XYLT2-related spondyloocular syndrome. European Journal of Medical Genetics, 2019, 62, 103585.	1.3	9
34	Dermal fibroblast transcriptome indicates contribution of wnt signaling pathways in the pathogenesis of apert syndrome. Turkish Journal of Pediatrics, 2017, 59, 619.	0.6	9
35	Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification. Journal of Human Genetics, 2018, 63, 1003-1007.	2.3	8
36	Kohlschütter-Tarnz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity. Journal of Child Neurology, 2021, 36, 816-822.	1.4	8

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37	Catelâ€“Manzke syndrome: A clinical report suggesting autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2011, 155, 2288-2292.	1.2	7
38	Molecular Etiology of Isolated Congenital Cataract Using Next-Generation Sequencing: Single Center Exome Sequencing Data from Turkey. Molecular Syndromology, 2020, 11, 302-308.	0.8	7
39	A rare cause of syndromic short stature: <scp>3M</scp> syndrome in three families. American Journal of Medical Genetics, Part A, 2021, 185, 461-468.	1.2	7
40	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	2.8	7
41	Fragile x-associated premature ovarian failure in a large Turkish cohort: Findings of Hacettepe Fragile X Registry. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 221, 76-80.	1.1	6
42	Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum. European Journal of Medical Genetics, 2019, 62, 103535.	1.3	6
43	Atypical Presentation of Sengers Syndrome: A Novel Mutation Revealed with Postmortem Genetic Testing. Fetal and Pediatric Pathology, 2020, 39, 163-171.	0.7	6
44	Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype. European Journal of Paediatric Neurology, 2021, 32, 46-55.	1.6	6
45	Spondyloepimetaphyseal dysplasia <scp>EXTL3â€“deficient</scp> type: Longâ€“term <scp>followâ€“up</scp> and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3104-3110.	1.2	6
46	Expanding the phenotypic spectrum of TNFRSF11A-associated dysosteosclerosis: a case with intracranial extramedullary hematopoiesis. Journal of Human Genetics, 2021, 66, 607-611.	2.3	6
47	Peters plus syndrome: a recognizable clinical entity. Turkish Journal of Pediatrics, 2020, 62, 136.	0.6	6
48	Diagnostic yield of microarrays in individuals with nonâ€“syndromic developmental delay and intellectual disability. Journal of Intellectual Disability Research, 2021, 65, 1033-1048.	2.0	6
49	Barraquerâ€“Simons syndrome: A rare clinical entity. American Journal of Medical Genetics, Part A, 2014, 164, 1756-1760.	1.2	5
50	ADA2 deficiency in a patient with Noonan syndromeâ€“like disorder with loose anagen hair: The coâ€“occurrence of two rare syndromes. American Journal of Medical Genetics, Part A, 2019, 179, 2474-2480.	1.2	5
51	An eightâ€“case 1q21 region series: novel aberrations and clinical variability with new features. Journal of Intellectual Disability Research, 2019, 63, 548-557.	2.0	5
52	Hyperinsulinemic Hypoglycemia in a Patient with Costello Syndrome: An Etiology to Consider in Hypoglycemia. Molecular Syndromology, 2020, 11, 207-216.	0.8	5
53	Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum. Cleft Palate-Craniofacial Journal, 2021, , 105566562110381.	0.9	5
54	Prenatal and Postnatal Follow-up in Trisomies 13 and 18: A 20-Year Experience in a Tertiary Center. American Journal of Perinatology, 2018, 35, 427-433.	1.4	4

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55	A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate. <i>European Journal of Medical Genetics</i> , 2019, 62, 21-26.	1.3	4
56	Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience. <i>Journal of Human Genetics</i> , 2021, 66, 585-596.	2.3	4
57	Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum. <i>Molecular Syndromology</i> , 2021, 12, 106-111.	0.8	4
58	Epigenotype and phenotype correlations in patients with beckwith-wiedemann syndrome. <i>Turkish Journal of Pediatrics</i> , 2018, 60, 506.	0.6	4
59	Partial monosomy 3q26.33-3q27.3 presenting with intellectual disability, facial dysmorphism, and diaphragm eventration. <i>Clinical Dysmorphology</i> , 2014, 23, 147-151.	0.3	3
60	A Revisited Diagnosis of Collagen VI Related Muscular Dystrophy in a Patient with a Novel COL6A2 Variant and 21q22.3 Deletion. <i>Neuropediatrics</i> , 2020, 51, 445-449.	0.6	3
61	Genetic IGF1R defects: new cases expand the spectrum of clinical features. <i>Journal of Endocrinological Investigation</i> , 2020, 43, 1739-1748.	3.3	3
62	Obstructive sleep apnea in children with Down syndrome: is it possible to predict severe apnea?. <i>European Journal of Pediatrics</i> , 2022, 181, 735-743.	2.7	3
63	Sleep disordered breathing in patients with Prader willi syndrome: Impact of underlying genetic mechanism. <i>Respiratory Medicine</i> , 2021, 187, 106567.	2.9	3
64	Anauxetic dysplasia: a rare clinical entity. <i>Turkish Journal of Pediatrics</i> , 2018, 60, 89.	0.6	3
65	Bilateral anterior segment dysgenesis in an infant with partial trisomy 16q and partial monosomy 3p. <i>Journal of AAPOS</i> , 2012, 16, 473-475.	0.3	2
66	Coexistence of Trisomy 13 and SRY (âˆ™) XX Ovotesticular Disorder of Sex Development. <i>Fetal and Pediatric Pathology</i> , 2017, 36, 445-451.	0.7	2
67	Mystery Case: Pontine tegmental cap dysplasia in a neonate. <i>Neurology</i> , 2018, 91, e2100-e2101.	1.1	2
68	Clinical and Molecular Spectrum of Four Patients Diagnosed with Mowat-Wilson Syndrome. <i>Molecular Syndromology</i> , 2020, 11, 296-301.	0.8	2
69	Genetic disorders with symptoms mimicking rheumatologic diseases: A single-center retrospective study. <i>European Journal of Medical Genetics</i> , 2021, 64, 104185.	1.3	2
70	A novel de novo mutation involving the MLL2 gene in a Kabuki syndrome patient presenting with seizures. <i>Turkish Journal of Pediatrics</i> , 2016, 58, 97-100.	0.6	2
71	Mucopolipidosis type III in an adolescent presenting with atypical facial features and skeletal deformities. <i>Genetic Counseling</i> , 2013, 24, 7-12.	0.1	2
72	A very rare case of a newborn with tetrasomy 9p and literature review. <i>Turkish Journal of Pediatrics</i> , 2022, 64, 171.	0.6	2

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73	Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions. <i>Molecular Syndromology</i> , 2022, 13, 537-542.	0.8	2
74	A Diagnosis to Consider in an Adult Patient with Facial Features and Intellectual Disability: Williams Syndrome. <i>Korean Journal of Family Medicine</i> , 2017, 38, 102.	1.2	1
75	Further delineation of spondyloepimetaphyseal dysplasia Fadenâ€Alkuraya type: A RSPRY1â€associated spondyloâ€epiâ€metaphyseal dysplasia with conoâ€brachydactyly and craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2009-2016.	1.2	1
76	Ophthalamo-acromelic syndrome in an infant. <i>European Journal of Medical Genetics</i> , 2019, 62, 103664.	1.3	1
77	Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20). <i>Molecular Syndromology</i> , 2020, 11, 38-42.	0.8	1
78	Three new cases of Crisponi /cold induced sweating syndrome (CS/CISS1) in Turkish families. <i>European Journal of Medical Genetics</i> , 2021, 64, 104229.	1.3	1
79	Main Physical Features, Echocardiographic and Renal Ultrasonographic Findings of Turner Syndrome in 107 Pediatric Patients. <i>Molecular Syndromology</i> , 2021, 12, 1-7.	0.8	1
80	Clinical and molecular evaluation of 16 patients with rett syndrome. <i>Turkish Journal of Pediatrics</i> , 2018, 60, 1.	0.6	1
81	Ä°skelet displazilerinde genel deÄ°ylendirme. <i>TÄ¼rk Ortopedi Ve Travmatoloji BirliÄ°yi DerneÄ°yi</i> , 2016, 15, .	0.1	1
82	Wildervanck syndrome: An uncommon cause of Duane syndrome. <i>Journal Francais D'Ophtalmologie</i> , 2014, 37, e123-e124.	0.4	0
83	Two adult siblings with progressive walking difficulty and visual disturbances. <i>Molecular Genetics and Metabolism</i> , 2015, 114, S108-S109.	1.1	0
84	Biallelic ITGB4 variants in familial pyloric atresia without epidermolysis bullosa: Report of two families with five siblings. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3427-3432.	1.2	0
85	The rare reason of pain in hip girdle: mucopolidosis type 3 gamma. <i>Turkish Journal of Pediatrics</i> , 2021, 63, 1091.	0.6	0
86	A de novo 11q23 deletion in a patient presenting with severe ophthalmologic findings, psychomotor retardation and facial dysmorphism. <i>Turkish Journal of Pediatrics</i> , 2014, 56, 80-4.	0.6	0
87	Recurrent fractures and an unusual diagnosis: Pycnodysostosis. <i>Journal of the National Medical Association</i> , 2022, 114, 295-297.	0.8	0
88	78. Menstruation Related Quality of Life in Adolescents With Genetic Syndromes Accompanying an Intellectual Disability. <i>Journal of Adolescent Health</i> , 2022, 70, S41.	2.5	0
89	Psychometric and Psychosocial Evaluation of Adolescents with Turner Syndrome in a Multidisciplinary Approach: A Preliminary Study. <i>Guncel Pediatri</i> , 2021, 19, 363-372.	0.1	0
90	Diagnostic distribution and postnatal evaluation of prenatally detected short femur: A single center experience. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	1.2	0