

Aslihan Tolun

List of Publications by Year in descending order

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65
papers

3,280
citations

257357

24
h-index

155592

55
g-index

66
all docs

66
docs citations

66
times ranked

5999
citing authors

#	ARTICLE	IF	CITATIONS
1	Y-Chromosomal Diversity in Europe Is Clinal and Influenced Primarily by Geography, Rather than by Language. <i>American Journal of Human Genetics</i> , 2000, 67, 1526-1543.	2.6	519
2	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511.	6.0	466
3	Mutations in SLC34A2 Cause Pulmonary Alveolar Microlithiasis and Are Possibly Associated with Testicular Microlithiasis. <i>American Journal of Human Genetics</i> , 2006, 79, 650-656.	2.6	226
4	Mutation in ATG5 reduces autophagy and leads to ataxia with developmental delay. <i>ELife</i> , 2016, 5, .	2.8	161
5	DNAJC6 is responsible for juvenile parkinsonism with phenotypic variability. <i>Parkinsonism and Related Disorders</i> , 2013, 19, 320-324.	1.1	154
6	The Y-Chromosome Tree Bursts into Leaf: 13,000 High-Confidence SNPs Covering the Majority of Known Clades. <i>Molecular Biology and Evolution</i> , 2015, 32, 661-673.	3.5	137
7	Sequence of inverted terminal repetitions from different adenoviruses: Demonstration of conserved sequences and homology between SA7 termini and SV40 DNA. <i>Cell</i> , 1979, 17, 705-713.	13.5	131
8	Direct repeats of the F plasmid incC region express F incompatibility. <i>Cell</i> , 1981, 24, 687-694.	13.5	105
9	Truncating mutations in <i>TAF4B</i> and <i>ZMYND15</i> causing recessive azoospermia. <i>Journal of Medical Genetics</i> , 2014, 51, 239-244.	1.5	104
10	<i>TBC1D24</i> genotype-phenotype correlation. <i>Neurology</i> , 2016, 87, 77-85.	1.5	97
11	Two complementary strand-specific termination sites for adenovirus DNA replication. <i>Cell</i> , 1976, 9, 259-268.	13.5	76
12	Large-scale recent expansion of European patrilineages shown by population resequencing. <i>Nature Communications</i> , 2015, 6, 7152.	5.8	69
13	Recessive truncating <i>NALCN</i> mutation in infantile neuroaxonal dystrophy with facial dysmorphism. <i>Journal of Medical Genetics</i> , 2013, 50, 515-520.	1.5	66
14	Homozygous WNT10b mutation and complex inheritance in Split-Hand/Foot Malformation. <i>Human Molecular Genetics</i> , 2008, 17, 2644-2653.	1.4	65
15	<i>TBC1D24</i> truncating mutation resulting in severe neurodegeneration. <i>Journal of Medical Genetics</i> , 2013, 50, 199-202.	1.5	65
16	Control of ciliogenesis by FOR20, a novel centrosome and pericentriolar satellite protein. <i>Journal of Cell Science</i> , 2010, 123, 2391-2401.	1.2	61
17	A frameshift mutation of ERLIN2 in recessive intellectual disability, motor dysfunction and multiple joint contractures. <i>Human Molecular Genetics</i> , 2011, 20, 1886-1892.	1.4	59
18	Highest heterogeneity for cystic fibrosis: 36 mutations account for 75% of all CF chromosomes in Turkish patients. <i>American Journal of Medical Genetics Part A</i> , 2002, 113, 250-257.	2.4	48

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19	Autosomal recessive spastic paraplegia (SPG45) with mental retardation maps to 10q24.3â€“q25.1. <i>Neurogenetics</i> , 2009, 10, 325-331.	0.7	48
20	Novel NDE1 homozygous mutation resulting in microhydranencephaly and not microlyssencephaly. <i>Neurogenetics</i> , 2012, 13, 189-194.	0.7	40
21	Is the novel SCKL3 at 14q23 the predominant Seckel locus?. <i>European Journal of Human Genetics</i> , 2003, 11, 851-857.	1.4	38
22	The Novel Genetic Disorder Microhydranencephaly Maps to Chromosome 16p13.3-12.1. <i>American Journal of Human Genetics</i> , 2000, 66, 1705-1709.	2.6	32
23	The phenotype caused by <i>PYCR1</i> mutations corresponds to geroderma osteodysplasticum rather than autosomal recessive cutis laxa type 2. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 134-140.	0.7	31
24	Adult phenotype and further phenotypic variability in SRD5A3-CDG. <i>BMC Medical Genetics</i> , 2014, 15, 10.	2.1	28
25	Novel Recessive Cone-Rod Dystrophy Caused by <i>POC1B</i> Mutation. <i>JAMA Ophthalmology</i> , 2014, 132, 1185.	1.4	27
26	Neuromuscular endplate pathology in recessive desminopathies. <i>Neurology</i> , 2016, 87, 799-805.	1.5	26
27	Identification of a locus for an autosomal recessive hyaline body myopathy at chromosome 3p22.2â€“p21.32. <i>Neuromuscular Disorders</i> , 2004, 14, 4-9.	0.3	25
28	Homozygous MYH7 R1820W mutation results in recessive myosin storage myopathy: Scapulooperoneal and respiratory weakness with dilated cardiomyopathy. <i>Neuromuscular Disorders</i> , 2015, 25, 340-344.	0.3	25
29	Homozygous mutation in <i>CEP19</i> , a gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly. <i>Journal of Medical Genetics</i> , 2018, 55, 189-197.	1.5	25
30	Severe neurodegenerative disease in brothers with homozygous mutation in POLR1A. <i>European Journal of Human Genetics</i> , 2017, 25, 315-323.	1.4	23
31	Repair of Radiation-Induced Strand Breaks as Related to the Inducible Inhibitor of Postirradiation DNA Degradation. <i>Biophysical Journal</i> , 1974, 14, 691-696.	0.2	22
32	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. <i>Modern Rheumatology</i> , 2015, 25, 315-321.	0.9	21
33	A novel recessive GUCY2D mutation causing coneâ€“rod dystrophy and not Leber's congenital amaurosis. <i>European Journal of Human Genetics</i> , 2010, 18, 1121-1126.	1.4	20
34	Progressive SCAR14 with unclear speech, developmental delay, tremor, and behavioral problems caused by a homozygous deletion of the SPTBN2 pleckstrin homology domain. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2494-2499.	0.7	19
35	<i>LACC1</i> Gene Defects in Familial Form of Juvenile Arthritis. <i>Journal of Rheumatology</i> , 2018, 45, 726-728.	1.0	17
36	A deletion in DRCTNNB1A associated with hypomyelination and juvenile onset cataract. <i>European Journal of Human Genetics</i> , 2008, 16, 261-264.	1.4	15

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37	Severe Form of Cockayne Syndrome With Varying Clinical Presentation and No Photosensitivity in a Family. <i>Journal of Child Neurology</i> , 2006, 21, 333-337.	0.7	14
38	Early-Onset Progressive Myoclonic Epilepsy With Dystonia Mapping to 16pter-p13.3. <i>Journal of Neurogenetics</i> , 2010, 24, 207-215.	0.6	14
39	A homozygous frameshift mutation of sepiapterin reductase gene causing parkinsonism with onset in childhood. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 191-193.	1.1	13
40	A homozygous 237-kb deletion at 1p31 identified as the locus for midline cleft of the upper and lower lip in a consanguineous family. <i>European Journal of Human Genetics</i> , 2014, 22, 333-337.	1.4	13
41	Homozygous <i>CHST11</i> mutation in chondrodysplasia, brachydactyly, overriding digits, clino-symphalangism and synpolydactyly. <i>Journal of Medical Genetics</i> , 2018, 55, 489-496.	1.5	12
42	<i>STUB1</i> polyadenylation signal variant AACAAA does not affect polyadenylation but decreases <i>STUB1</i> translation causing SCAR16. <i>Human Mutation</i> , 2018, 39, 1344-1348.	1.1	11
43	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. <i>EMBO Journal</i> , 2022, 41, e105531.	3.5	11
44	Linked homozygous <i>BMPR1B</i> and <i>PDHA2</i> variants in a consanguineous family with complex digit malformation and male infertility. <i>European Journal of Human Genetics</i> , 2018, 26, 876-885.	1.4	10
45	Deletion pattern in the dystrophin gene in Turks and a comparison with Europeans and Indians. <i>Annals of Human Genetics</i> , 2000, 64, 33-40.	0.3	10
46	Familial microhydranencephaly, a family that does not map to 16p13.13-p12.2: relationship with hereditary fetal brain degeneration and fetal brain disruption sequence. <i>Clinical Dysmorphology</i> , 2010, 19, 107-118.	0.1	8
47	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. <i>Modern Rheumatology</i> , 2013, , 1.	0.9	8
48	CRADD and USP44 mutations in intellectual disability, mild lissencephaly, brain atrophy, developmental delay, strabismus, behavioural problems and skeletal anomalies. <i>European Journal of Medical Genetics</i> , 2021, 64, 104181.	0.7	8
49	A Novel ATP6V0A2 Mutation Causing Recessive Cutis Laxa with Unusual Manifestations of Bleeding Diathesis and Defective Wound Healing. <i>Turkish Journal of Haematology</i> , 2019, 36, 29-36.	0.2	7
50	Complex postaxial polydactyly types A and B with camptodactyly, hypoplastic third toe, zygodactyly and other digit anomalies caused by a novel <i>GLI3</i> mutation. <i>European Journal of Medical Genetics</i> , 2017, 60, 268-274.	0.7	6
51	Novel <i>EDAR</i> mutation in tooth agenesis and variable associated features. <i>European Journal of Medical Genetics</i> , 2020, 63, 103926.	0.7	6
52	Very late-onset pyridoxine-dependent seizures not linking to the known 5q31 locus. <i>Pediatrics International</i> , 2008, 50, 703-705.	0.2	5
53	<i>RBBP8</i> syndrome with microcephaly, intellectual disability, short stature and brachydactyly. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3148-3152.	0.7	5
54	Biallelic <i>ZNF407</i> mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. <i>Journal of Human Genetics</i> , 2020, 65, 1115-1123.	1.1	5

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55	The first adolescent case of Fraser syndrome 3, with a novel nonsense variant in <i>GRIP1</i> . American Journal of Medical Genetics, Part A, 2021, 185, 1858-1863.	0.7	4
56	DIRECT REPEATS OF NUCLEOTIDE SEQUENCES ARE INVOLVED IN PLASMID REPLICATION AND INCOMPATIBILITY. , 1981, , 113-124.		4
57	A Two-Base Pair Deletion in IQ Repeats in ASPM Underlies Microcephaly in a Pakistani Family. Genetic Testing and Molecular Biomarkers, 2022, 26, 37-42.	0.3	4
58	KERATIN 17-related recessive atypical pachyonychia congenita with variable hair and tooth anomalies. European Journal of Human Genetics, 2022, 30, 1292-1296.	1.4	4
59	Homozygous deletion of MYADML2 in cranial asymmetry, reduced bone maturation, multiple dislocations, lumbar lordosis, and prominent clavicles. Journal of Human Genetics, 2021, 66, 171-179.	1.1	2
60	Other autoinflammatory disease genes in an FMF-prevalent population: a homozygous MVK mutation and a novel heterozygous TNFRSF1A mutation in two different Turkish families with clinical FMF. Clinical and Experimental Rheumatology, 2017, 35 Suppl 108, 75-81.	0.4	2
61	FAM160B1 deficit associated with microcephaly, severe intellectual disability, ataxia, behavioral abnormalities and speech problems. Clinical Genetics, 2019, 96, 456-460.	1.0	1
62	A homozygous ROR2 variant in a family with atypical Robinow syndrome and tetramelic transverse deficiency of autopods. American Journal of Medical Genetics, Part A, 2021, , .	0.7	1
63	Plasmid Incompatibility and Replication. , 1985, , 1-7.		1
64	Response to Kouwenberg et al. "Recognizable Phenotype With Common Occurrence of Microcephaly, Psychomotor Retardation, But No Spontaneous Bone Fractures in ARCL2B Due to <i>PYCR1</i> Mutations" American Journal of Medical Genetics, Part A, 2011, 155, 2333-2334.	0.7	0
65	A role of autophagy in spinocerebellar ataxia"Rare exception or general principle?. Autophagy, 2016, 12, 1208-1209.	4.3	0