Keith Vaux

List of Publications by Year
in descending order

21 papers	2,583 citations	430874 18 h-index	752698 20 g-index
22 all docs	22 docs citations	22 times ranked	5341 citing authors

#	Article	IF	Citations
1	Prevalence of Fetal Alcohol Spectrum Disorders in 4 US Communities. JAMA - Journal of the American Medical Association, 2018, 319, 474.	7.4	562
2	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	12.6	466
3	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	2.5	382
4	Inactivating mutations in MFSD2A, required for omega-3 fatty acid transport in brain, cause a lethal microcephaly syndrome. Nature Genetics, 2015, 47, 809-813.	21.4	180
5	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	12.6	174
6	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
7	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. Cell, 2013, 154, 505-517.	28.9	94
8	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	6.2	88
9	The genetic landscape of autism spectrum disorders. Developmental Medicine and Child Neurology, 2014, 56, 12-18.	2.1	83
10	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
11	Use of gabapentin in the treatment of childhood reflex sympathetic dystrophy. Pediatric Neurology, 2000, 22, 220-221.	2.1	62
12	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
13	The Safe and Effective Use of Propofol Sedation in Children Undergoing Diagnostic and Therapeutic Procedures: Experience in a Pediatric ICU and a Review of the Literature. Pediatric Emergency Care, 2003, 19, 385-392.	0.9	61
14	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
15	Cyclophosphamide, methotrexate, and cytarabine embropathy: Is apoptosis the common pathway?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2003, 67, 403-408.	1.6	44
16	Birth outcomes in women who have taken adalimumab in pregnancy: A prospective cohort study. PLoS ONE, 2019, 14, e0223603.	2.5	40
17	The role of molecular testing and enzyme analysis in the management of hypomorphic citrullinemia. American Journal of Medical Genetics, Part A, 2008, 146A, 2885-2890.	1.2	24
18	Preventive Control of AIDS by the Dental Profession: A Survey of Practices in a Large Urban Area. Journal of Public Health Dentistry, 1990, 50, 7-12.	1.2	20

#	Article	IF	Citations
19	Neural responses to affective speech, including motherese, map onto clinical and social eye tracking profiles in toddlers with ASD. Nature Human Behaviour, 2022, 6, 443-454.	12.0	14
20	Placenta Accreta in a Separate Uterine Horn. Pediatric and Developmental Pathology, 2010, 13, 63-65.	1.0	4
21	Inside â€œOutsideâ€•Job: Unexpected Geometric Skin Ulcerations Overlying Orthopedic Hardware After Multimodal Laser Scar Revision. Dermatologic Surgery, 2018, 44, 1231-1233.	0.8	0