Stephen F Kingsmore

List of Publications by Year
in descending order

60	5,225	147726	123376
papers	citations	31 h-index	61 g-index

67	67	67	8113
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Retrospective identification of prenatal fetal anomalies associated with diagnostic neonatal genomic sequencing results. Prenatal Diagnosis, 2022, 42, 705-716.	1.1	2
2	2022: A pivotal year for diagnosis and treatment of rare genetic diseases. Journal of Physical Education and Sports Management, 2022, , mcs.a006204.	0.5	6
3	The Role of Genome Sequencing in Neonatal Intensive Care Units. Annual Review of Genomics and Human Genetics, 2022, 23, 427-448.	2.5	23
4	Rapid whole genome sequencing impacts care and resource utilization in infants with congenital heart disease. Npj Genomic Medicine, 2021, 6, 29.	1.7	27
5	Rapid Sequencing-Based Diagnosis of Thiamine Metabolism Dysfunction Syndrome. New England Journal of Medicine, 2021, 384, 2159-2161.	13.9	48
6	Project Baby Bear: Rapid precision care incorporating rWGS in 5 California childrenâ€™s hospitals demonstrates improved clinical outcomes and reduced costs of care. American Journal of Human Genetics, 2021, 108, 1231-1238.	2.6	140
7	Integrative genetic, genomic and transcriptomic analysis of heat shock protein and nuclear hormone receptor gene associations with spontaneous preterm birth. Scientific Reports, 2021, 11, 17115.	1.6	12
8	Rapid whole-genome sequencing in critically Ill children: shifting from unease to evidence, education, and equitable implementation. Journal of Pediatrics, 2021, 238, 343.	0.9	5
9	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	3.6	53
10	Cost Efficacy of Rapid Whole Genome Sequencing in the Pediatric Intensive Care Unit. Frontiers in Pediatrics, 2021, 9, 809536.	0.9	18
11	Measurement of genetic diseases as a cause of mortality in infants receiving whole genome sequencing. Npj Genomic Medicine, 2020, 5, 49.	1.7	29
12	Is Rapid Exome Sequencing Standard of Care in the Neonatal and Pediatric Intensive Care Units?. Journal of Pediatrics, 2020, 226, 14-15.	0.9	5
13	Partially automated whole-genome sequencing reanalysis of previously undiagnosed pediatric patients can efficiently yield new diagnoses. Npj Genomic Medicine, 2020, 5, 33.	1.7	36
14	Moving Genomics to Routine Care. Circulation Genomic and Precision Medicine, 2020, 13, 406-416.	1.6	11
15	An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. American Journal of Human Genetics, 2020, 107, 942-952.	2.6	110
16	A Prospective Study of Parental Perceptions of Rapid Whole-Genome and -Exome Sequencing among Seriously Ill Infants. American Journal of Human Genetics, 2020, 107, 953-962.	2.6	65
17	Mortality in a neonate with molybdenum cofactor deficiency illustrates the need for a comprehensive rapid precision medicine system. Journal of Physical Education and Sports Management, 2020, 6, a004705.	0.5	16
18	Clinical utility of ultra-rapid whole-genome sequencing in an infant with atypical presentation of WT1-associated nephrotic syndrome type 4. Journal of Physical Education and Sports Management, 2020, 6, a005470.	0.5	5

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19	A Randomized, Controlled Trial of the Analytic and Diagnostic Performance of Singleton and Trio, Rapid Genome and Exome Sequencing in Ill Infants. American Journal of Human Genetics, 2019, 105, 719-733.	2.6	238
20	Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation. Science Translational Medicine, 2019, 11, .	5.8	203
21	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	5.8	28
22	Rapid Whole Genome Sequencing Has Clinical Utility in Children in the PICU*. Pediatric Critical Care Medicine, 2019, 20, 1007-1020.	0.2	105
23	Rapid whole-genome sequencing identifies a novel AIRE variant associated with autoimmune polyendocrine syndrome type 1. Journal of Physical Education and Sports Management, 2018, 4, a002485.	0.5	11
24	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	6.0	174
25	Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. Npj Genomic Medicine, 2018, 3, 10.	1.7	314
26	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. Npj Genomic Medicine, 2018, 3, 6.	1.7	156
27	The case for early use of rapid whole-genome sequencing in management of critically ill infants: late diagnosis of Coffinâ€“Siris syndrome in an infant with left congenital diaphragmatic hernia, congenital heart disease, and recurrent infections. Journal of Physical Education and Sports Management, 2018, 4, a002469.	0.5	29
28	Evaluating the discriminating capacity of cell death (apoptotic) biomarkers in sepsis. Journal of Intensive Care, 2018, 6, 72.	1.3	12
29	Concomitant diagnosis of immune deficiency and Pseudomonas sepsis in a 19 month old with ecthyma gangrenosum by host whole-genome sequencing. Journal of Physical Education and Sports Management, 2018, 4, a003244.	0.5	17
30	Novel Factor XIII variant identified through whole-genome sequencing in a child with intracranial hemorrhage. Journal of Physical Education and Sports Management, 2018, 4, a003525.	0.5	10
31	Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. Npj Genomic Medicine, 2018, 3, 16.	1.7	420
32	Whole exome sequencing reveals HSPA1L as a genetic risk factor for spontaneous preterm birth. PLoS Genetics, 2018, 14, e1007394.	1.5	35
33	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	1.0	174
34	Rapid whole-genome sequencing identifies a novel homozygous NPC1 variant associated with Niemannâ€“Pick type C1 disease in a 7-week-old male with cholestasis. Journal of Physical Education and Sports Management, 2017, 3, a001966.	0.5	15
35	Rapid whole-genome sequencing identifies a novel GABRA1 variant associated with West syndrome. Journal of Physical Education and Sports Management, 2017, 3, a001776.	0.5	23
36	Whole genome sequencing of an African American family highlights toll like receptor 6 variants in Kawasaki disease susceptibility. PLoS ONE, 2017, 12, e0170977.	1.1	14

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37	Constellation: a tool for rapid, automated phenotype assignment of a highly polymorphic pharmacogene, CYP2D6, from whole-genome sequences. Npj Genomic Medicine, 2016, 1, 15007.	1.7	93
38	Host gene expression classifiers diagnose acute respiratory illness etiology. Science Translational Medicine, 2016, 8, 322ra11.	5.8	202
39	Whole-Exome Sequencing and Whole-Genome Sequencing in Critically Ill Neonates Suspected to Have Single-Gene Disorders. Cold Spring Harbor Perspectives in Medicine, 2016, 6, a023168.	2.9	83
40	Newborn testing and screening by whole-genome sequencing. Genetics in Medicine, 2016, 18, 214-216.	1.1	28
41	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	0.6	436
42	AlstrÃ¶m Syndrome: Mutation Spectrum ofALMS1. Human Mutation, 2015, 36, 660-668.	1.1	117
43	Renal systems biology of patients with systemic inflammatory response syndrome. Kidney International, 2015, 88, 804-814.	2.6	38
44	Rapid whole genome sequencing and precision neonatology. Seminars in Perinatology, 2015, 39, 623-631.	1.1	162
45	A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. Genome Medicine, 2015, 7, 100.	3.6	237
46	CLPB Variants Associated with Autosomal-Recessive Mitochondrial Disorder with Cataract, Neutropenia, Epilepsy, and Methylglutaconic Aciduria. American Journal of Human Genetics, 2015, 96, 258-265.	2.6	58
47	Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. Lancet Respiratory Medicine,the, 2015, 3, 377-387.	5.2	322
48	Human metabolic response to systemic inflammation: assessment of the concordance between experimental endotoxemia and clinical cases of sepsis/SIRS. Critical Care, 2015, 19, 71.	2.5	62
49	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	9.4	65
50	A novel epileptic encephalopathy mutation in KCNB1 disrupts Kv2.1 ion selectivity, expression, and localization. Journal of General Physiology, 2015, 146, 399-410.	0.9	79
51	A patient with polymerase E1 deficiency (POLE1): clinical features and overlap with DNA breakage/instability syndromes. BMC Medical Genetics, 2015, 16, 31.	2.1	26
52	Emergency medical genomes: a breakthrough application of precision medicine. Genome Medicine, 2015, 7, 82.	3.6	25
53	Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Science Translational Medicine, 2014, 6, 265ra168.	5.8	440
54	N-of-1 genomic medicine for the rare pediatric genetic diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1279-1290.	0.5	7

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55	Expansion of CCR4+ activated T cells is associated with memory B cell reduction in DOCK8-deficient patients. Clinical Immunology, 2014, 152, 164-170.	1.4	11
56	Use of Dried Blood Spots for High through-Put, Rapid Turnaround Mutational Analysis in Patients with Hemophilia. Blood, 2014, 124, 5034-5034.	0.6	0
57	Molecular diagnosis of infantile onset inflammatory bowel disease by exome sequencing. Genomics, 2013, 102, 442-447.	1.3	35
58	Next-generation community genetics for low- and middle-income countries. Genome Medicine, 2012, 4, 25.	3.6	51
59	An Unusual Presentation of Congenital Dyserythropoietic Anemia Type II (CDAII) Associated with Severe Anemia in a Patient with a Novel Mutation of the SEC23B Gene. Blood, 2012, 120, 990-990.	0.6	1
60	Adopting orphans: comprehensive genetic testing of Mendelian diseases of childhood by next-generation sequencing. Expert Review of Molecular Diagnostics, 2011, 11, 855-868.	1.5	45