Sebastian Lunke

List of Publications by Year
in descending order

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66 papers	2,716 citations	257450 24 h-index	197818 49 g-index
68 all docs	68 docs citations	68 times ranked	5611 citing authors

#	Article	IF	Citations
1	Tracking the origins and drivers of subclonal metastatic expansion in prostate cancer. Nature Communications, 2015, 6, 6605.	12.8	312
2	Genome-wide analysis distinguishes hyperglycemia regulated epigenetic signatures of primary vascular cells. Genome Research, 2011, 21, 1601-1615.	5.5	198
3	Circulating tumour cells from patients with colorectal cancer have cancer stem cell hallmarks inex vivoculture. Gut, 2017, 66, 1802-1810.	12.1	163
4	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. JAMA - Journal of the American Medical Association, 2020, 323, 2503.	7.4	160
5	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. Genetics in Medicine, 2018, 20, 1554-1563.	2.4	125
6	Does genomic sequencing early in the diagnostic trajectory make a difference? AÂfollow-up study of clinical outcomes and cost-effectiveness. Genetics in Medicine, 2019, 21, 173-180.	2.4	118
7	Survival motor neuron gene 2 silencing by DNA methylation correlates with spinal muscular atrophy disease severity and can be bypassed by histone deacetylase inhibition. Human Molecular Genetics, 2009, 18, 304-317.	2.9	116
8	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	7.6	105
9	Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. European Journal of Human Genetics, 2018, 26, 644-651.	2.8	102
10	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	1.6	93
11	Contraction-induced Interleukin-6 Gene Transcription in Skeletal Muscle Is Regulated by c-Jun Terminal Kinase/Activator Protein-1. Journal of Biological Chemistry, 2012, 287, 10771-10779.	3.4	87
12	Vascular histone deacetylation by pharmacological HDAC inhibition. Genome Research, 2014, 24, 1271-1284.	5.5	79
13	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
14	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. Genetics in Medicine, 2021, 23, 183-191.	2.4	70
15	Response to Cetuximab With or Without Irinotecan in Patients With Refractory Metastatic Colorectal Cancer Harboring the KRAS G13D Mutation: Australasian Gastro-Intestinal Trials Group ICECREAM Study. Journal of Clinical Oncology, 2016, 34, 2258-2264.	1.6	52
16	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
17	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1508.	1.2	44
18	Role of Histone Acetylation in the Stimulatory Effect of Valproic Acid on Vascular Endothelial Tissue-Type Plasminogen Activator Expression. PLoS ONE, 2012, 7, e31573.	2.5	41

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19	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. European Journal of Human Genetics, 2020, 28, 587-596.	2.8	38
20	A head-to-head evaluation of the diagnostic efficacy and costs of trio versus singleton exome sequencing analysis. European Journal of Human Genetics, 2019, 27, 1791-1799.	2.8	37
21	Exploring the utility of human DNA methylation arrays for profiling mouse genomic DNA. Genomics, 2013, 102, 38-46.	2.9	36
22	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
23	Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. European Journal of Human Genetics, 2019, 27, 1493-1501.	2.8	29
24	The emerging role of epigenetic modifications and chromatin remodeling in spinal muscular atrophy. Journal of Neurochemistry, 2009, 109, 1557-1569.	3.9	28
25	Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. Genetics in Medicine, 2020, 22, 1976-1985.	2.4	28
26	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. Nature Communications, 2021, 12, 2678.	12.8	26
27	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. Genetics in Medicine, 2020, 22, 1986-1993.	2.4	25
28	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	2.8	24
29	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	2.0	24
30	Exome sequencing in newborns with congenital deafness as a model for genomic newborn screening: the Baby Beyond Hearing project. Genetics in Medicine, 2020, 22, 937-944.	2.4	22
31	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. European Journal of Human Genetics, 2019, 27, 1821-1826.	2.8	19
32	IREB2-associated neurodegeneration. Brain, 2019, 142, e40-e40.	7.6	19
33	Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. Npj Genomic Medicine, 2021, 6, 5.	3.8	19
34	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1692-1709.	6.2	18
35	ERCC1mutations impede DNA damage repair and cause liver and kidney dysfunction in patients. Journal of Experimental Medicine, 2021, 218, .	8.5	18
36	Is faster better? An economic evaluation of rapid and ultra-rapid genomic testing in critically ill infants and children. Genetics in Medicine, 2022, 24, 1037-1044.	2.4	18

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37	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
38	Examining the impact of regular aspirin use and <scp>PIK3CA</scp> mutations on survival in stage 2 colon cancer. Internal Medicine Journal, 2017, 47, 88-98.	0.8	16
39	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. BMJ Paediatrics Open, 2017, 1, e000119.	1.4	16
40	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.6	16
41	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
42	The expandingLARS2phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. Human Mutation, 2020, 41, 1425-1434.	2.5	15
43	The feasibility of molecular testing on cell blocks created from brush tip washings in the assessment of peripheral lung lesions. Journal of Thoracic Disease, 2016, 8, 2551-2555.	1.4	14
44	A novel approach to offering additional genomic findingsâ€"A protocol to test a twoâ€step approach in the healthcare system. Journal of Genetic Counseling, 2019, 28, 388-397.	1.6	14
45	Clinical and laboratory reporting impact of ACMG-AMP and modified ClinGen variant classification frameworks in MYH7-related cardiomyopathy. Genetics in Medicine, 2021, 23, 1108-1115.	2.4	14
46	Epigenetic evidence of an Ac/Dc axis by VPA and SAHA. Clinical Epigenetics, 2021, 13, 58.	4.1	13
47	Applicability of Histone Deacetylase Inhibition for the Treatment of Spinal Muscular Atrophy. Neurotherapeutics, 2013, 10, 677-687.	4.4	12
48	High-Throughput Amplicon-Based Copy Number Detection of 11 Genes in Formalin-Fixed Paraffin-Embedded Ovarian Tumour Samples by MLPA-Seq. PLoS ONE, 2015, 10, e0143006.	2.5	11
49	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. Allergy, Asthma and Clinical Immunology, 2018, 14, 65.	2.0	10
50	Rapid Identification of Biallelic <i>SPTB</i> Mutation in a Neonate with Severe Congenital Hemolytic Anemia and Liver Failure. Molecular Syndromology, 2020, 11, 50-55.	0.8	9
51	Whole exome sequencing reveals a de novo missense variant in EEF1A2 in a Rett syndromeâ€like patient. Clinical Case Reports (discontinued), 2019, 7, 2476-2482.	0.5	8
52	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. Human Mutation, 2020, 41, 1884-1891.	2.5	8
53	Paediatric genomic testing: Navigating medicare rebatable genomic testing. Journal of Paediatrics and Child Health, 2021, 57, 477-483.	0.8	8
54	AmpliVar: Mutation Detection in High-Throughput Sequence from Amplicon-Based Libraries. Human Mutation, 2015, 36, 411-418.	2.5	7

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55	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. Obstetrical and Gynecological Survey, 2020, 75, 662-664.	0.4	7
56	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	9.0	7
57	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. International Journal of Molecular Sciences, 2022, 23, 986.	4.1	5
58	MSH2-deficient prostate tumours have a distinct immune response and clinical outcome compared to MSH2-deficient colorectal or endometrial cancer. Prostate Cancer and Prostatic Diseases, 2021, 24, 1167-1180.	3.9	4
59	Ethylmalonic encephalopathy masquerading as meningococcemia. Journal of Physical Education and Sports Management, 2022, , mcs.a006193.	1.2	3
60	Can Rapid Nanopore Sequencing Bring Genomic Testing to the Bedside?. Clinical Chemistry, 2022, 68, 1484-1485.	3.2	3
61	Intratumorous heterogeneity for RAS mutations in a treatment-naÃ¯ve colorectal tumour. Journal of Clinical Pathology, 2017, 70, 720-723.	2.0	2
62	Use of ultraâ€rapid wholeâ€exome sequencing to diagnose congenital central hypoventilation syndrome. Pediatric Pulmonology, 2020, 55, 855-857.	2.0	2
63	Distinct diagnostic trajectories in <scp>NBAS</scp>â€associated acute liver failure highlights the need for timely functional studies. JIMD Reports, 2022, 63, 240-249.	1.5	2
64	Rapid mitochondrial genome (MTDNA) sequencing: facilitating rapid diagnosis of mitochondrial diseases in paediatric acute care. Pathology, 2019, 51, S118-S119.	0.6	1
65	Rapid genomic testing in acute paediatric care: Is it worth the trouble?. Pathology, 2018, 50, S59.	0.6	0
66	Clinical Utility of Real-Time Targeted Molecular Profiling in the Clinical Management of Ovarian Cancer: The ALLOCATE Study. JCO Precision Oncology, 2019, 3, 1-18.	3.0	0