Elisa De Franco

List of Publications by Year
in descending order

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Version: 2024-02-01

69 papers	2,890 citations	331538 21 h-index	182361 51 g-index
74 all docs	74 docs citations	74 times ranked	4331 citing authors

#	Article	IF	Citations
1	Importance of Immediate Thiamine Therapy in Children with Suspected Thiamine-Responsive Megaloblastic Anemiaâ€"Report on Two Patients Carrying a Novel SLC19A2 Gene Mutation. Journal of Pediatric Genetics, 2022, 11, 236-239.	0.3	3
2	Clinical and molecular characteristics of infantile-onset diabetes mellitus in Egypt. Annals of Pediatric Endocrinology and Metabolism, 2022, , .	0.8	3
3	SavvyCNV: Genome-wide CNV calling from off-targetÂreads. PLoS Computational Biology, 2022, 18, e1009940.	1.5	18
4	Genetic Etiology of Neonatal Diabetes Mellitus in Vietnamese Infants and Characteristics of Those With INS Gene Mutations. Frontiers in Endocrinology, 2022, 13, 866573.	1.5	1
5	Congenital beta cell defects are not associated with markers of islet autoimmunity, even in the context of high genetic risk for type 1 diabetes. Diabetologia, 2022, , 1.	2.9	1
6	A biallelic lossâ€ofâ€function PDIA6 variant in a second patient with polycystic kidney disease, infancyâ€onset diabetes, and microcephaly. Clinical Genetics, 2022, 102, 457-458.	1.0	0
7	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated ABCC8 Permanent Neonatal Diabetes. Diabetes Care, 2021, 44, 35-42.	4.3	24
8	Genotype and Phenotype Heterogeneity in Neonatal Diabetes: A Single Centre Experience in Turkey. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 80-87.	0.4	3
9	Pathological Î²-Cell Endoplasmic Reticulum Stress in Type 2 Diabetes: Current Evidence. Frontiers in Endocrinology, 2021, 12, 650158.	1.5	53
10	Case Report: Extended Clinical Spectrum of the Neonatal Diabetes With Congenital Hypothyroidism Syndrome. Frontiers in Endocrinology, 2021, 12, 665336.	1.5	8
11	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. Cell Reports, 2021, 35, 108981.	2.9	9
12	Case Report: Neonatal Diabetes Mellitus Caused by a Novel GLIS3 Mutation in Twins. Frontiers in Endocrinology, 2021, 12, 673755.	1.5	10
13	In celebration of a century with insulin â€“ Update of insulin gene mutations in diabetes. Molecular Metabolism, 2021, 52, 101280.	3.0	20
14	Identification of<scp>GCKâ€</scp>maturityâ€onset diabetes of the young in cases of neonatal hyperglycemia: A case series and review of clinical features. Pediatric Diabetes, 2021, 22, 876-881.	1.2	8
15	Monogenic diabetes in Pakistani infants and children: challenges in a resource poor country. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1095-1103.	0.4	4
16	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. American Journal of Human Genetics, 2021, 108, 1342-1349.	2.6	9
17	Genetic and clinical heterogeneity of permanent neonatal diabetes mellitus: a single tertiary centre experience. Acta Diabetologica, 2021, 58, 1689-1700.	1.2	8
18	Molecular Genetics, Clinical Characteristics, and Treatment Outcomes of KATP-Channel Neonatal Diabetes Mellitus in Vietnam National Childrenâ€™s Hospital. Frontiers in Endocrinology, 2021, 12, 727083.	1.5	4

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19	Neonatal diabetes caused by disrupted pancreatic and Î²â€cell development. Diabetic Medicine, 2021, 38, e14728.	1.2	6
20	Transient neonatal diabetes due to a disease causing novel variant in the ATP-binding cassette subfamily C member 8 (ABCC8) gene unmasks maturity-onset diabetes of the young (MODY) diabetes cases within a family. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 273-276.	0.4	1
21	From Biology to Genes and Back Again: Gene Discovery for Monogenic Forms of Beta-Cell Dysfunction in Diabetes. Journal of Molecular Biology, 2020, 432, 1535-1550.	2.0	19
22	Transient Neonatal Diabetes: An Etiologic Clue for the Adult Diabetologist. Canadian Journal of Diabetes, 2020, 44, 128-130.	0.4	5
23	A neuromuscular disorder with homozygosity for PIEZO2 gene variants: an important differential diagnosis for kyphoscoliotic Ehlers-Danlos Syndrome. Clinical Dysmorphology, 2020, 29, 69-72.	0.1	2
24	De Novo Mutations in EIF2B1 Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. Diabetes, 2020, 69, 477-483.	0.3	29
25	Type 1 diabetes can present before the age of 6Âmonths and is characterised by autoimmunity and rapid loss of beta cells. Diabetologia, 2020, 63, 2605-2615.	2.9	24
26	Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4351-e4359.	1.8	10
27	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.	1.1	90
28	<scp>HNRNPH1</scp>â€related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clinical Genetics, 2020, 98, 91-98.	1.0	25
29	Clinical Characteristics, Molecular Features, and Long-Term Follow-Up of 15 Patients with Neonatal Diabetes: A Single-Centre Experience. Hormone Research in Paediatrics, 2020, 93, 423-432.	0.8	2
30	A Non-Coding Disease Modifier of Pancreatic Agenesis Identified by Genetic Correction in a Patient-Derived iPSC Line. Cell Stem Cell, 2020, 27, 137-146.e6.	5.2	19
31	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, 2020, 130, 6338-6353.	3.9	58
32	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	1.4	9
33	Wolcott-Rallison syndrome in Iran: a common cause of neonatal diabetes. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 607-613.	0.4	11
34	Practical management in Wolcottâ€Rallison syndrome with associated hypothyroidism, neutropenia, and recurrent liver failure: A case report. Clinical Case Reports (discontinued), 2019, 7, 1133-1138.	0.2	6
35	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. American Journal of Human Genetics, 2019, 104, 985-989.	2.6	43
36	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. Diabetes, 2019, 68, 1528-1535.	0.3	22

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37	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	2.6	18
38	Identification of novel variants in neonatal diabetes mellitus genes in Egyptian patients with permanent NDM. International Journal of Diabetes in Developing Countries, 2019, 39, 53-59.	0.3	3
39	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. Genetics in Medicine, 2019, 21, 982-986.	1.1	22
40	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. Diabetologia, 2018, 61, 1027-1036.	2.9	26
41	A type 1 diabetes genetic risk score can discriminate monogenic autoimmunity with diabetes from early-onset clustering of polygenic autoimmunity with diabetes. Diabetologia, 2018, 61, 862-869.	2.9	33
42	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 195-204.	0.4	16
43	Emergence of insulin resistance following empirical glibenclamide therapy: a case report of neonatal diabetes with a recessive INS gene mutation. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 345-348.	0.4	8
44	Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation inNEUROD1. Pediatric Diabetes, 2018, 19, 898-904.	1.2	22
45	Monogenic Diabetes Not Caused By Mutations in Mody Genes: A Very Heterogenous Group of Diabetes. Experimental and Clinical Endocrinology and Diabetes, 2018, 126, 612-618.	0.6	12
46	Marked intrafamilial variability of exocrine and endocrine pancreatic phenotypes due to a splice site mutation in GATA6. Biotechnology and Biotechnological Equipment, 2018, 32, 124-129.	0.5	0
47	A Novel KCNJ11 Mutation Associated with Transient Neonatal Diabetes. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 175-178.	0.4	9
48	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. Lancet Diabetes and Endocrinology,the, 2018, 6, 637-646.	5.5	120
49	Neonatal Diabetes: Two Cases with Isolated Pancreas Agenesis due to Homozygous PTF1A Enhancer Mutations and One with Developmental Delay, Epilepsy, and Neonatal Diabetes Syndrome due to KCNJ11 Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 168-174.	0.4	19
50	Dominant ER Stressâ€“Inducing WFS1 Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. Diabetes, 2017, 66, 2044-2053.	0.3	77
51	Recessively InheritedLRBAMutations Cause Autoimmunity Presenting as Neonatal Diabetes. Diabetes, 2017, 66, 2316-2322.	0.3	59
52	Clinical and molecular characterization of children with neonatal diabetes mellitus at a tertiary care center in northern India. Indian Pediatrics, 2017, 54, 467-471.	0.2	13
53	Case report: maternal mosaicism resulting in inheritance of a novel GATA6 mutation causing pancreatic agenesis and neonatal diabetes mellitus. Diagnostic Pathology, 2017, 12, 1.	0.9	33
54	The Clinical Course of Patients with Preschool Manifestation of Type 1 Diabetes Is Independent of the HLA DR-DQ Genotype. Genes, 2017, 8, 146.	1.0	9

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55	Pancreatic Agenesis due to Compound Heterozygosity for a Novel Enhancer and Truncating Mutation in the PTF1A Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 274-277.	0.4	23
56	An ABCC8 Nonsense Mutation Causing Neonatal Diabetes Through Altered Transcript Expression. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 260-264.	0.4	13
57	An emerging, recognizable facial phenotype in association with mutations in GLIâ€similar 3 (GLIS3). American Journal of Medical Genetics, Part A, 2016, 170, 1918-1923.	0.7	16
58	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
59	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. Diabetes, 2016, 65, 2810-2815.	0.3	22
60	Single patient in GCK-MODY family successfully re-diagnosed into GCK-PNDM through targeted next-generation sequencing technology. Acta Diabetologica, 2016, 53, 337-338.	1.2	3
61	Genome, Exome, and Targeted Next-Generation Sequencing in Neonatal Diabetes. Pediatric Clinics of North America, 2015, 62, 1037-1053.	0.9	16
62	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet, The, 2015, 386, 957-963.	6.3	250
63	Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability. Human Molecular Genetics, 2014, 23, 6432-6440.	1.4	41
64	GATA4 Mutations Are a Cause of Neonatal and Childhood-Onset Diabetes. Diabetes, 2014, 63, 2888-2894.	0.3	108
65	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	9.4	255
66	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. Nature Genetics, 2014, 46, 812-814.	9.4	411
67	Analysis of Transcription Factors Key for Mouse Pancreatic Development Establishes NKX2-2 and MNX1 Mutations as Causes of Neonatal Diabetes in Man. Cell Metabolism, 2014, 19, 146-154.	7.2	123
68	GATA6 Mutations Cause a Broad Phenotypic Spectrum of Diabetes From Pancreatic Agenesis to Adult-Onset Diabetes Without Exocrine Insufficiency. Diabetes, 2013, 62, 993-997.	0.3	128
69	GATA6 haploinsufficiency causes pancreatic agenesis in humans. Nature Genetics, 2012, 44, 20-22.	9.4	249