Pelin Ozlem Simsek Kiper

List of Publications by Year
in descending order

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	Obstructive sleep apnea in children with Down syndrome: is it possible to predict severe apnea?. European Journal of Pediatrics, 2022, 181, 735-743.	2.7	3
2	A very rare case of a newborn with tetrasomy 9p and literature review. Turkish Journal of Pediatrics, 2022, 64, 171.	0.6	2
3	HEATR3 variants impair nuclear import of uL18 (RPL5) and drive Diamond-Blackfan anemia. Blood, 2022, 139, 3111-3126.	1.4	15
4	Recurrent fractures and an unusual diagnosis: Pycnodysostosis. Journal of the National Medical Association, 2022, 114, 295-297.	0.8	0
5	78. Menstruation Related Quality of Life in Adolescents With Genetic Syndromes Accompanying an Intellectual Disability. Journal of Adolescent Health, 2022, 70, S41.	2.5	0
6	Diagnostic distribution and postnatal evaluation of prenatally detected short femur: A single center experience. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
7	Typical Face, Developmental Delay, and Hearing Loss in a Patient with 3M Syndrome: The Co-Occurrence of Two Rare Conditions. Molecular Syndromology, 2022, 13, 537-542.	0.8	2
8	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
9	Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience. Journal of Human Genetics, 2021, 66, 585-596.	2.3	4
10	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
11	Two Siblings with Kaufman Oculocerebrofacial Syndrome Resembling Oculoauriculovertebral Spectrum. Molecular Syndromology, 2021, 12, 106-111.	0.8	4
12	Diagnostic yield of wholeâ€exome sequencing in nonâ€syndromic intellectual disability. Journal of Intellectual Disability Research, 2021, 65, 577-588.	2.0	11
13	Further expanding the mutational spectrum of <scp>brain abnormalities, neurodegeneration, and dysosteosclerosis</scp>: A rare disorder with neurologic regression and skeletal features. American Journal of Medical Genetics, Part A, 2021, 185, 1888-1896.	1.2	14
14	Genetic disorders with symptoms mimicking rheumatologic diseases: A single-center retrospective study. European Journal of Medical Genetics, 2021, 64, 104185.	1.3	2
15	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
16	KohlschÃ¼tter-TÃ¶nz Syndrome With a Novel ROGD1 Variant in 3 Individuals: A Rare Clinical Entity. Journal of Child Neurology, 2021, 36, 816-822.	1.4	8
17	Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype. European Journal of Paediatric Neurology, 2021, 32, 46-55.	1.6	6
18	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

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19	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
20	Three new cases of Crisponi /cold induced sweating syndrome (CS/CISS1) in Turkish families. European Journal of Medical Genetics, 2021, 64, 104229.	1.3	1
21	Main Physical Features, Echocardiographic and Renal Ultrasonographic Findings of Turner Syndrome in 107 Pediatric Patients. Molecular Syndromology, 2021, 12, 1-7.	0.8	1
22	Biallelic ITGB4 variants in familial pyloric atresia without epidermolysis bullosa: Report of two families with five siblings. American Journal of Medical Genetics, Part A, 2021, 185, 3427-3432.	1.2	0
23	Investigation of Genetic Causes in a Developmental Disorder: Oculoauriculovertebral Spectrum. Cleft Palate-Craniofacial Journal, 2021, , 105566562110381.	0.9	5
24	Sleep disordered breathing in patients with Prader willi syndrome: Impact of underlying genetic mechanism. Respiratory Medicine, 2021, 187, 106567.	2.9	3
25	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
26	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
27	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
28	The rare reason of pain in hip girdle: mucolipidosis type 3 gamma. Turkish Journal of Pediatrics, 2021, 63, 1091.	0.6	0
29	Psychometric and Psychosocial Evaluation of Adolescents with Turner Syndrome in a Multidisiplinary Approach: A Preliminary Study. Guncel Pediatri, 2021, 19, 363-372.	0.1	0
30	Atypical Presentation of Sengers Syndrome: A Novel Mutation Revealed with Postmortem Genetic Testing. Fetal and Pediatric Pathology, 2020, 39, 163-171.	0.7	6
31	A Revisited Diagnosis of Collagen VI Related Muscular Dystrophy in a Patient with a Novel COL6A2 Variant and 21q22.3 Deletion. Neuropediatrics, 2020, 51, 445-449.	0.6	3
32	Hyperinsulinemic Hypoglycemia in a Patient with Costello Syndrome: An Etiology to Consider in Hypoglycemia. Molecular Syndromology, 2020, 11, 207-216.	0.8	5
33	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
34	Clinical and Molecular Spectrum of Four Patients Diagnosed with Mowat-Wilson Syndrome. Molecular Syndromology, 2020, 11, 296-301.	0.8	2
35	Further Phenotypic Delineation of Partial Trisomy 17q and Partial Monosomy 20q due to Rare t(17;20). Molecular Syndromology, 2020, 11, 38-42.	0.8	1
36	Genetic IGF1R defects: new cases expand the spectrum of clinical features. Journal of Endocrinological Investigation, 2020, 43, 1739-1748.	3.3	3

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37	Peters plus syndrome: a recognizable clinical entity. Turkish Journal of Pediatrics, 2020, 62, 136.	0.6	6
38	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
39	Ophthalmo-acromelic syndrome in an infant. European Journal of Medical Genetics, 2019, 62, 103664.	1.3	1
40	Further expanding the mutational spectrum and investigation of genotypeâ€“phenotype correlation in 3M syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1157-1172.	1.2	14
41	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	6.2	92
42	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
43	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
44	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
45	Intrafamilial variability of XYLT2-related spondyloocular syndrome. European Journal of Medical Genetics, 2019, 62, 103585.	1.3	9
46	A novel NKX3-2 mutation associated with perinatal lethal phenotype of spondylo-megaepiphyseal-metaphyseal dysplasia in a neonate. European Journal of Medical Genetics, 2019, 62, 21-26.	1.3	4
47	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
48	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
49	Mystery Case: Pontine tegmental cap dysplasia in a neonate. Neurology, 2018, 91, e2100-e2101.	1.1	2
50	Further delineation of spondyloepimetaphyseal dysplasia Fadenâ€Alkuraya type: A RSPRY1â€associated spondyloâ€epiâ€metaphyseal dysplasia with conoâ€brachydactyly and craniosynostosis. American Journal of Medical Genetics, Part A, 2018, 176, 2009-2016.	1.2	1
51	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	2.8	18
52	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
53	Clinical and molecular evaluation of 16 patients with rett syndrome. Turkish Journal of Pediatrics, 2018, 60, 1.	0.6	1
54	Anauxetic dysplasia: a rare clinical entity. Turkish Journal of Pediatrics, 2018, 60, 89.	0.6	3

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55	Epigenotype and phenotype correlations in patients with beckwith-wiedemann syndrome. Turkish Journal of Pediatrics, 2018, 60, 506.	0.6	4
56	HERC1 mutations in idiopathic intellectual disability. European Journal of Medical Genetics, 2017, 60, 279-283.	1.3	37
57	Homozygous indel mutation in CDH11 as the probable cause of Elsahyâ€“Waters syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3143-3152.	1.2	13
58	Coexistence of Trisomy 13 and SRY (â^') XX Ovotesticular Disorder of Sex Development. Fetal and Pediatric Pathology, 2017, 36, 445-451.	0.7	2
59	A Diagnosis to Consider in an Adult Patient with Facial Features and Intellectual Disability: Williams Syndrome. Korean Journal of Family Medicine, 2017, 38, 102.	1.2	1
60	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
61	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
62	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	27.0	119
63	Expanding the clinical and mutational spectrum of the Ehlersâ€“Danlos syndrome, dermatosparaxis type. Genetics in Medicine, 2016, 18, 882-891.	2.4	37
64	A novel de novo mutation involving the MLL2 gene in a Kabuki syndrome patient presenting with seizures. Turkish Journal of Pediatrics, 2016, 58, 97-100.	0.6	2
65	Ä°skelet displazilerinde genel deÄŸerlendirme. TÃ¼rk Ortopedi Ve Travmatoloji BirliÄŸi DerneÄŸi, 2016, 15, .	0.1	1
66	Experience of a skeletal dysplasia registry in Turkey: A fiveâ€years retrospective analysis. American Journal of Medical Genetics, Part A, 2015, 167, 2065-2074.	1.2	10
67	Two adult siblings with progressive walking difficulty and visual disturbances. Molecular Genetics and Metabolism, 2015, 114, S108-S109.	1.1	0
68	Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Human Genetics, 2015, 134, 553-568.	3.8	59
69	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
70	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
71	Positive effects of an angiotensin II type 1 receptor antagonist in Camuratiâ€“Engelmann disease: A single case observation. American Journal of Medical Genetics, Part A, 2014, 164, 2667-2671.	1.2	21
72	TMCO1 deficiency causes autosomal recessive cerebrofaciothoracic dysplasia. American Journal of Medical Genetics, Part A, 2014, 164, 291-304.	1.2	25

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73	Partial monosomy 3q26.33-3q27.3 presenting with intellectual disability, facial dysmorphism, and diaphragm eventration. Clinical Dysmorphology, 2014, 23, 147-151.	0.3	3
74	Wildervanck syndrome: An uncommon cause of Duane syndrome. Journal Francais D'Ophtalmologie, 2014, 37, e123-e124.	0.4	0
75	Barraquerâ€“Simons syndrome: A rare clinical entity. American Journal of Medical Genetics, Part A, 2014, 164, 1756-1760.	1.2	5
76	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
77	Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features. Orphanet Journal of Rare Diseases, 2014, 9, 60.	2.7	35
78	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
79	Etiological yield of SNP microarrays in idiopathic intellectual disability. European Journal of Paediatric Neurology, 2014, 18, 327-337.	1.6	26
80	A de novo 11q23 deletion in a patient presenting with severe ophthalmologic findings, psychomotor retardation and facial dysmorphism. Turkish Journal of Pediatrics, 2014, 56, 80-4.	0.6	0
81	A Homozygous Deletion in GRID2 Causes a Human Phenotype With Cerebellar Ataxia and Atrophy. Journal of Child Neurology, 2013, 28, 926-932.	1.4	67
82	Clinical and molecular analysis of RASopathies in a group of Turkish patients. Clinical Genetics, 2013, 83, 181-186.	2.0	24
83	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	2.9	190
84	Functional analysis of a duplication (p.E63_D69dup) in the switch II region of HRAS: new aspects of the molecular pathogenesis underlying Costello syndrome. Human Molecular Genetics, 2013, 22, 1643-1653.	2.9	26
85	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
86	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	2.5	29
87	Mucolipidosis type III in an adolescent presenting with atypical facial features and skeletal deformities. Genetic Counseling, 2013, 24, 7-12.	0.1	2
88	Bilateral anterior segment dysgenesis in an infant with partial trisomy 16q and partial monosomy 3p. Journal of AAPOS, 2012, 16, 473-475.	0.3	2
89	A mutation screen in patients with Kabuki syndrome. Human Genetics, 2011, 130, 715-724.	3.8	106
90	Catelâ€“Manzke syndrome: A clinical report suggesting autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2011, 155, 2288-2292.	1.2	7