Aslihan Tolun

List of Publications by Year in descending order

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65 papers	3,280 citations	257450 24 h-index	155660 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. American Journal of Human Genetics, 2000, 67, 1526-1543.	6.2	519
2	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	12.6	466
3	Mutations in SLC34A2 Cause Pulmonary Alveolar Microlithiasis and Are Possibly Associated with Testicular Microlithiasis. American Journal of Human Genetics, 2006, 79, 650-656.	6.2	226
4	Mutation in ATG5 reduces autophagy and leads to ataxia with developmental delay. ELife, 2016, 5, .	6.0	161
5	DNAJC6 is responsible for juvenile parkinsonism with phenotypic variability. Parkinsonism and Related Disorders, 2013, 19, 320-324.	2.2	154
6	The Y-Chromosome Tree Bursts into Leaf: 13,000 High-Confidence SNPs Covering the Majority of Known Clades. Molecular Biology and Evolution, 2015, 32, 661-673.	8.9	137
7	Sequence of inverted terminal repetitions from different adenoviruses: Demonstration of conserved sequences and homology between SA7 termini and SV40 DNA. Cell, 1979, 17, 705-713.	28.9	131
8	Direct repeats of the F plasmid incC region express F incompatibility. Cell, 1981, 24, 687-694.	28.9	105
9	Truncating mutations inTAF4BandZMYND15causing recessive azoospermia. Journal of Medical Genetics, 2014, 51, 239-244.	3.2	104
10	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
11	Two complementary strand-specific termination sites for adenovirus DNA replication. Cell, 1976, 9, 259-268.	28.9	76
12	Large-scale recent expansion of European patrilineages shown by population resequencing. Nature Communications, 2015, 6, 7152.	12.8	69
13	Recessive truncatingNALCNmutation in infantile neuroaxonal dystrophy with facial dysmorphism. Journal of Medical Genetics, 2013, 50, 515-520.	3.2	66
14	Homozygous WNT10b mutation and complex inheritance in Split-Hand/Foot Malformation. Human Molecular Genetics, 2008, 17, 2644-2653.	2.9	65
15	TBC1D24truncating mutation resulting in severe neurodegeneration. Journal of Medical Genetics, 2013, 50, 199-202.	3.2	65
16	Control of ciliogenesis by FOR20, a novel centrosome and pericentriolar satellite protein. Journal of Cell Science, 2010, 123, 2391-2401.	2.0	61
17	A frameshift mutation of ERLIN2 in recessive intellectual disability, motor dysfunction and multiple joint contractures. Human Molecular Genetics, 2011, 20, 1886-1892.	2.9	59
18	Highest heterogeneity for cystic fibrosis: 36 mutations account for 75% of all CF chromosomes in Turkish patients. American Journal of Medical Genetics Part A, 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. Neurogenetics, 2009, 10, 325-331.	1.4	48
20	Novel NDE1 homozygous mutation resulting in microhydranencephaly and not microlyssencephaly. Neurogenetics, 2012, 13, 189-194.	1.4	40
21	Is the novel SCKL3 at 14q23 the predominant Seckel locus?. European Journal of Human Genetics, 2003, 11, 851-857.	2.8	38
22	The Novel Genetic Disorder Microhydranencephaly Maps to Chromosome 16p13.3-12.1. American Journal of Human Genetics, 2000, 66, 1705-1709.	6.2	32
23	The phenotype caused by PYCR1 mutations corresponds to geroderma osteodysplasticum rather than autosomal recessive cutis laxa type 2. American Journal of Medical Genetics, Part A, 2011, 155, 134-140.	1.2	31
24	Adult phenotype and further phenotypic variability in SRD5A3-CDG. BMC Medical Genetics, 2014, 15, 10.	2.1	28
25	Novel Recessive Cone-Rod Dystrophy Caused byPOC1BMutation. JAMA Ophthalmology, 2014, 132, 1185.	2.5	27
26	Neuromuscular endplate pathology in recessive desminopathies. Neurology, 2016, 87, 799-805.	1.1	26
27	Identification of a locus for an autosomal recessive hyaline body myopathy at chromosome 3p22.2â€“p21.32. Neuromuscular Disorders, 2004, 14, 4-9.	0.6	25
28	Homozygous MYH7 R1820W mutation results in recessive myosin storage myopathy: Scapuloperoneal and respiratory weakness with dilated cardiomyopathy. Neuromuscular Disorders, 2015, 25, 340-344.	0.6	25
29	Homozygous mutation inCEP19,a gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly. Journal of Medical Genetics, 2018, 55, 189-197.	3.2	25
30	Severe neurodegenerative disease in brothers with homozygous mutation in POLR1A. European Journal of Human Genetics, 2017, 25, 315-323.	2.8	23
31	Repair of Radiation-Induced Strand Breaks as Related to the Inducible Inhibitor of Postirradiation DNA Degradation. Biophysical Journal, 1974, 14, 691-696.	0.5	22
32	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. Modern Rheumatology, 2015, 25, 315-321.	1.8	21
33	A novel recessive GUCY2D mutation causing coneâ€“rod dystrophy and not Leber's congenital amaurosis. European Journal of Human Genetics, 2010, 18, 1121-1126.	2.8	20
34	Progressive SCAR14 with unclear speech, developmental delay, tremor, and behavioral problems caused by a homozygous deletion of the SPTBN2 pleckstrin homology domain. American Journal of Medical Genetics, Part A, 2017, 173, 2494-2499.	1.2	19
35	LACC1 Gene Defects in Familial Form of Juvenile Arthritis. Journal of Rheumatology, 2018, 45, 726-728.	2.0	17
36	A deletion in DRCTNNB1A associated with hypomyelination and juvenile onset cataract. European Journal of Human Genetics, 2008, 16, 261-264.	2.8	15

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37	Severe Form of Cockayne Syndrome With Varying Clinical Presentation and No Photosensitivity in a Family. Journal of Child Neurology, 2006, 21, 333-337.	1.4	14
38	Early-Onset Progressive Myoclonic Epilepsy With Dystonia Mapping to 16pter-p13.3. Journal of Neurogenetics, 2010, 24, 207-215.	1.4	14
39	A homozygous frameshift mutation of sepiapterin reductase gene causing parkinsonism with onset in childhood. Parkinsonism and Related Disorders, 2012, 18, 191-193.	2.2	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. European Journal of Human Genetics, 2014, 22, 333-337.	2.8	13
41	Homozygous CHST11 mutation in chondrodysplasia, brachydactyly, overriding digits, clino-symphalangism and synpolydactyly. Journal of Medical Genetics, 2018, 55, 489-496.	3.2	12
42	STUB1polyadenylation signal variant AACAAA does not affect polyadenylation but decreasesSTUB1translation causing SCAR16. Human Mutation, 2018, 39, 1344-1348.	2.5	11
43	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. EMBO Journal, 2022, 41, e105531.	7.8	11
44	Linked homozygous BMPR1B and PDHA2 variants in a consanguineous family with complex digit malformation and male infertility. European Journal of Human Genetics, 2018, 26, 876-885.	2.8	10
45	Deletion pattern in the dystrophin gene in Turks and a comparison with Europeans and Indians. Annals of Human Genetics, 2000, 64, 33-40.	0.8	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. Clinical Dysmorphology, 2010, 19, 107-118.	0.3	8
47	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. Modern Rheumatology, 2013, , 1.	1.8	8
48	CRADD and USP44 mutations in intellectual disability, mild lissencephaly, brain atrophy, developmental delay, strabismus, behavioural problems and skeletal anomalies. European Journal of Medical Genetics, 2021, 64, 104181.	1.3	8
49	A Novel ATP6V0A2 Mutation Causing Recessive Cutis Laxa with Unusual Manifestations of Bleeding Diathesis and Defective Wound Healing. Turkish Journal of Haematology, 2019, 36, 29-36.	0.5	7
50	Complex postaxial polydactyly types A and B with camptodactyly, hypoplastic third toe, zygodactyly and other digit anomalies caused by a novel GLI3 mutation. European Journal of Medical Genetics, 2017, 60, 268-274.	1.3	6
51	Novel EDAR mutation in tooth agenesis and variable associated features. European Journal of Medical Genetics, 2020, 63, 103926.	1.3	6
52	Veryâ€lateâ€onset pyridoxineâ€dependent seizures not linking to the known 5q31 locus. Pediatrics International, 2008, 50, 703-705.	0.5	5
53	RBBP8 syndrome with microcephaly, intellectual disability, short stature and brachydactyly. American Journal of Medical Genetics, Part A, 2015, 167, 3148-3152.	1.2	5
54	Biallelic ZNF407 mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. Journal of Human Genetics, 2020, 65, 1115-1123.	2.3	5

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55	The first adolescent case of Fraser syndrome 3, with a novel nonsense variant in GRIP1. American Journal of Medical Genetics, Part A, 2021, 185, 1858-1863.	1.2	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.	0.7	4
58	KERATIN 17-related recessive atypical pachyonychia congenita with variable hair and tooth anomalies. European Journal of Human Genetics, 2022, 30, 1292-1296.	2.8	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.	2.3	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.	0.8	2
61	FAM160B1 deficit associated with microcephaly, severe intellectual disability, ataxia, behavioral abnormalities and speech problems. Clinical Genetics, 2019, 96, 456-460.	2.0	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, , .	1.2	1
63	Plasmid Incompatibility and Replication. , 1985, , 1-7.		1
64	Response to Kouwenberg et al. â€œRecognizable Phenotype With Common Occurrence of Microcephaly, Psychomotor Retardation, But No Spontaneous Bone Fractures in ARCL2B Due to PYCR1 Mutationsâ€• American Journal of Medical Genetics, Part A, 2011, 155, 2333-2334.	1.2	0
65	A role of autophagy in spinocerebellar ataxiaâ€”Rare exception or general principle?. Autophagy, 2016, 12, 1208-1209.	9.1	0