Davor Lessel

List of Publications by Year
in descending order

85 papers	5,177 citations	134610 34 h-index	120465 65 g-index
102 all docs	102 docs citations	102 times ranked	13264 citing authors

#	Article	IF	Citations
1	Werner syndrome in a Lebanese family. American Journal of Medical Genetics, Part A, 2022, , .	0.7	3
2	Aggressive variants of prostate cancer: underlying mechanisms of neuroendocrine transdifferentiation. Journal of Experimental and Clinical Cancer Research, 2022, 41, 46.	3.5	43
3	Biallelic CACNA2D1 loss-of-function variants cause early-onset developmental epileptic encephalopathy. Brain, 2022, 145, 2721-2729.	3.7	15
4	Biallelic PAN2 variants in individuals with a syndromic neurodevelopmental disorder and multiple congenital anomalies. European Journal of Human Genetics, 2022, 30, 611-618.	1.4	4
5	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males. Human Genetics, 2022, 141, 257-272.	1.8	8
6	Intake Patterns of Specific Alcoholic Beverages by Prostate Cancer Status. Cancers, 2022, 14, 1981.	1.7	0
7	Association Study between Polymorphisms in DNA Methylationâ€“Related Genes and Testicular Germ Cell Tumor Risk. Cancer Epidemiology Biomarkers and Prevention, 2022, 31, 1769-1779.	1.1	4
8	Brain Abnormalities in Patients with Germline Variants in H3F3: Novel Imaging Findings and Neurologic Symptoms Beyond Somatic Variants and Brain Tumors. American Journal of Neuroradiology, 2022, 43, 1048-1053.	1.2	2
9	Trans-ancestry genome-wide association meta-analysis of prostate cancer identifies new susceptibility loci and informs genetic risk prediction. Nature Genetics, 2021, 53, 65-75.	9.4	264
10	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
11	A Novel Homozygous WRN Mutation Identified in a Middle Aged Man With Diabetes Mellitus Complicated By Multiple Features of Accelerated Aging. Journal of the Endocrine Society, 2021, 5, A361-A361.	0.1	0
12	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	3.6	16
13	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
14	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269.	0.7	8
15	Identification of 22 susceptibility loci associated with testicular germ cell tumors. Nature Communications, 2021, 12, 4487.	5.8	27
16	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. Clinical Genetics, 2021, 100, 766-770.	1.0	5
17	Whole-Exome Sequencing in Critically Ill Neonates and Infants: Diagnostic Yield and Predictability of Monogenic Diagnosis. Neonatology, 2021, 118, 454-461.	0.9	16
18	A novel homozygous synonymous variant further expands the phenotypic spectrum of POLR3A â€•related pathologies. American Journal of Medical Genetics, Part A, 2021, 188, 216.	0.7	5

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19	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
20	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
21	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43
22	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
23	The CHEK2 Variant C.349A>G Is Associated with Prostate Cancer Risk and Carriers Share a Common Ancestor. Cancers, 2020, 12, 3254.	1.7	16
24	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
25	Nine newly identified individuals refine the phenotype associated with MYT1L mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1021-1031.	0.7	19
26	Two novel cases further expand the phenotype of TOR1AIP1-associated nuclear envelopathies. Human Genetics, 2020, 139, 483-498.	1.8	11
27	Genetic identification of cell types underlying brain complex traits yields insights into the etiology of Parkinsonâ€™s disease. Nature Genetics, 2020, 52, 482-493.	9.4	216
28	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. International Journal of Epidemiology, 2020, 49, 1022-1031.	0.9	34
29	Mutations in genes encoding regulators of mRNA decapping and translation initiation: links to intellectual disability. Biochemical Society Transactions, 2020, 48, 1199-1211.	1.6	9
30	Abstract 1203: Identification of 22 novel loci associated with susceptibility to testicular germ cell tumors. , 2020, , .		1
31	Runs of homozygosity and testicular cancer risk. Andrology, 2019, 7, 555-564.	1.9	5
32	SPRTN protease and checkpoint kinase 1 cross-activation loop safeguards DNA replication. Nature Communications, 2019, 10, 3142.	5.8	36
33	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
34	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
35	Association of Inherited Pathogenic Variants in Checkpoint Kinase 2 (CHEK2) With Susceptibility to Testicular Germ Cell Tumors. JAMA Oncology, 2019, 5, 514.	3.4	43
36	Fatal Myelotoxicity Following Palliative Chemotherapy With Cisplatin and Gemcitabine in a Patient With Stage IV Cholangiocarcinoma Linked to Post Mortem Diagnosis of Fanconi Anemia. Frontiers in Oncology, 2019, 9, 420.	1.3	14

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37	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. American Journal of Human Genetics, 2019, 104, 749-757.	2.6	41
38	Circulating Metabolic Biomarkers of Screen-Detected Prostate Cancer in the ProtecT Study. Cancer Epidemiology Biomarkers and Prevention, 2019, 28, 208-216.	1.1	21
39	A recurrent de-novo ANO3 mutation causes early-onset generalized dystonia. Journal of the Neurological Sciences, 2019, 396, 199-201.	0.3	13
40	Genome-wide association analysis suggests novel loci for Hashimotoâ€™s thyroiditis. Journal of Endocrinological Investigation, 2019, 42, 567-576.	1.8	17
41	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	1.1	68
42	Exome Sequencing in Children. Deutsches Ärzteblatt International, 2019, 116, 197-204.	0.6	25
43	Hereditary Syndromes With Signs of Premature Aging. Deutsches Ärzteblatt International, 2019, 116, 489-496.	0.6	12
44	Abstract 2684: Identification of 14 novel genetic loci for testicular germ cell tumor susceptibility. , 2019, , .		0
45	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
46	Genome-wide association study identifies susceptibility loci for B-cell childhood acute lymphoblastic leukemia. Nature Communications, 2018, 9, 1340.	5.8	58
47	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. Neuron, 2018, 98, 743-753.e4.	3.8	63
48	Analyses of LMNA-negative juvenile progeroid cases confirms biallelic POLR3A mutations in Wiedemannâ€“Rautenstrauch-like syndrome and expands the phenotypic spectrum of PYCR1 mutations. Human Genetics, 2018, 137, 921-939.	1.8	17
49	<scp>CTC</scp>1 mutations in a Brazilian family with progeroid features and recurrent bone fractures. Molecular Genetics & Genomic Medicine, 2018, 6, 1148-1156.	0.6	19
50	Germline variation at 8q24 and prostate cancer risk in men of European ancestry. Nature Communications, 2018, 9, 4616.	5.8	43
51	Activating Mutations in PAK1, Encoding p21-Activated Kinase 1, Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 579-591.	2.6	54
52	Large-scale transcriptome-wide association study identifies new prostate cancer risk regions. Nature Communications, 2018, 9, 4079.	5.8	121
53	Identification of multiple risk loci and regulatory mechanisms influencing susceptibility to multiple myeloma. Nature Communications, 2018, 9, 3707.	5.8	86
54	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	3.7	81

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55	Association analyses of more than 140,000 men identify 63 new prostate cancer susceptibility loci. Nature Genetics, 2018, 50, 928-936.	9.4	652
56	Integrated Molecular Characterization of Testicular Germ Cell Tumors. Cell Reports, 2018, 23, 3392-3406.	2.9	324
57	Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. Nature Communications, 2018, 9, 2256.	5.8	88
58	Inherited defects in checkpoint kinase 2 (CHEK2) to confer increased susceptibility to testicular germ cell tumors.. Journal of Clinical Oncology, 2018, 36, 1515-1515.	0.8	1
59	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
60	Compound heterozygous GATA5 mutations in a girl with hydrops fetalis, congenital heart defects and genital anomalies. Human Genetics, 2017, 136, 339-346.	1.8	11
61	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	0.7	69
62	Meta-analysis of five genome-wide association studies identifies multiple new loci associated with testicular germ cell tumor. Nature Genetics, 2017, 49, 1141-1147.	9.4	105
63	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
64	Survival beyond the perinatal period expands the phenotypes caused by mutations in GLE1. American Journal of Medical Genetics, Part A, 2017, 173, 3098-3103.	0.7	10
65	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
66	Genome-wide association study of classical Hodgkin lymphoma identifies key regulators of disease susceptibility. Nature Communications, 2017, 8, 1892.	5.8	40
67	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. American Journal of Human Genetics, 2017, 101, 1013-1020.	2.6	53
68	First de novo ANK3 nonsense mutation in a boy with intellectual disability, speech impairment and autistic features. European Journal of Medical Genetics, 2017, 60, 494-498.	0.7	21
69	WRNMutation Update: Mutation Spectrum, Patient Registries, and Translational Prospects. Human Mutation, 2017, 38, 7-15.	1.1	79
70	Dysfunction of the MDM2/p53 axis is linked to premature aging. Journal of Clinical Investigation, 2017, 127, 3598-3608.	3.9	54
71	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	2.6	81
72	The analysis of heterotaxy patients reveals new loss-of-function variants of GRK5. Scientific Reports, 2016, 6, 33231.	1.6	4

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73	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. Nature Genetics, 2016, 48, 856-866.	9.4	520
74	Phenotype and genotype in patients with Larsen syndrome: clinical homogeneity and allelic heterogeneity in seven patients. BMC Medical Genetics, 2016, 17, 27.	2.1	18
75	POLD1Germline Mutations in Patients Initially Diagnosed with Werner Syndrome. Human Mutation, 2015, 36, 1070-1079.	1.1	56
76	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	2.6	71
77	Suppressor of cytokine signaling 1gene mutation status as a prognostic biomarker in classical Hodgkin lymphoma. Oncotarget, 2015, 6, 29097-29110.	0.8	26
78	Atypical Aicardiâ€Goutieres syndrome: Is the WRN locus a modifier?. American Journal of Medical Genetics, Part A, 2014, 164, 2510-2513.	0.7	14
79	Carpal Tunnel Syndrome Is Associated With High Fibrinogen and Fibrinogen Deposits. Neurosurgery, 2014, 75, 276-285.	0.6	3
80	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	9.4	165
81	Chromatin retention of DNA damage sensors DDB2 and XPC through loss of p97 segregase causes genotoxicity. Nature Communications, 2014, 5, 3695.	5.8	92
82	Ethnicâ€specific <scp>WRN</scp> mutations in <scp>S</scp>outh <scp>A</scp>sian <scp>W</scp>erner syndrome patients: potential founder effect in patients with <scp>I</scp>ndian or <scp>P</scp>akistani ancestry. Molecular Genetics & Genomic Medicine, 2013, 1, 7-14.	0.6	16
83	Replication of genetic susceptibility loci for testicular germ cell cancer in the Croatian population. Carcinogenesis, 2012, 33, 1548-1552.	1.3	15
84	Coronary artery disease in a Werner syndromeâ€like form of progeria characterized by low levels of progerin, a splice variant of lamin A. American Journal of Medical Genetics, Part A, 2011, 155, 3002-3006.	0.7	55
85	WRN mutations in Werner syndrome patients: genomic rearrangements, unusual intronic mutations and ethnic-specific alterations. Human Genetics, 2010, 128, 103-111.	1.8	87