Daniela del Gaudio

List of Publications by Year
in descending order

29 papers	1,381 citations	516710 16 h-index	501196 28 g-index
31 all docs	31 docs citations	31 times ranked	2959 citing authors

#	Article	IF	Citations
1	Novel KDM6A Kabuki Syndrome Mutation With Hyperinsulinemic Hypoglycemia and Pulmonary Hypertension Requiring ECMO. Journal of the Endocrine Society, 2022, 6, bvac015.	0.2	7
2	Feasibility and limitations of cultured skin fibroblasts for germline genetic testing in hematologic disorders. Human Mutation, 2022, 43, 950-962.	2.5	15
3	Exome sequencing identifies PDâ€Ł2 as a potential predisposition gene for lymphoma. Hematological Oncology, 2022, 40, 475-478.	1.7	1
4	Response to Mounts and Besser. Genetics in Medicine, 2021, 23, 240-242.	2.4	1
5	Germline variants drive myelodysplastic syndrome in young adults. Leukemia, 2021, 35, 2439-2444.	7.2	43
6	Novel compound heterozygous LRBA deletions in a 6-month-old with neonatal diabetes. Diabetes Research and Clinical Practice, 2021, 175, 108798.	2.8	3
7	Further delineation of a recognizable type of syndromic short stature caused by biallelic SEMA3A lossâ€ofâ€function variants. American Journal of Medical Genetics, Part A, 2021, 185, 889-893.	1.2	3
8	Adapting ACMG/AMP sequence variant classification guidelines for single-gene copy number variants. Genetics in Medicine, 2020, 22, 336-344.	2.4	82
9	Telomere biology disorder prevalence and phenotypes in adults with familial hematologic and/or pulmonary presentations. Blood Advances, 2020, 4, 4873-4886.	5.2	23
10	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. Brain, 2020, 143, 3564-3573.	7.6	23
11	CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1288-1295.	2.4	39
12	Update of variants identified in the pancreatic Î²â€cell K _{ATP} channel genes KCNJ11 and ABCC8 in individuals with congenital hyperinsulinism and diabetes. Human Mutation, 2020, 41, 884-905.	2.5	90
13	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	6.2	8
14	Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1133-1141.	2.4	89
15	Assessing the Feasibility and Limitations of Cultured Skin Fibroblasts for Germline Genetic Testing in Hematologic Disorders. Blood, 2020, 136, 35-36.	1.4	2
16	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 10 affected individuals. Genetics in Medicine, 2019, 21, 233-242.	2.4	39
17	GCK-MODY in the US Monogenic Diabetes Registry: Description of 27 unpublished variants. Diabetes Research and Clinical Practice, 2019, 151, 231-236.	2.8	14
18	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. Genetics in Medicine, 2019, 21, 195-206.	2.4	65

#	Article	IF	Citations
19	Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. Journal of Human Genetics, 2018, 63, 349-356.	2.3	10
20	Pancreatic Histopathology of Human Monogenic Diabetes Due to Causal Variants in KCNJ11, HNF1A, GATA6, and LMNA. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 35-45.	3.6	17
21	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	2.4	42
22	FOXP3 mutations causing earlyâ€onset insulinâ€requiring diabetes but without other features of immune dysregulation, polyendocrinopathy, enteropathy, Xâ€linked syndrome. Pediatric Diabetes, 2018, 19, 388-392.	2.9	25
23	Reprogramming human T cell function and specificity with non-viral genome targeting. Nature, 2018, 559, 405-409.	27.8	630
24	Alu -mediated deletion of PIGL in a Patient with CHIME syndrome. , 2017, 173, 1378-1382.		14
25	Improved molecular diagnosis of patients with neonatal diabetes using a combined next-generation sequencing and MS-MLPA approach. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 523-31.	0.9	16
26	A novel mutation in the promoter of RARS2 causes pontocerebellar hypoplasia in two siblings. Journal of Human Genetics, 2015, 60, 363-369.	2.3	26
27	Copy number analysis of NIPBL in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. Molecular Genetics & Genomic Medicine, 2014, 2, 115-123.	1.2	12
28	Clinical utility of next-generation sequencing for the molecular diagnosis of monogenic diabetes. Personalized Medicine, 2014, 11, 155-165.	1.5	0
29	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. Gene, 2014, 537, 279-284.	2.2	31