Zehra Oya Uyguner

List of Publications by Year
in descending order

33	427	933447 10	794594 19
papers	citations	h-index	g-index

34	34	34	1079
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional loss of ubiquitinâ€specific protease 14 may lead to a novel distal arthrogryposis phenotype. Clinical Genetics, 2022, 101, 421-428.	2.0	1
2	Longâ€term followâ€up findings in a Turkish girl with osteogenesis imperfecta type <scp>XX</scp> caused by a homozygous <scp>MESD</scp> variant. American Journal of Medical Genetics, Part A, 2022, 188, 1639-1646.	1.2	2
3	Clinical and molecular evaluation of MEFV gene variants in the Turkish population: a study by the National Genetics Consortium. Functional and Integrative Genomics, 2022, 22, 291-315.	3.5	7
4	Evaluation of growth, puberty, osteoporosis, and the response to longâ€term bisphosphonate therapy in four patients with osteoporosisâ€pseudoglioma syndrome. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
5	A novel PSEN2 p.Ser175Phe variant in a family with Alzheimerâ€™s disease. Neurological Sciences, 2021, 42, 2497-2504.	1.9	1
6	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 2488-2495.	1.2	8
7	Long-Term Follow-Up Outcomes of 19 Patients with Osteogenesis Imperfecta Type XI and Bruck Syndrome Type I Caused by FKBP10 Variants. Calcified Tissue International, 2021, 109, 633-644.	3.1	4
8	Clinical and molecular genetic findings of hereditary Parkinson's patients from Turkey. Parkinsonism and Related Disorders, 2021, 93, 35-39.	2.2	1
9	Different phenotypes of transthyretin-associated familial amyloid polyneuropathy (TTR-FAP) due to a mutation in p.Glu109Gln in members of the same family. Ä°stanbul Kuzey Klinikleri, 2020, 8, 398-401.	0.3	0
10	Clinical and Genetic Investigation of Premature Ovarian Insufficiency Cases from Turkey. Journal of Gynecology Obstetrics and Human Reproduction, 2019, 48, 817-823.	1.3	6
11	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. Neuromuscular Disorders, 2019, 29, 601-613.	0.6	18
12	Mutation spectrum and pivotal features for differential diagnosis of Mucopolysaccharidosis IVA patients with severe and attenuated phenotype. Gene, 2019, 704, 59-67.	2.2	7
13	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. Journal of Pediatrics, 2019, 204, 305-313.e14.	1.8	24
14	Prevalence, clinical characteristics and long-term outcomes of classical 11 Î²-hydroxylase deficiency (11BOHD) in Turkish population and novel mutations in CYP11B1 gene. Journal of Steroid Biochemistry and Molecular Biology, 2018, 181, 88-97.	2.5	23
15	Precocious or early puberty in patients with combined pituitary hormone deficiency due to POU1F1 gene mutation: case report and review of possible mechanisms. Hormones, 2018, 17, 581-588.	1.9	13
16	Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. Molecular Cytogenetics, 2018, 11, 45.	0.9	18
17	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 2018, 557, 564-569.	27.8	141
18	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	3.8	24

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19	Ungual squamous cell carcinoma in a patient with Mal de Meleda. JDDG - Journal of the German Society of Dermatology, 2016, 14, 514-516.	0.8	1
20	Unguales Plattenepithelkarzinom bei einem Patienten mit Mal de Meleda. JDDG - Journal of the German Society of Dermatology, 2016, 14, 514-516.	0.8	2
21	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	1.2	55
22	Idiopathic angioedema with F12 mutation: is it a new entity?. Annals of Allergy, Asthma and Immunology, 2015, 114, 154-156.	1.0	10
23	A new hereditary congenital facial palsy case supports arg5 in HOX-DNA binding domain as possible hot spot for mutations. European Journal of Medical Genetics, 2015, 58, 358-363.	1.3	9
24	Twins with hereditary sensory and autonomic neuropathy type IV with preserved periodontal sensation. European Journal of Medical Genetics, 2014, 57, 240-246.	1.3	9
25	Galactosemia and phantom absence seizures. Journal of Pediatric Neurosciences, 2014, 9, 253.	0.3	1
26	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 308.	3.8	0
27	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309.	3.8	0
28	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309.	3.8	0
29	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309-10.	3.8	0
30	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 310.	3.8	0
31	Gene symbol: COL11A2. Disease: Otospondylomegaepiphyseal dysplasia. Human Genetics, 2008, 124, 310-1.	3.8	0
32	A new locus for autosomal recessive non-syndromic mental retardation maps to 1p21.1-p13.3. Clinical Genetics, 2007, 71, 212-219.	2.0	19
33	The R110C mutation in Notch3 causes variable clinical features in two Turkish families with CADASIL syndrome. Journal of the Neurological Sciences, 2006, 246, 123-130.	0.6	15