

Zehra Oya Uyguner

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

427
citations

933447

10
h-index

794594

19
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34
all docs

34
docs citations

34
times ranked

1079
citing authors

#	ARTICLE	IF	CITATIONS
1	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. <i>Nature</i> , 2018, 557, 564-569.	27.8	141
2	Mutations in <i>CDK5</i> & <i>RAP2</i> cause Seckel syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 467-480.	1.2	55
3	Identification of likely pathogenic and known variants in <i>TSPEAR</i> , <i>LAMB3</i> , <i>BCOR</i> , and <i>WNT10A</i> in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	3.8	24
4	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. <i>Journal of Pediatrics</i> , 2019, 204, 305-313.e14.	1.8	24
5	Prevalence, clinical characteristics and long-term outcomes of classical 11 β -hydroxylase deficiency (11BOHD) in Turkish population and novel mutations in <i>CYP11B1</i> gene. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2018, 181, 88-97.	2.5	23
6	A new locus for autosomal recessive non-syndromic mental retardation maps to 1p21.1-p13.3. <i>Clinical Genetics</i> , 2007, 71, 212-219.	2.0	19
7	Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases. <i>Molecular Cytogenetics</i> , 2018, 11, 45.	0.9	18
8	Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling. <i>Neuromuscular Disorders</i> , 2019, 29, 601-613.	0.6	18
9	The R110C mutation in <i>Notch3</i> causes variable clinical features in two Turkish families with CADASIL syndrome. <i>Journal of the Neurological Sciences</i> , 2006, 246, 123-130.	0.6	15
10	Precocious or early puberty in patients with combined pituitary hormone deficiency due to <i>POU1F1</i> gene mutation: case report and review of possible mechanisms. <i>Hormones</i> , 2018, 17, 581-588.	1.9	13
11	Idiopathic angioedema with F12 mutation: is it a new entity?. <i>Annals of Allergy, Asthma and Immunology</i> , 2015, 114, 154-156.	1.0	10
12	Twins with hereditary sensory and autonomic neuropathy type IV with preserved periodontal sensation. <i>European Journal of Medical Genetics</i> , 2014, 57, 240-246.	1.3	9
13	A new hereditary congenital facial palsy case supports <i>arg5</i> in <i>HOX</i> -DNA binding domain as possible hot spot for mutations. <i>European Journal of Medical Genetics</i> , 2015, 58, 358-363.	1.3	9
14	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2488-2495.	1.2	8
15	Mutation spectrum and pivotal features for differential diagnosis of Mucopolysaccharidosis IVA patients with severe and attenuated phenotype. <i>Gene</i> , 2019, 704, 59-67.	2.2	7
16	Clinical and molecular evaluation of <i>MEFV</i> gene variants in the Turkish population: a study by the National Genetics Consortium. <i>Functional and Integrative Genomics</i> , 2022, 22, 291-315.	3.5	7
17	Clinical and Genetic Investigation of Premature Ovarian Insufficiency Cases from Turkey. <i>Journal of Gynecology Obstetrics and Human Reproduction</i> , 2019, 48, 817-823.	1.3	6
18	Long-Term Follow-Up Outcomes of 19 Patients with Osteogenesis Imperfecta Type XI and Bruck Syndrome Type I Caused by <i>FKBP10</i> Variants. <i>Calcified Tissue International</i> , 2021, 109, 633-644.	3.1	4

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19	Unguales Plattenepithelkarzinom bei einem Patienten mit Mal de Meleda. JDDG - Journal of the German Society of Dermatology, 2016, 14, 514-516.	0.8	2
20	Long-term follow-up findings in a Turkish girl with osteogenesis imperfecta type <sc>XX</sc> caused by a homozygous <sc><i>MESD</i></sc> variant. American Journal of Medical Genetics, Part A, 2022, 188, 1639-1646.	1.2	2
21	Evaluation of growth, puberty, osteoporosis, and the response to long-term bisphosphonate therapy in four patients with osteoporosis-pseudoglioma syndrome. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
22	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
23	A novel PSEN2 p.Ser175Phe variant in a family with Alzheimer's disease. Neurological Sciences, 2021, 42, 2497-2504.	1.9	1
24	Galactosemia and phantom absence seizures. Journal of Pediatric Neurosciences, 2014, 9, 253.	0.3	1
25	Clinical and molecular genetic findings of hereditary Parkinson's patients from Turkey. Parkinsonism and Related Disorders, 2021, 93, 35-39.	2.2	1
26	Functional loss of ubiquitin-specific protease 14 may lead to a novel distal arthrogryposis phenotype. Clinical Genetics, 2022, 101, 421-428.	2.0	1
27	Different phenotypes of transthyretin-associated familial amyloid polyneuropathy (TTR-FAP) due to a mutation in p.Glu109Gln in members of the same family. İstanbul Kuzey Klinikleri, 2020, 8, 398-401.	0.3	0
28	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 308.	3.8	0
29	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309.	3.8	0
30	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309.	3.8	0
31	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 309-10.	3.8	0
32	Gene symbol: SERPING1. Disease: Angioneurotic oedema. Human Genetics, 2008, 124, 310.	3.8	0
33	Gene symbol: COL11A2. Disease: Otospondylomegalepiphyseal dysplasia. Human Genetics, 2008, 124, 310-1.	3.8	0