Martin Jakob Larsen

List of Publications by Year
in descending order

46	1,347	448610 19	425179 34
papers	citations	h-index	g-index

48	48	48	3253
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders. Scientific Reports, 2022, 12, 902.	1.6	9
2	Detection of DZIP1L mutations by whole-exome sequencing in consanguineous families with polycystic kidney disease. Pediatric Nephrology, 2022, 37, 2657-2665.	0.9	5
3	Total number of reads affects the accuracy of fetal fraction estimates in NIPT. Molecular Genetics & Genomic Medicine, 2021, 9, e1653.	0.6	8
4	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
5	Global Gene Expression Profiling and Transcription Factor Network Analysis of Cognitive Aging in Monozygotic Twins. Frontiers in Genetics, 2021, 12, 675587.	1.1	6
6	OTEH-4. Deeper insight into intratumoral heterogeneity by MRI and PET-guided stereotactic biopsies from glioblastoma patients. Neuro-Oncology Advances, 2021, 3, ii11-ii11.	0.4	0
7	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	1.0	6
8	Differential lncRNA expression profiling of cognitive function in middle and old aged monozygotic twins using generalized association analysis. Journal of Psychiatric Research, 2021, 140, 197-204.	1.5	3
9	Comparison of the Metastasis Predictive Potential of mRNA and Long Non-Coding RNA Profiling in Systemically Untreated Breast Cancer. Cancers, 2021, 13, 4907.	1.7	0
10	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. Brain, 2020, 143, 94-111.	3.7	18
11	Chromosomal translocation disrupting the SMAD4 gene resulting in the combined phenotype of Juvenile polyposis syndrome and Hereditary Hemorrhagic Telangiectasia. Molecular Genetics & Genomic Medicine, 2020, 8, e1498.	0.6	6
12	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
13	Biallelic variants in <scp>GLE1</scp> with survival beyond neonatal period. Clinical Genetics, 2020, 98, 622-625.	1.0	1
14	Differential long noncoding RNA profiling of BMI in twins. Epigenomics, 2020, 12, 1531-1541.	1.0	4
15	Exome sequencing revealed DNA variants in NCOR1, IGF2BP1, SGLT2 and NEK11 as potential novel causes of ketotic hypoglycemia in children. Scientific Reports, 2020, 10, 2114.	1.6	6
16	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	2.6	18
17	A Genome-Wide Integrative Association Study of DNA Methylation and Gene Expression Data and Later Life Cognitive Functioning in Monozygotic Twins. Frontiers in Neuroscience, 2020, 14, 233.	1.4	5
18	Is MED13L-related intellectual disability a recognizable syndrome?. European Journal of Medical Genetics, 2019, 62, 129-136.	0.7	21

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19	Global expression profiling of cognitive level and decline in middle-aged monozygotic twins. Neurobiology of Aging, 2019, 84, 141-147.	1.5	10
20	The Optimal Sequencing Depth of Tumor Biopsies for Identifying Clonal Cell Populations. Journal of Molecular Diagnostics, 2019, 21, 790-795.	1.2	2
21	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408.	1.0	10
22	Subtypes in BRCA-mutated breast cancer. Human Pathology, 2019, 84, 192-201.	1.1	22
23	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	1.4	32
24	Association of miR-548c-5p, miR-7-5p, miR-210-3p, miR-128-3p with recurrence in systemically untreated breast cancer. Oncotarget, 2018, 9, 9030-9042.	0.8	22
25	Compound heterozygous mutations in two different domains of ALDH18A1 do not affect the amino acid levels in a patient with hereditary spastic paraplegia. Neurogenetics, 2018, 19, 145-149.	0.7	11
26	Identification of metastasis driver genes by massive parallel sequencing of successive steps of breast cancer progression. PLoS ONE, 2018, 13, e0189887.	1.1	24
27	Genomic Analyses of Breast Cancer Progression Reveal Distinct Routes of Metastasis Emergence. Scientific Reports, 2017, 7, 43813.	1.6	24
28	Investigating a case of possible field cancerization in oral squamous cell carcinoma by the use of next-generation sequencing. Oral Oncology, 2017, 68, 74-80.	0.8	15
29	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
30	The subclonal structure and genomic evolution of oral squamous cell carcinoma revealed by ultra-deep sequencing. Oncotarget, 2017, 8, 16571-16580.	0.8	25
31	Bone structure in two adult subjects with impaired minor spliceosome function resulting from RNU4ATAC mutations causing microcephalic osteodysplastic primordial dwarfism type 1 (MOPD1). Bone, 2016, 92, 145-149.	1.4	8
32	Molecular Concordance Between Primary Breast Cancer and Matched Metastases. Breast Journal, 2016, 22, 420-430.	0.4	44
33	DamX Controls Reversible Cell Morphology Switching in Uropathogenic Escherichia coli. MBio, 2016, 7, .	1.8	55
34	Neonatal High Bone Mass With First Mutation of the NF-ÎºB Complex: Heterozygous De Novo Missense (p.Asp512Ser) RELA (Rela/p65). Journal of Bone and Mineral Research, 2016, 31, 163-172.	3.1	21
35	Evaluation of Nine Somatic Variant Callers for Detection of Somatic Mutations in Exome and Targeted Deep Sequencing Data. PLoS ONE, 2016, 11, e0151664.	1.1	144
36	Long non-coding RNA expression profiles predict metastasis in lymph node-negative breast cancer independently of traditional prognostic markers. Breast Cancer Research, 2015, 17, 55.	2.2	49

#	Article	IF	Citations
37	Global gene expression profiling of telangiectasial tissue from patients with hereditary hemorrhagic telangiectasia. Microvascular Research, 2015, 99, 118-126.	1.1	13
38	Clonal expansion and linear genome evolution through breast cancer progression from pre-invasive stages to asynchronous metastasis. Oncotarget, 2015, 6, 5634-5649.	0.8	42
39	Long Non-Coding RNA Expression Profiles in Hereditary Haemorrhagic Telangiectasia. PLoS ONE, 2014, 9, e90272.	1.1	21
40	Immunodeficiency Associated with a Nonsense Mutation of IKBKB. Journal of Clinical Immunology, 2014, 34, 916-921.	2.0	44
41	Microarray-Based RNA Profiling of Breast Cancer: Batch Effect Removal Improves Cross-Platform Consistency. BioMed Research International, 2014, 2014, 1-11.	0.9	21
42	RNA profiling reveals familial aggregation of molecular subtypes in non-BRCA1/2 breast cancer families. BMC Medical Genomics, 2014, 7, 9.	0.7	18
43	Use of next generation sequencing in head and neck squamous cell carcinomas: A review. Oral Oncology, 2014, 50, 1035-1040.	0.8	21
44	Hereditary Breast Cancer: Clinical, Pathological and Molecular Characteristics. Breast Cancer: Basic and Clinical Research, 2014, 8, BCBCR.S18715.	0.6	71
45	Long non-coding RNA HOTAIR is an independent prognostic marker of metastasis in estrogen receptor-positive primary breast cancer. Breast Cancer Research and Treatment, 2013, 142, 529-536.	1.1	242
46	Classifications within Molecular Subtypes Enables Identification of BRCA1/BRCA2 Mutation Carriers by RNA Tumor Profiling. PLoS ONE, 2013, 8, e64268.	1.1	89