Sebastian Lunke

List of Publications by Year
in descending order

66	2,716	293460	223390
papers	citations	24 h-index	49 g-index

68	68	68	6067
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
2	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. International Journal of Molecular Sciences, 2022, 23, 986.	1.8	5
3	Ethylmalonic encephalopathy masquerading as meningococcemia. Journal of Physical Education and Sports Management, 2022, , mcs.a006193.	0.5	3
4	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.	1.1	18
5	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	4.5	7
6	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.	0.7	2
7	Can Rapid Nanopore Sequencing Bring Genomic Testing to the Bedside?. Clinical Chemistry, 2022, 68, 1484-1485.	1.5	3
8	Clinical impact of genomic testing in patients with suspected monogenic kidney disease. Genetics in Medicine, 2021, 23, 183-191.	1.1	70
9	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.	0.3	16
10	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.	1.1	14
11	Paediatric genomic testing: Navigating medicare rebatable genomic testing. Journal of Paediatrics and Child Health, 2021, 57, 477-483.	0.4	8
12	Epigenetic evidence of an Ac/Dc axis by VPA and SAHA. Clinical Epigenetics, 2021, 13, 58.	1.8	13
13	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. Nature Communications, 2021, 12, 2678.	5.8	26
14	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.	2.0	4
15	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.	2.6	36
16	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1692-1709.	2.6	18
17	Learning from scaling up ultra-rapid genomic testing for critically ill children to a national level. Npj Genomic Medicine, 2021, 6, 5.	1.7	19
18	ERCC1mutations impede DNA damage repair and cause liver and kidney dysfunction in patients. Journal of Experimental Medicine, 2021, 218, .	4.2	18

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19	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
20	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. European Journal of Human Genetics, 2020, 28, 587-596.	1.4	38
21	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.	1.1	8
22	A cost-effectiveness analysis of genomic sequencing in a prospective versus historical cohort of complex pediatric patients. Genetics in Medicine, 2020, 22, 1986-1993.	1.1	25
23	Parental experiences of ultrarapid genomic testing for their critically unwell infants and children. Genetics in Medicine, 2020, 22, 1976-1985.	1.1	28
24	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1508.	0.6	44
25	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.	3.8	160
26	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.	0.3	9
27	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.	1.1	22
28	Use of ultraâ€rapid wholeâ€exome sequencing to diagnose congenital central hypoventilation syndrome. Pediatric Pulmonology, 2020, 55, 855-857.	1.0	2
29	The expandingLARS2phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. Human Mutation, 2020, 41, 1425-1434.	1.1	15
30	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.	0.2	7
31	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	1.1	15
32	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.	1.1	118
33	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.	1.4	37
34	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.	1.4	19
35	IREB2-associated neurodegeneration. Brain, 2019, 142, e40-e40.	3.7	19
36	Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. European Journal of Human Genetics, 2019, 27, 1493-1501.	1.4	29

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37	Rapid mitochondrial genome (MTDNA) sequencing: facilitating rapid diagnosis of mitochondrial diseases in paediatric acute care. Pathology, 2019, 51, S118-S119.	0.3	1
38	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.	0.9	14
39	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.	0.2	8
40	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.	1.5	0
41	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.	1.4	102
42	Meeting the challenges of implementing rapid genomic testing in acute pediatric care. Genetics in Medicine, 2018, 20, 1554-1563.	1.1	125
43	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. Seminars in Pediatric Neurology, 2018, 26, 2-9.	1.0	24
44	Rapid genomic testing in acute paediatric care: Is it worth the trouble?. Pathology, 2018, 50, S59.	0.3	0
45	Pitfalls of immunotherapy: lessons from a patient with CTLA-4 haploinsufficiency. Allergy, Asthma and Clinical Immunology, 2018, 14, 65.	0.9	10
46	Circulating tumour cells from patients with colorectal cancer have cancer stem cell hallmarks inex vivoculture. Gut, 2017, 66, 1802-1810.	6.1	163
47	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	0.8	93
48	Intratumorous heterogeneity for RAS mutations in a treatment-naÃ¯ve colorectal tumour. Journal of Clinical Pathology, 2017, 70, 720-723.	1.0	2
49	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.	1.4	24
50	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.	0.5	16
51	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. BMJ Paediatrics Open, 2017, 1, e000119.	0.6	16
52	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	3.7	105
53	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.	0.6	14
54	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.	0.8	52

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55	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	3.6	78
56	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.	1.1	11
57	Tracking the origins and drivers of subclonal metastatic expansion in prostate cancer. Nature Communications, 2015, 6, 6605.	5.8	312
58	AmpliVar: Mutation Detection in High-Throughput Sequence from Amplicon-Based Libraries. Human Mutation, 2015, 36, 411-418.	1.1	7
59	Vascular histone deacetylation by pharmacological HDAC inhibition. Genome Research, 2014, 24, 1271-1284.	2.4	79
60	Applicability of Histone Deacetylase Inhibition for the Treatment of Spinal Muscular Atrophy. Neurotherapeutics, 2013, 10, 677-687.	2.1	12
61	Exploring the utility of human DNA methylation arrays for profiling mouse genomic DNA. Genomics, 2013, 102, 38-46.	1.3	36
62	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.	1.6	87
63	Role of Histone Acetylation in the Stimulatory Effect of Valproic Acid on Vascular Endothelial Tissue-Type Plasminogen Activator Expression. PLoS ONE, 2012, 7, e31573.	1.1	41
64	Genome-wide analysis distinguishes hyperglycemia regulated epigenetic signatures of primary vascular cells. Genome Research, 2011, 21, 1601-1615.	2.4	198
65	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.	1.4	116
66	The emerging role of epigenetic modifications and chromatin remodeling in spinal muscular atrophy. Journal of Neurochemistry, 2009, 109, 1557-1569.	2.1	28