Donald G Basel

List of Publications by Year
in descending order

36 papers	679 citations	567281 15 h-index	580821 25 g-index
37 all docs	37 docs citations	37 times ranked	1363 citing authors

#	Article	IF	Citations
1	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	6.2	92
2	Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial. Lancet, The, 2020, 396, 684-692.	13.7	92
3	Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. Prenatal Diagnosis, 2015, 35, 1073-1078.	2.3	88
4	Analyzing the Genetic Spectrum of Vascular Anomalies with Overgrowth viaÂCancer Genomics. Journal of Investigative Dermatology, 2018, 138, 957-967.	0.7	45
5	Survival Benefit for Individuals With Constitutional Mismatch Repair Deficiency Undergoing Surveillance. Journal of Clinical Oncology, 2021, 39, 2779-2790.	1.6	40
6	Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study. Genetics in Medicine, 2021, 23, 2443-2447.	2.4	36
7	Ending a Diagnostic Odyssey. Pediatric Clinics of North America, 2017, 64, 265-272.	1.8	32
8	Genetic Testing Protocol Reduces Costs and Increases Rate of Genetic Diagnosis in Infants with Congenital Heart Disease. Pediatric Cardiology, 2017, 38, 1465-1470.	1.3	27
9	How Doctors Think. Pediatric Clinics of North America, 2017, 64, 1-15.	1.8	20
10	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
11	Bacteremia in Patients with Heterotaxy: A Review and Implications for Management. Congenital Heart Disease, 2016, 11, 537-547.	0.2	18
12	CafÃ© au lait spots: When and how to pursue their genetic origins. Clinics in Dermatology, 2020, 38, 421-431.	1.6	18
13	Mitochondrial DNA Depletion Syndromes. Clinics in Perinatology, 2020, 47, 123-141.	2.1	17
14	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	3.8	17
15	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	2.5	15
16	Adaptive Behavior in Young Children with Neurofibromatosis Type 1. International Journal of Pediatrics (United Kingdom), 2013, 2013, 1-7.	0.8	14
17	Benign Joint Hypermobility Minimally Impacts Autonomic Abnormalities in Pediatric Subjects with Chronic Functional Pain Disorders. Journal of Pediatrics, 2016, 177, 49-52.	1.8	13
18	Molecular mechanics and dynamic simulations of well-known Kabuki syndrome-associated KDM6A variants reveal putative mechanisms of dysfunction. Orphanet Journal of Rare Diseases, 2021, 16, 66.	2.7	11

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19	Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder. Human Genetics, 2019, 138, 1259-1266.	3.8	10
20	Photodynamic Therapy for Benign Cutaneous Neurofibromas Using Aminolevulinic Acid Topical Application and 633â€‰nm Red Light Illumination. Photobiomodulation, Photomedicine, and Laser Surgery, 2021, 39, 411-417.	1.4	9
21	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. European Journal of Medical Genetics, 2020, 63, 103817.	1.3	6
22	Liver failure and xâ€linked immunodeficiency type 47. Pediatric Transplantation, 2020, 24, e13808.	1.0	6
23	De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445.	1.1	5
24	Rapid Exome and Genome Sequencing in the Intensive Care Unit. Critical Care Clinics, 2022, 38, 173-184.	2.6	5
25	A Rare Combination of Functional Disomy Xp, Deletion Xq13.2-q28 Spanning the XIST Gene, and Duplication 3q25.33-q29 in a Female with der(X)t(X;3)(q13.2;q25.33). Journal of Pediatric Genetics, 2018, 07, 023-028.	0.7	4
26	Hemophagocytic lymphohistiocytosis mimicking neonatal hemochromatosis. Pediatric Hematology and Oncology, 2019, 36, 451-456.	0.8	4
27	Inheritance of a Balanced t(12;20)(q24.33;p12.2) and Unbalanced der(13)t(7;13)(p21.3;q33.2) from a Maternally Derived Double Balanced Translocation Carrier. Journal of Pediatric Genetics, 2018, 07, 035-039.	0.7	3
28	Undiagnosed and Rare Diseases in Perinatal Medicine. Clinics in Perinatology, 2020, 47, 1-14.	2.1	3
29	Severe Neonatal RYR1 Myopathy With Pathological Features of Congenital Muscular Dystrophy. Journal of Neuropathology and Experimental Neurology, 2019, 78, 283-287.	1.7	3
30	Adaptive Behavior and Executive Functioning in Children with Neurofibromatosis Type 1 Using a Mixed Design. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 637-643.	1.1	3
31	Dysmorphology in a Genomic Era. Clinics in Perinatology, 2020, 47, 15-23.	2.1	2
32	Persistent and Stable Growth Promoting Effects of Vosoritide in Children With Achondroplasia for up to 2 Years: Results From the Ongoing Phase 3 Extension Study. Journal of the Endocrine Society, 2021, 5, A670-A671.	0.2	2
33	Clinical Implications of Mosaicism and Low-Level Mosaicism in Neurocutaneous Disorders. Current Genetic Medicine Reports, 2020, 8, 132-139.	1.9	0
34	SAT-LB18 A Randomized Controlled Trial of Vosoritide in Children With Achondroplasia. Journal of the Endocrine Society, 2020, 4, .	0.2	0
35	RARE-17. SURVIVAL BENEFIT FOR INDIVIDUALS WITH CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY SYNDROME AND BRAIN TUMORS WHO UNDERGO SURVEILLANCE PROTOCOL. A REPORT FROM THE INTERNATIONAL REPLICATION REPAIR CONSORTIUM. Neuro-Oncology, 2020, 22, iii445-iii446.	1.2	0
36	Mosaic Trisomy 9p in a Patient with Mild Dysmorphic Features and Normal Intelligence. Journal of the Association of Genetic Technologists, 2017, 43, 56-58.	0.1	0