

# Daniela del Gaudio

## List of Publications by Year in descending order

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Version: 2024-02-01

29  
papers

1,381  
citations

516710

16  
h-index

501196

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g-index

31  
all docs

31  
docs citations

31  
times ranked

2959  
citing authors

#	ARTICLE	IF	CITATIONS
1	Reprogramming human T cell function and specificity with non-viral genome targeting. <i>Nature</i> , 2018, 559, 405-409.	27.8	630
2	Update of variants identified in the pancreatic $\beta$ -cell K <sup>ATP</sup> channel genes <i>KCNJ11</i> and <i>ABCC8</i> in individuals with congenital hyperinsulinism and diabetes. <i>Human Mutation</i> , 2020, 41, 884-905.	2.5	90
3	Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2020, 22, 1133-1141.	2.4	89
4	Adapting ACMG/AMP sequence variant classification guidelines for single-gene copy number variants. <i>Genetics in Medicine</i> , 2020, 22, 336-344.	2.4	82
5	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. <i>Genetics in Medicine</i> , 2019, 21, 195-206.	2.4	65
6	Germline variants drive myelodysplastic syndrome in young adults. <i>Leukemia</i> , 2021, 35, 2439-2444.	7.2	43
7	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. <i>Genetics in Medicine</i> , 2018, 20, 464-469.	2.4	42
8	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 10 affected individuals. <i>Genetics in Medicine</i> , 2019, 21, 233-242.	2.4	39
9	CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2020, 22, 1288-1295.	2.4	39
10	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. <i>Gene</i> , 2014, 537, 279-284.	2.2	31
11	A novel mutation in the promoter of RARS2 causes pontocerebellar hypoplasia in two siblings. <i>Journal of Human Genetics</i> , 2015, 60, 363-369.	2.3	26
12	<i>FOXP3</i> mutations causing early-onset insulin-requiring diabetes but without other features of immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. <i>Pediatric Diabetes</i> , 2018, 19, 388-392.	2.9	25
13	Telomere biology disorder prevalence and phenotypes in adults with familial hematologic and/or pulmonary presentations. <i>Blood Advances</i> , 2020, 4, 4873-4886.	5.2	23
14	Variants in the SK2 channel gene ( <i>KCNN2</i> ) lead to dominant neurodevelopmental movement disorders. <i>Brain</i> , 2020, 143, 3564-3573.	7.6	23
15	Pancreatic Histopathology of Human Monogenic Diabetes Due to Causal Variants in <i>KCNJ11</i> , <i>HNF1A</i> , <i>GATA6</i> , and <i>LMNA</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 35-45.	3.6	17
16	Improved molecular diagnosis of patients with neonatal diabetes using a combined next-generation sequencing and MS-MLPA approach. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 523-31.	0.9	16
17	Feasibility and limitations of cultured skin fibroblasts for germline genetic testing in hematologic disorders. <i>Human Mutation</i> , 2022, 43, 950-962.	2.5	15
18	Alu-mediated deletion of <i>PIGL</i> in a Patient with CHIME syndrome. , 2017, 173, 1378-1382.		14

#	ARTICLE	IF	CITATIONS
19	GCK-MODY in the US Monogenic Diabetes Registry: Description of 27 unpublished variants. <i>Diabetes Research and Clinical Practice</i> , 2019, 151, 231-236.	2.8	14
20	Copy number analysis of <i>NIPBL</i> in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 115-123.	1.2	12
21	Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. <i>Journal of Human Genetics</i> , 2018, 63, 349-356.	2.3	10
22	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 405-411.	6.2	8
23	Novel <i>KDM6A</i> Kabuki Syndrome Mutation With Hyperinsulinemic Hypoglycemia and Pulmonary Hypertension Requiring ECMO. <i>Journal of the Endocrine Society</i> , 2022, 6, bvac015.	0.2	7
24	Novel compound heterozygous LRBA deletions in a 6-month-old with neonatal diabetes. <i>Diabetes Research and Clinical Practice</i> , 2021, 175, 108798.	2.8	3
25	Further delineation of a recognizable type of syndromic short stature caused by biallelic <i>SEMA3A</i> loss-of-function variants. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 889-893.	1.2	3
26	Assessing the Feasibility and Limitations of Cultured Skin Fibroblasts for Germline Genetic Testing in Hematologic Disorders. <i>Blood</i> , 2020, 136, 35-36.	1.4	2
27	Response to Mounts and Besser. <i>Genetics in Medicine</i> , 2021, 23, 240-242.	2.4	1
28	Exome sequencing identifies <i>PDÉL2</i> as a potential predisposition gene for lymphoma. <i>Hematological Oncology</i> , 2022, 40, 475-478.	1.7	1
29	Clinical utility of next-generation sequencing for the molecular diagnosis of monogenic diabetes. <i>Personalized Medicine</i> , 2014, 11, 155-165.	1.5	0