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	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
2	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. EMBO Journal, 2022, 41, e105531.	7.8	11
3	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
4	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
5	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
6	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
7	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
8	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
9	Novel EDAR mutation in tooth agenesis and variable associated features. European Journal of Medical Genetics, 2020, 63, 103926.	1.3	6
10	FAM160B1 deficit associated with microcephaly, severe intellectual disability, ataxia, behavioral abnormalities and speech problems. Clinical Genetics, 2019, 96, 456-460.	2.0	1
11	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
12	Homozygous CHST11 mutation in chondrodysplasia, brachydactyly, overriding digits, clino-symphalangism and synpolydactyly. Journal of Medical Genetics, 2018, 55, 489-496.	3.2	12
13	LACC1 Gene Defects in Familial Form of Juvenile Arthritis. Journal of Rheumatology, 2018, 45, 726-728.	2.0	17
14	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
15	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
16	STUB1polyadenylation signal variant AACAAA does not affect polyadenylation but decreasesSTUB1translation causing SCAR16. Human Mutation, 2018, 39, 1344-1348.	2.5	11
17	Severe neurodegenerative disease in brothers with homozygous mutation in POLR1A. European Journal of Human Genetics, 2017, 25, 315-323.	2.8	23
18	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

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19	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
20	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
21	A role of autophagy in spinocerebellar ataxiaâ€"Rare exception or general principle?. Autophagy, 2016, 12, 1208-1209.	9.1	0
22	Neuromuscular endplate pathology in recessive desminopathies. Neurology, 2016, 87, 799-805.	1.1	26
23	TBC1D24 genotypeâ€"phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
24	Mutation in ATG5 reduces autophagy and leads to ataxia with developmental delay. ELife, 2016, 5, .	6.0	161
25	RBBP8 syndrome with microcephaly, intellectual disability, short stature and brachydactyly. American Journal of Medical Genetics, Part A, 2015, 167, 3148-3152.	1.2	5
26	Large-scale recent expansion of European patrilineages shown by population resequencing. Nature Communications, 2015, 6, 7152.	12.8	69
27	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. Modern Rheumatology, 2015, 25, 315-321.	1.8	21
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	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
30	Novel Recessive Cone-Rod Dystrophy Caused byPOC1BMutation. JAMA Ophthalmology, 2014, 132, 1185.	2.5	27
31	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
32	Adult phenotype and further phenotypic variability in SRD5A3-CDG. BMC Medical Genetics, 2014, 15, 10.	2.1	28
33	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	12.6	466
34	Truncating mutations inTAF4BandZMYND15causing recessive azoospermia. Journal of Medical Genetics, 2014, 51, 239-244.	3.2	104
35	A novel recessive 15-hydroxyprostaglandin dehydrogenase mutation in a family with primary hypertrophic osteoarthropathy. Modern Rheumatology, 2013, , 1.	1.8	8
36	DNAJC6 is responsible for juvenile parkinsonism with phenotypic variability. Parkinsonism and Related Disorders, 2013, 19, 320-324.	2.2	154

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37	TBC1D24truncating mutation resulting in severe neurodegeneration. Journal of Medical Genetics, 2013, 50, 199-202.	3.2	65
38	Recessive truncatingNALCNmutation in infantile neuroaxonal dystrophy with facial dysmorphism. Journal of Medical Genetics, 2013, 50, 515-520.	3.2	66
39	Novel NDE1 homozygous mutation resulting in microhydranencephaly and not microlyssencephaly. Neurogenetics, 2012, 13, 189-194.	1.4	40
40	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
41	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
42	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
43	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
44	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
45	Control of ciliogenesis by FOR20, a novel centrosome and pericentriolar satellite protein. Journal of Cell Science, 2010, 123, 2391-2401.	2.0	61
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	Early-Onset Progressive Myoclonic Epilepsy With Dystonia Mapping to 16pter-p13.3. Journal of Neurogenetics, 2010, 24, 207-215.	1.4	14
48	Autosomal recessive spastic paraplegia (SPG45) with mental retardation maps to 10q24.3â€“q25.1. Neurogenetics, 2009, 10, 325-331.	1.4	48
49	A deletion in DRCTNNB1A associated with hypomyelination and juvenile onset cataract. European Journal of Human Genetics, 2008, 16, 261-264.	2.8	15
50	Veryâ€lateâ€onset pyridoxineâ€dependent seizures not linking to the known 5q31 locus. Pediatrics International, 2008, 50, 703-705.	0.5	5
51	Homozygous WNT10b mutation and complex inheritance in Split-Hand/Foot Malformation. Human Molecular Genetics, 2008, 17, 2644-2653.	2.9	65
52	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
53	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
54	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

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55	Is the novel SCKL3 at 14q23 the predominant Seckel locus?. European Journal of Human Genetics, 2003, 11, 851-857.	2.8	38
56	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
57	The Novel Genetic Disorder Microhydranencephaly Maps to Chromosome 16p13.3-12.1. American Journal of Human Genetics, 2000, 66, 1705-1709.	6.2	32
58	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
59	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
60	Plasmid Incompatibility and Replication. , 1985, , 1-7.		1
61	Direct repeats of the F plasmid incC region express F incompatibility. Cell, 1981, 24, 687-694.	28.9	105
62	DIRECT REPEATS OF NUCLEOTIDE SEQUENCES ARE INVOLVED IN PLASMID REPLICATION AND INCOMPATIBILITY. , 1981, , 113-124.		4
63	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
64	Two complementary strand-specific termination sites for adenovirus DNA replication. Cell, 1976, 9, 259-268.	28.9	76
65	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