

# Sebastian Lunke

## List of Publications by Year in descending order

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Version: 2024-02-01

66  
papers

2,716  
citations

293460

24  
h-index

223390

49  
g-index

68  
all docs

68  
docs citations

68  
times ranked

6067  
citing authors

#	ARTICLE	IF	CITATIONS
1	Tracking the origins and drivers of subclonal metastatic expansion in prostate cancer. <i>Nature Communications</i> , 2015, 6, 6605.	5.8	312
2	Genome-wide analysis distinguishes hyperglycemia regulated epigenetic signatures of primary vascular cells. <i>Genome Research</i> , 2011, 21, 1601-1615.	2.4	198
3	Circulating tumour cells from patients with colorectal cancer have cancer stem cell hallmarks in <i>ex vivo</i> culture. <i>Gut</i> , 2017, 66, 1802-1810.	6.1	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. <i>JAMA - Journal of the American Medical Association</i> , 2020, 323, 2503.	3.8	160
5	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. <i>Genetics in Medicine</i> , 2018, 20, 1554-1563.	1.1	125
6	Does genomic sequencing early in the diagnostic trajectory make a difference? A follow-up study of clinical outcomes and cost-effectiveness. <i>Genetics in Medicine</i> , 2019, 21, 173-180.	1.1	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. <i>Human Molecular Genetics</i> , 2009, 18, 304-317.	1.4	116
8	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. <i>Brain</i> , 2017, 140, 1595-1610.	3.7	105
9	Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. <i>European Journal of Human Genetics</i> , 2018, 26, 644-651.	1.4	102
10	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. <i>Epilepsy Research</i> , 2017, 131, 1-8.	0.8	93
11	Contraction-induced Interleukin-6 Gene Transcription in Skeletal Muscle Is Regulated by c-Jun Terminal Kinase/Activator Protein-1. <i>Journal of Biological Chemistry</i> , 2012, 287, 10771-10779.	1.6	87
12	Vascular histone deacetylation by pharmacological HDAC inhibition. <i>Genome Research</i> , 2014, 24, 1271-1284.	2.4	79
13	Cpipe: a shared variant detection pipeline designed for diagnostic settings. <i>Genome Medicine</i> , 2015, 7, 68.	3.6	78
14	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. <i>Genetics in Medicine</i> , 2021, 23, 183-191.	1.1	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. <i>Journal of Clinical Oncology</i> , 2016, 34, 2258-2264.	0.8	52
16	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	1.1	45
17	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1508.	0.6	44
18	Role of Histone Acetylation in the Stimulatory Effect of Valproic Acid on Vascular Endothelial Tissue-Type Plasminogen Activator Expression. <i>PLoS ONE</i> , 2012, 7, e31573.	1.1	41

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19	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. <i>European Journal of Human Genetics</i> , 2020, 28, 587-596.	1.4	38
20	A head-to-head evaluation of the diagnostic efficacy and costs of trio versus singleton exome sequencing analysis. <i>European Journal of Human Genetics</i> , 2019, 27, 1791-1799.	1.4	37
21	Exploring the utility of human DNA methylation arrays for profiling mouse genomic DNA. <i>Genomics</i> , 2013, 102, 38-46.	1.3	36
22	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. <i>American Journal of Human Genetics</i> , 2021, 108, 1551-1557.	2.6	36
23	Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. <i>European Journal of Human Genetics</i> , 2019, 27, 1493-1501.	1.4	29
24	The emerging role of epigenetic modifications and chromatin remodeling in spinal muscular atrophy. <i>Journal of Neurochemistry</i> , 2009, 109, 1557-1569.	2.1	28
25	Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. <i>Genetics in Medicine</i> , 2020, 22, 1976-1985.	1.1	28
26	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. <i>Nature Communications</i> , 2021, 12, 2678.	5.8	26
27	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. <i>Genetics in Medicine</i> , 2020, 22, 1986-1993.	1.1	25
28	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. <i>European Journal of Human Genetics</i> , 2017, 25, 1268-1272.	1.4	24
29	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 2-9.	1.0	24
30	Exome sequencing in newborns with congenital deafness as a model for genomic newborn screening: the Baby Beyond Hearing project. <i>Genetics in Medicine</i> , 2020, 22, 937-944.	1.1	22
31	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1821-1826.	1.4	19
32	IREB2-associated neurodegeneration. <i>Brain</i> , 2019, 142, e40-e40.	3.7	19
33	Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. <i>Npj Genomic Medicine</i> , 2021, 6, 5.	1.7	19
34	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 1692-1709.	2.6	18
35	ERCC1 mutations impede DNA damage repair and cause liver and kidney dysfunction in patients. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	18
36	Is faster better? An economic evaluation of rapid and ultra-rapid genomic testing in critically ill infants and children. <i>Genetics in Medicine</i> , 2022, 24, 1037-1044.	1.1	18

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37	Missense variants in <i>TAF1</i> and developmental phenotypes: Challenges of determining pathogenicity. <i>Human Mutation</i> , 2020, 41, 449-464.	1.1	17
38	Examining the impact of regular aspirin use and <i>PIK3CA</i> mutations on survival in stage 2 colon cancer. <i>Internal Medicine Journal</i> , 2017, 47, 88-98.	0.5	16
39	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. <i>BMJ Paediatrics Open</i> , 2017, 1, e000119.	0.6	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. <i>Journal of the Neurological Sciences</i> , 2021, 420, 117260.	0.3	16
41	Biallelic loss of function variants in <i>PPP1R21</i> cause a neurodevelopmental syndrome with impaired endocytic function. <i>Human Mutation</i> , 2019, 40, 267-280.	1.1	15
42	The expanding <i>LARS2</i> phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. <i>Human Mutation</i> , 2020, 41, 1425-1434.	1.1	15
43	The feasibility of molecular testing on cell blocks created from brush tip washings in the assessment of peripheral lung lesions. <i>Journal of Thoracic Disease</i> , 2016, 8, 2551-2555.	0.6	14
44	A novel approach to offering additional genomic findings – A protocol to test a two-step approach in the healthcare system. <i>Journal of Genetic Counseling</i> , 2019, 28, 388-397.	0.9	14
45	Clinical and laboratory reporting impact of ACMG-AMP and modified ClinGen variant classification frameworks in MYH7-related cardiomyopathy. <i>Genetics in Medicine</i> , 2021, 23, 1108-1115.	1.1	14
46	Epigenetic evidence of an Ac/Dc axis by VPA and SAHA. <i>Clinical Epigenetics</i> , 2021, 13, 58.	1.8	13
47	Applicability of Histone Deacetylase Inhibition for the Treatment of Spinal Muscular Atrophy. <i>Neurotherapeutics</i> , 2013, 10, 677-687.	2.1	12
48	High-Throughput Amplicon-Based Copy Number Detection of 11 Genes in Formalin-Fixed Paraffin-Embedded Ovarian Tumour Samples by MLPA-Seq. <i>PLoS ONE</i> , 2015, 10, e0143006.	1.1	11
49	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. <i>Allergy, Asthma and Clinical Immunology</i> , 2018, 14, 65.	0.9	10
50	Rapid Identification of Biallelic <i>SPTB</i> Mutation in a Neonate with Severe Congenital Hemolytic Anemia and Liver Failure. <i>Molecular Syndromology</i> , 2020, 11, 50-55.	0.3	9
51	Whole exome sequencing reveals a de novo missense variant in <i>EEF1A2</i> in a Rett syndrome-like patient. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 2476-2482.	0.2	8
52	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous <i>ASNS</i> splicing variant in a critically ill neonate. <i>Human Mutation</i> , 2020, 41, 1884-1891.	1.1	8
53	Paediatric genomic testing: Navigating medicare rebatable genomic testing. <i>Journal of Paediatrics and Child Health</i> , 2021, 57, 477-483.	0.4	8
54	AmpliVar: Mutation Detection in High-Throughput Sequence from Amplicon-Based Libraries. <i>Human Mutation</i> , 2015, 36, 411-418.	1.1	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. <i>Obstetrical and Gynecological Survey</i> , 2020, 75, 662-664.	0.2	7
56	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. <i>JAMA Neurology</i> , 2022, 79, 405.	4.5	7
57	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. <i>International Journal of Molecular Sciences</i> , 2022, 23, 986.	1.8	5
58	MSH2-deficient prostate tumours have a distinct immune response and clinical outcome compared to MSH2-deficient colorectal or endometrial cancer. <i>Prostate Cancer and Prostatic Diseases</i> , 2021, 24, 1167-1180.	2.0	4
59	Ethylmalonic encephalopathy masquerading as meningococemia. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006193.	0.5	3
60	Can Rapid Nanopore Sequencing Bring Genomic Testing to the Bedside?. <i>Clinical Chemistry</i> , 2022, 68, 1484-1485.	1.5	3
61	Intratumorous heterogeneity for RAS mutations in a treatment-naïve colorectal tumour. <i>Journal of Clinical Pathology</i> , 2017, 70, 720-723.	1.0	2
62	Use of ultra-rapid whole-exome sequencing to diagnose congenital central hypoventilation syndrome. <i>Pediatric Pulmonology</i> , 2020, 55, 855-857.	1.0	2
63	Distinct diagnostic trajectories in <sc>NBAS</sc>-associated acute liver failure highlights the need for timely functional studies. <i>JIMD Reports</i> , 2022, 63, 240-249.	0.7	2
64	Rapid mitochondrial genome (MTDNA) sequencing: facilitating rapid diagnosis of mitochondrial diseases in paediatric acute care. <i>Pathology</i> , 2019, 51, S118-S119.	0.3	1
65	Rapid genomic testing in acute paediatric care: Is it worth the trouble?. <i>Pathology</i> , 2018, 50, S59.	0.3	0
66	Clinical Utility of Real-Time Targeted Molecular Profiling in the Clinical Management of Ovarian Cancer: The ALLOCATE Study. <i>JCO Precision Oncology</i> , 2019, 3, 1-18.	1.5	0