Fabrizio Barbetti

List of Publications by Citations

105 papers	4,305 citations	35 h-index	64 g-index
110 ext. papers	4,878 ext. citations	6.7 avg, IF	4.58 L-index

#	Paper	IF	Citations
105	Neonatal diabetes mellitus due to complete glucokinase deficiency. *New England Journal of Medicine*, **2001**, 344, 1588-92	59.2	350
104	Mutations in the insulin receptor gene. *Endocrine Reviews*, **1992**, 13, 566-95	27.2	236
103	The genetic abnormality in the beta cell determines the response to an oral glucose load. *Diabetologia*, **2002**, 45, 427-35	10.3	197
102	Fecal lactate and ulcerative colitis. *Gastroenterology*, **1988**, 95, 1564-8	13.3	179
101	Lack of the architectural factor HMGA1 causes insulin resistance and diabetes in humans and mice. *Nature Medicine*, **2005**, 11, 765-73	50.5	172
100	Seven mutations in the human insulin gene linked to permanent neonatal/infancy-onset diabetes mellitus. *Journal of Clinical Investigation*, **2008**, 118, 2148-56	15.9	158
99	Antibodies from patients with rheumatoid arthritis target citrullinated histone 4 contained in neutrophils extracellular traps. *Annals of the Rheumatic Diseases*, **2014**, 73, 1414-22	2.4	153
98	The second activating glucokinase mutation (A456V): implications for glucose homeostasis and diabetes therapy. *Diabetes*, **2002**, 51, 1240-6	0.9	137
97	Permanent diabetes mellitus in the first year of life. *Diabetologia*, **2002**, 45, 798-804	10.3	130
96	KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes. *Human Mutation*, **2005**, 25, 22-7	4.7	118
95	Insulin gene mutations as cause of diabetes in children negative for five type 1 diabetes autoantibodies. *Diabetes Care*, **2009**, 32, 123-5	14.6	104
94	Minimal incidence of neonatal/infancy onset diabetes in Italy is 1:90,000 live births. *Acta Diabetologica*, **2012**, 49, 405-8	3.9	99
93	Role of transglutaminase 2 in glucose tolerance: knockout mice studies and a putative mutation in a MODY patient. *FASEB Journal*, **2002**, 16, 1371-8	0.9	99
92	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. *American Journal of Human Genetics*, **2015**, 97, 177-85	11	91
91	Mutant INS-gene induced diabetes of youth: proinsulin cysteine residues impose dominant-negative inhibition on wild-type proinsulin transport. *PLoS ONE*, **2010**, 5, e13333	3.7	85
90	The G53D mutation in Kir6.2 (KCNJ11) is associated with neonatal diabetes and motor dysfunction in adulthood that is improved with sulfonylurea therapy. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 1054-61	5.6	82
89	INS-gene mutations: from genetics and beta cell biology to clinical disease. *Molecular Aspects of Medicine*, **2015**, 42, 3-18	16.7	78

88	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	18.1	77
87	Maturity-onset diabetes of the young in children with incidental hyperglycemia: a multicenter Italian study of 172 families. *Diabetes Care*, **2009**, 32, 1864-6	14.6	77
86	Insights into the structure and regulation of glucokinase from a novel mutation (V62M), which causes maturity-onset diabetes of the young. *Journal of Biological Chemistry*, **2005**, 280, 14105-13	5.4	76
85	An ATP-binding mutation (G334D) in KCNJ11 is associated with a sulfonylurea-insensitive form of developmental delay, epilepsy, and neonatal diabetes. *Diabetes*, **2007**, 56, 328-36	0.9	73
84	Syndromes of Autoimmunity and Hypoglycemia: Autoantibodies Directed Against insulinand Its Receptor. *Endocrinology and Metabolism Clinics of North America*, **1989**, 18, 123-143	5.5	69
83	Further evidence that mutations in INS can be a rare cause of Maturity-Onset Diabetes of the Young (MODY). *BMC Medical Genetics*, **2010**, 11, 42	2.1	62
82	Monogenic Diabetes Accounts for 6.3% of Cases Referred to 15 Italian Pediatric Diabetes Centers During 2007 to 2012. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1826-1834	5.6	60
81	Mutations at the same residue (R50) of Kir6.2 (KCNJ11) that cause neonatal diabetes produce different functional effects. *Diabetes*, **2006**, 55, 1705-12	0.9	56
80	Sulfonylurea treatment outweighs insulin therapy in short-term metabolic control of patients with permanent neonatal diabetes mellitus due to activating mutations of the KCNJ11 (KIR6.2) gene. *Diabetologia*, **2006**, 49, 2210-3	10.3	52
79	Permanent diabetes during the first year of life: multiple gene screening in 54 patients. *Diabetologia*, **2011**, 54, 1693-701	10.3	51
78	Impaired cleavage of preproinsulin signal peptide linked to autosomal-dominant diabetes. *Diabetes* , **2012**, 61, 828-37	0.9	49
77	Genetic basis of endocrine disease. 1. Molecular genetics of insulin resistant diabetes mellitus. *Journal of Clinical Endocrinology and Metabolism*, **1991**, 73, 1158-63	5.6	47
76	Sexual dimorphism of body composition and insulin sensitivity across pubertal development in obese Caucasian subjects. *European Journal of Endocrinology*, **2009**, 160, 769-75	6.5	46
75	Renal cysts and diabetes syndrome linked to mutations of the hepatocyte nuclear factor-1 beta gene: description of a new family with associated liver involvement. *American Journal of Kidney Diseases*, **2002**, 40, 397-402	7.4	46
74	Cell Replacement after Gene Editing of a Neonatal Diabetes-Causing Mutation at the Insulin Locus. *Stem Cell Reports*, **2018**, 11, 1407-1415	8	42
73	Glucose tolerance status in 510 children and adolescents attending an obesity clinic in Central Italy. *Pediatric Diabetes*, **2010**, 11, 47-54	3.6	40
72	Transient neonatal diabetes mellitus is associated with a recurrent (R201H) KCNJ11 (KIR6.2) mutation. *Diabetologia*, **2005**, 48, 2439-41	10.3	39
71	School and pre-school children with type 1 diabetes during Covid-19 quarantine: The synergic effect of parental care and technology. *Diabetes Research and Clinical Practice*, **2020**, 166, 108302	7.4	35

70	Missense Mutations in the Human Insulin Promoter Factor-1 Gene and Their Relation to Maturity-Onset Diabetes of the Young and Late-Onset Type 2 Diabetes Mellitus in Caucasians. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 1323-1326	5.6	34
69	Missense mutations in the human insulin promoter factor-1 gene and their relation to maturity-onset diabetes of the young and late-onset type 2 diabetes mellitus in caucasians. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 1323-6	5.6	33
68	Two unrelated patients with familial hyperproinsulinemia due to a mutation substituting histidine for arginine at position 65 in the proinsulin molecule: identification of the mutation by direct sequencing of genomic deoxyribonucleic acid amplified by polymerase chain reaction. *Journal of Clinical Endocrinology and Metabolism*, **1990**, 71, 164-9	5.6	32
67	Role of the ENPP1 K121Q polymorphism in glucose homeostasis. *Diabetes*, **2008**, 57, 3360-4	0.9	31
66	A convenient microwave-assisted synthesis of N-glycosyl amino acids. *Tetrahedron Letters*, **2007**, 48, 2901-2904	1	31
65	N-Fmoc-Protected ω-Azido- and ω-Alkynyl-L-amino Acids as Building Blocks for the Synthesis of "Clickable" Peptides. *European Journal of Organic Chemistry*, **2008**, 2008, 5308-5314	3.2	29
64	Genetic causes and treatment of neonatal diabetes and early childhood diabetes. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2018**, 32, 575-591	6.5	29
63	Six cases with severe insulin resistance (SIR) associated with mutations of insulin receptor: Is a Bartter-like syndrome a feature of congenital SIR?. *Acta Diabetologica*, **2013**, 50, 951-7	3.9	28
62	Serological Proteome Analysis (SERPA) as a tool for the identification of new candidate autoantigens in type 1 diabetes. *Journal of Proteomics*, **2013**, 82, 263-73	3.9	28
61	No beta cell desensitisation after a median of 68 months on glibenclamide therapy in patients with KCNJ11-associated permanent neonatal diabetes. *Diabetologia*, **2011**, 54, 2736-8	10.3	27
60	Obese children with low birth weight demonstrate impaired beta-cell function during oral glucose tolerance test. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4448-52	5.6	27
59	Opposite clinical phenotypes of glucokinase disease: Description of a novel activating mutation and contiguous inactivating mutations in human glucokinase (GCK) gene. *Molecular Endocrinology*, **2009**, 23, 1983-9		27
58	Identification of candidate children for maturity-onset diabetes of the young type 2 (MODY2) gene testing: a seven-item clinical flowchart (7-iF). *PLoS ONE*, **2013**, 8, e79933	3.7	24
57	Increased OB gene expression leads to elevated plasma leptin concentrations in patients with chronic primary hyperinsulinemia. *Diabetes*, **1998**, 47, 1625-9	0.9	23
56	Successful treatment of young infants presenting neonatal diabetes mellitus with continuous subcutaneous insulin infusion before genetic diagnosis. *Acta Diabetologica*, **2016**, 53, 559-65	3.9	22
55	Insulin therapy in neonatal diabetes mellitus: a review of the literature. *Diabetes Research and Clinical Practice*, **2017**, 129, 126-135	7.4	20
54	IGF2 methylation is associated with lipid profile in obese children. *Hormone Research in Paediatrics*, **2013**, 79, 361-7	3.3	20
53	Biosensor analysis of anti-citrullinated protein/peptide antibody affinity. *Analytical Biochemistry*, **2014**, 465, 96-101	3.1	19

52	Low prevalence of HNF1A mutations after molecular screening of multiple MODY genes in 58 Italian families recruited in the pediatric or adult diabetes clinic from a single Italian hospital. *Diabetes Care*, **2014**, 37, e258-60	14.6	19
51	Mutations in the insulin receptor gene in patients with genetic syndromes of insulin resistance and acanthosis nigricans. *Journal of Investigative Dermatology*, **1992**, 98, 77S-81S	4.3	19
50	Survey on etiological diagnosis of diabetes in 1244 Italian diabetic children and adolescents: impact of access to genetic testing. *Diabetes Research and Clinical Practice*, **2015**, 107, e15-8	7.4	17
49	Glyburide ameliorates motor coordination and glucose homeostasis in a child with diabetes associated with the KCNJ11/S225T, del226-232 mutation. *Pediatric Diabetes*, **2012**, 13, 656-60	3.6	17
48	Three novel missense mutations in the glucokinase gene (G80S; E221K; G227C) in Italian subjects with maturity-onset diabetes of the young (MODY). *Human Mutation*, **1998**, 12, 136-136	4.7	17
47	Diabetes associated with dominant insulin gene mutations: outcome of 24-month, sensor-augmented insulin pump treatment. *Acta Diabetologica*, **2016**, 53, 499-501	3.9	15
46	Ketogenic diet in a patient with congenital hyperinsulinism: a novel approach to prevent brain damage. *Orphanet Journal of Rare Diseases*, **2015**, 10, 120	4.2	15
45	Congenital hyperinsulinism and glucose hypersensitivity in homozygous and heterozygous carriers of Kir6.2 (KCNJ11) mutation V290M mutation: K(ATP) channel inactivation mechanism and clinical management. *Diabetes*, **2011**, 60, 209-17	0.9	15
44	Designed glucopeptides mimetics of myelin protein epitopes as synthetic probes for the detection of autoantibodies, biomarkers of multiple sclerosis. *Journal of Medicinal Chemistry*, **2012**, 55, 10437-47	8.3	14
43	Thyroid function tests in obese prepubertal children: correlations with insulin sensitivity and body fat distribution. *Hormone Research in Paediatrics*, **2012**, 78, 100-5	3.3	14
42	TRIB3 R84 variant affects glucose homeostasis by altering the interplay between insulin sensitivity and secretion. *Diabetologia*, **2010**, 53, 1354-61	10.3	14
41	Mutational analysis of the coding regions of the genes encoding protein kinase B-alpha and -beta, phosphoinositide-dependent protein kinase-1, phosphatase targeting to glycogen, protein phosphatase inhibitor-1, and glycogenin: lessons from a search for genetic variability of the insulin-stimulated glycogen synthesis pathway of skeletal muscle in NIDDM patients. *Diabetes*	0.9	14
40	Focal congenital hyperinsulinism managed by medical treatment: a diagnostic algorithm based on molecular genetic screening. *Clinical Endocrinology*, **2014**, 81, 679-88	3.4	13
39	Can HbA1c combined with fasting plasma glucose help to assess priority for GCK-MODY vs HNF1A-MODY genetic testing?. *Acta Diabetologica*, **2018**, 55, 981-983	3.9	11
38	Diagnosis of neonatal and infancy-onset diabetes. *Endocrine Development*, **2007**, 11, 83-93		11
37	Case Report: When an Induced Illness Looks Like a Rare Disease. *Pediatrics*, **2015**, 136, e1361-5	7.4	10
36	Metabolic syndrome in italian obese children and adolescents: stronger association with central fat depot than with insulin sensitivity and birth weight. *International Journal of Hypertension*, **2011**, 2011, 257168	2.4	10
35	Functional characterization of a novel KCNJ11 in frame mutation-deletion associated with infancy-onset diabetes and a mild form of intermediate DEND: a battle between K(ATP) gain of channel activity and loss of channel expression. *PLoS ONE*, **2013**, 8, e63758	3.7	8

34	A possible role of transglutaminase 2 in the nucleus of INS-1E and of cells of human pancreatic islets. *Journal of Proteomics*, **2014**, 96, 314-27	3.9	7
33	Cardiovascular fitness, insulin resistance and metabolic syndrome in severely obese prepubertal Italian children. *Hormone Research in Paediatrics*, **2008**, 70, 349-56	3.3	7
32	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated Permanent Neonatal Diabetes. *Diabetes Care*, **2021**, 44, 35-42	14.6	7
31	Severe insulin resistance in disguise: A familial case of reactive hypoglycemia associated with a novel heterozygous INSR mutation. *Pediatric Diabetes*, **2018**, 19, 670-674	3.6	6
30	Growth hormone does not inhibit its own secretion during prolonged hypoglycemia in man. *Journal of Clinical Endocrinology and Metabolism*, **1990**, 70, 1371-4	5.6	6
29	Hyperglucagonemia in an animal model of insulin- deficient diabetes: what therapy can improve it?. *Clinical Diabetes and Endocrinology*, **2016**, 2, 11	4.7	5
28	The expression of four pyridoxal kinase (PDXK) human variants in Drosophila impacts on genome integrity. *Scientific Reports*, **2019**, 9, 14188	4.9	5
27	Evaluation of new immunological targets in neuromyelitis optica. *Journal of Peptide Science*, **2013**, 19, 25-32	2.1	5
26	Search for genetic variants in the p66Shc longevity gene by PCR-single strand conformational polymorphism in patients with early-onset cardiovascular disease. *BMC Genetics*, **2006**, 7, 14	2.6	5
25	Single-strand conformation polymorphism analysis of the glucose transporter gene GLUT1 in maturity-onset diabetes of the young. *Journal of Molecular Medicine*, **2001**, 79, 270-4	5.5	5
24	Role of proline 193 in the insulin receptor post-translational processing. *Diabetologia*, **1999**, 42, 435-42	10.3	5
23	Reduced replication fork speed promotes pancreatic endocrine differentiation and controls graft size. *JCI Insight*, **2021**, 6,	9.9	5
22	Prevalence of elevated 1-h plasma glucose and its associations in obese youth. *Diabetes Research and Clinical Practice*, **2016**, 116, 202-4	7.4	5
21	Neonatal Diabetes: Permanent Neonatal Diabetes and Transient Neonatal Diabetes. *Frontiers in Diabetes*, **2017**, 1-25	0.6	4
20	No sign of proliferative retinopathy in 15 patients with permanent neonatal diabetes with a median diabetes duration of 24 years. *Diabetes Care*, **2014**, 37, e181-2	14.6	4
19	Mutations in IAPP and NEUROG3 genes are not a common cause of permanent neonatal/infancy/childhood-onset diabetes. *Diabetic Medicine*, **2009**, 26, 660-1	3.5	3
18	Insulin: still a miracle after all these years. *Journal of Clinical Investigation*, **2019**, 129, 3045-3047	15.9	3
17	Congenital diabetes mellitus. *Minerva Pediatrica*, **2020**, 72, 240-249	1.6	3

List of Publications

16	GCK Mutation in a Child with Maturity Onset Diabetes of the Young, Type 2. *Iranian Journal of Pediatrics*, **2013**, 23, 226-8	1	3
15	MEHMO syndrome and the link between brain, pituitary and pancreas. *EBioMedicine*, **2019**, 42, 26-27	8.8	2
14	Macrosomia, transient neonatal hypoglycemia, and monogenic diabetes in a family with heterozygous mutation R154X of HNF4A gene. *Journal of Endocrinological Investigation*, **2011**, 34, 252-3	5.2	2
13	Sulfonylurea treatment in a girl with neonatal diabetes (KCNJ11 R201H) and celiac disease: impact of low compliance to the gluten free diet. *Diabetes Research and Clinical Practice*, **2009**, 84, 332-4	7.4	2
12	Differences between transient neonatal diabetes mellitus subtypes can guide diagnosis and therapy. *European Journal of Endocrinology*, **2021**, 184, 575-585	6.5	2
11	Type 2 diabetes in pediatrics. *Minerva Pediatrics*, **2021**,	1.5	1
10	Pre-diabetes in Italian obese children and youngsters. *Journal of Endocrinological Investigation*, **2011**, 34, e275-80	5.2	1
9	The application of precision medicine in monogenic diabetes.. *Expert Review of Endocrinology and Metabolism*, **2022**, 1-19	4.1	0
8	1453-P: Adaption of the ACMG/AMP Variant Interpretation Guidelines for GCK, HNF1A, HNF4A-MODY: Recommendations from the ClinGen Monogenic Diabetes Expert Panel. *Diabetes*, **2020**, 69, 1453-P	0.9	0
7	1636-P: Transient Neonatal Diabetes: Clinical Differences between Patients Bearing KATP Mutations and 6q24 Defects May Guide Genetic Screening. *Diabetes*, **2020**, 69, 1636-P	0.9	0
6	Contribution of ONECUT1 variants to different forms of non-autoimmune diabetes mellitus in Italian patients.. *Acta Diabetologica*, **2022**, 1	3.9	0
5	Intrafamilial Variability of Early-Onset Diabetes due to an INS Mutation. *Case Reports in Genetics*, **2011**, 2011, 258978	0.7	
4	Clinical and molecular evaluation of Italian patients affected by Pelizaeus-Merzbacher disease. *Journal of Inherited Metabolic Disease*, **1996**, 19, 197-200	5.4	
3	1264-P: Distinguishing between Obese Patients with Type 1 Diabetes (T1DMob) and Type 2 Diabetes in Adolescence (T2DMad) at Presentation. *Diabetes*, **2020**, 69, 1264-P	0.9	
2	Not Autoimmune Diabetes Mellitus in Paediatrics **2017**, 137-146		
1	Case report: coeliac disease as a cause of secondary failure of glibenclamide therapy in a patient with permanent neonatal diabetes due to KCNJ11/R201C mutation. *Diabetologia*, **2021**, 64, 1703-1706	10.3	