# Fabrizio Barbetti

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/9425583/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 108 papers | 5,317 citations | 101496 36 h-index | 85498 71 g-index |
| 110 all docs | 110 docs citations | 110 times ranked | 5536 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Neonatal Diabetes Mellitus Due to Complete Glucokinase Deficiency. New England Journal of Medicine, 2001, 344, 1588-1592. | 13.9 | 386 |
| 2 | Mutations in the Insulin Receptor Gene. Endocrine Reviews, 1992, 13, 566-595. | 8.9 | 287 |
| 3 | The genetic abnormality in the beta cell determines the response to an oral glucose load. Diabetologia, 2002, 45, 427-435. | 2.9 | 235 |
| 4 | Fecal Lactate and Ulcerative Colitis. Gastroenterology, 1988, 95, 1564-1568. | 0.6 | 225 |
| 5 | Antibodies from patients with rheumatoid arthritis target citrullinated histone 4 contained in neutrophils extracellular traps. Annals of the Rheumatic Diseases, 2014, 73, 1414-1422. | 0.5 | 209 |
| 6 | Lack of the architectural factor HMGA1 causes insulin resistance and diabetes in humans and mice. Nature Medicine, 2005, 11, 765-773. | 15.2 | 204 |
| 7 | Seven mutations in the human insulin gene linked to permanent neonatal/infancy-onset diabetes mellitus. Journal of Clinical Investigation, 2008, 118, 2148-56. | 3.9 | 189 |
| 8 | The Second Activating Glucokinase Mutation (A456V): Implications for Glucose Homeostasis and Diabetes Therapy. Diabetes, 2002, 51, 1240-1246. | 0.3 | 162 |
| 9 | Permanent diabetes mellitus in the first year of life. Diabetologia, 2002, 45, 798-804. | 2.9 | 150 |
| 10 | KCNJ11activating mutations in Italian patients with permanent neonatal diabetes. Human Mutation, 2005, 25, 22-27. | 1.1 | 131 |
| 11 | Minimal incidence of neonatal/infancy onset diabetes in Italy is 1:90,000 live births. Acta Diabetologica, 2012, 49, 405-408. | 1.2 | 130 |
| 12 | Insulin Gene Mutations as Cause of Diabetes in Children Negative for Five Type 1 Diabetes Autoantibodies. Diabetes Care, 2009, 32, 123-125. | 4.3 | 126 |
| 13 | 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 |
| 14 | Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. American Journal of Human Genetics, 2015, 97, 177-185. | 2.6 | 114 |
| 15 | Role of transglutaminase 2 in glucose tolerance: knockout mice studies and a putative mutation in a MODY patient. FASEB Journal, 2002, 16, 1371-1378. | 0.2 | 107 |
| 16 | INS-gene mutations: From genetics and beta cell biology to clinical disease. Molecular Aspects of Medicine, 2015, 42, 3-18. | 2.7 | 106 |
| 17 | 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, 1054-1061. | 1.8 | 100 |
| 18 | Mutant INS-Gene Induced Diabetes of Youth: Proinsulin Cysteine Residues Impose Dominant-Negative Inhibition on Wild-Type Proinsulin Transport. PLoS ONE, 2010, 5, e13333. | 1.1 | 100 |

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|---|---|---|---|
| 19 | Maturity-Onset Diabetes of the Young in Children With Incidental Hyperglycemia:. Diabetes Care, 2009, 32, 1864-1866. | 4.3 | 97 |
| 20 | 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. | 1.8 | 88 |
| 21 | 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, 14105-14113. | 1.6 | 87 |
| 22 | Syndromes of Autoimmunity and Hypoglycemia: Autoantibodies Directed Against insulinand Its Receptor. Endocrinology and Metabolism Clinics of North America, 1989, 18, 123-143. | 1.2 | 84 |
| 23 | An ATP-Binding Mutation (G334D) in KCNJ11 Is Associated With a Sulfonylurea-Insensitive Form of Developmental Delay, Epilepsy, and Neonatal Diabetes. Diabetes, 2007, 56, 328-336. | 0.3 | 82 |
| 24 | Further evidence that mutations in INScan be a rare cause of Maturity-Onset Diabetes of the Young (MODY). BMC Medical Genetics, 2010, 11, 42. | 2.1 | 67 |
| 25 | Î² Cell Replacement after Gene Editing of a Neonatal Diabetes-Causing Mutation at the Insulin Locus. Stem Cell Reports, 2018, 11, 1407-1415. | 2.3 | 67 |
| 26 | Mutations at the Same Residue (R50) of Kir6.2 (KCNJ11) That Cause Neonatal Diabetes Produce Different Functional Effects. Diabetes, 2006, 55, 1705-1712. | 0.3 | 64 |
| 27 | Permanent diabetes during the first year of life: multiple gene screening in 54 patients. Diabetologia, 2011, 54, 1693-1701. | 2.9 | 63 |
| 28 | Impaired Cleavage of Preproinsulin Signal Peptide Linked to Autosomal-Dominant Diabetes. Diabetes, 2012, 61, 828-837. | 0.3 | 61 |
| 29 | 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. | 1.1 | 61 |
| 30 | 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, 2210-2213. | 2.9 | 55 |
| 31 | GENETIC BASIS OF ENDOCRINE DISEASE 1 Molecular Genetics of Insulin Resistant Diabetes Mellitus. Journal of Clinical Endocrinology and Metabolism, 1991, 73, 1158-1163. | 1.8 | 54 |
| 32 | Renal cysts and diabetes syndrome linked to mutations of the hepatocyte nuclear factor-1Î² gene: Description of a new family with associated liver involvement. American Journal of Kidney Diseases, 2002, 40, 397-402. | 2.1 | 54 |
| 33 | Glucose tolerance status in 510 children and adolescents attending an obesity clinic in Central Italy. Pediatric Diabetes, 2010, 11, 47-54. | 1.2 | 52 |
| 34 | Sexual dimorphism of body composition and insulin sensitivity across pubertal development in obese Caucasian subjects. European Journal of Endocrinology, 2009, 160, 769-775. | 1.9 | 51 |
| 35 | Transient neonatal diabetes mellitus is associated with a recurrent (R201H) KCNJ11 (KIR6.2) mutation. Diabetologia, 2005, 48, 2439-2441. | 2.9 | 41 |
| 36 | A convenient microwave-assisted synthesis of N-glycosyl amino acids. Tetrahedron Letters, 2007, 48, 2901-2904. | 0.7 | 38 |

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|---|---|---|---|
| 37 | 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, 951-957. | 1.2 | 37 |
| 38 | Genetic causes and treatment of neonatal diabetes and early childhood diabetes. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 575-591. | 2.2 | 37 |
| 39 | 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. | 1.8 | 37 |
| 40 | 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. | 1.8 | 36 |
| 41 | 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, 164-169. | 1.8 | 35 |
| 42 | Role of the ENPP1 K121Q Polymorphism in Glucose Homeostasis. Diabetes, 2008, 57, 3360-3364. | 0.3 | 35 |
| 43 | 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. | 1.1 | 33 |
| 44 | Obese Children with Low Birth Weight Demonstrate Impaired Î²-Cell Function during Oral Glucose Tolerance Test. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4448-4452. | 1.8 | 32 |
| 45 | Serological Proteome Analysis (SERPA) as a tool for the identification of new candidate autoantigens in type 1 diabetes. Journal of Proteomics, 2013, 82, 263-273. | 1.2 | 32 |
| 46 | N<sup>Î±</sup>â€Fmocâ€Protected Ï‰â€Azidoâ€•and Ï‰â€Alkynylâ€<scp>L</scp>â€amino Acids as Building Blocks for the Synthesis of â€œClickableâ€•Peptides. European Journal of Organic Chemistry, 2008, 2008, 5308-5314. | 1.2 | 30 |
| 47 | 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, 1983-1989. | 3.7 | 30 |
| 48 | No beta cell desensitisation after a median of 68Âmonths on glibenclamide therapy in patients with KCNJ11-associated permanent neonatal diabetes. Diabetologia, 2011, 54, 2736-2738. | 2.9 | 30 |
| 49 | Glyburide ameliorates motor coordination and glucose homeostasis in a child with diabetes associated with theKCNJ11/S225T, del226-232 mutation. Pediatric Diabetes, 2012, 13, 656-660. | 1.2 | 28 |
| 50 | Successful treatment of young infants presenting neonatal diabetes mellitus with continuous subcutaneous insulin infusion before genetic diagnosis. Acta Diabetologica, 2016, 53, 559-565. | 1.2 | 28 |
| 51 | Mutations in the Insulin Receptor Gene in Patients with Genetic Syndromes of Insulin Resistance and Acanthosis Nigricans. Journal of Investigative Dermatology, 1992, 98, S77-S81. | 0.3 | 25 |
| 52 | <b><i>IGF2</i></b> Methylation Is Associated with Lipid Profile in Obese Children. Hormone Research in Paediatrics, 2013, 79, 361-367. | 0.8 | 25 |
| 53 | Insulin therapy in neonatal diabetes mellitus: a review of the literature. Diabetes Research and Clinical Practice, 2017, 129, 126-135. | 1.1 | 25 |
| 54 | Increased OB gene expression leads to elevated plasma leptin concentrations in patients with chronic primary hyperinsulinemia. Diabetes, 1998, 47, 1625-1629. | 0.3 | 24 |

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|---|---|---|---|
| 55 | 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-e18. | 1.1 | 24 |
| 56 | 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 |
| 57 | 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-e260. | 4.3 | 23 |
| 58 | 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, 10437-10447. | 2.9 | 22 |
| 59 | Diabetes associated with dominant insulin gene mutations: outcome of 24-month, sensor-augmented insulin pump treatment. Acta Diabetologica, 2016, 53, 499-501. | 1.2 | 22 |
| 60 | Reduced replication fork speed promotes pancreatic endocrine differentiation and controls graft size. JCI Insight, 2021, 6, . | 2.3 | 22 |
| 61 | Biosensor analysis of anti-citrullinated protein/peptide antibody affinity. Analytical Biochemistry, 2014, 465, 96-101. | 1.1 | 20 |
| 62 | 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. | 1.1 | 19 |
| 63 | Ketogenic diet in a patient with congenital hyperinsulinism: a novel approach to prevent brain damage. Orphanet Journal of Rare Diseases, 2015, 10, 120. | 1.2 | 19 |
| 64 | Congenital Hyperinsulinism and Glucose Hypersensitivity in Homozygous and Heterozygous Carriers of Kir6.2 (KCNJ11) Mutation V290M Mutation. Diabetes, 2011, 60, 209-217. | 0.3 | 17 |
| 65 | 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, 1999, 48, 403-407. | 0.3 | 16 |
| 66 | TRIB3 R84 variant affects glucose homeostasis by altering the interplay between insulin sensitivity and secretion. Diabetologia, 2010, 53, 1354-1361. | 2.9 | 16 |
| 67 | 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, 1-6. | 0.5 | 16 |
| 68 | Thyroid Function Tests in Obese Prepubertal Children: Correlations with Insulin Sensitivity and Body Fat Distribution. Hormone Research in Paediatrics, 2012, 78, 100-105. | 0.8 | 16 |
| 69 | 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 KATP Gain of Channel Activity and Loss of Channel Expression. PLoS ONE, 2013, 8, e63758. | 1.1 | 16 |
| 70 | Focal congenital hyperinsulinism managed by medical treatment: a diagnostic algorithm based on molecular genetic screening. Clinical Endocrinology, 2014, 81, 679-688. | 1.2 | 16 |
| 71 | 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. | 1.2 | 14 |
| 72 | Differences between transient neonatal diabetes mellitus subtypes can guide diagnosis and therapy. European Journal of Endocrinology, 2021, 184, 575-585. | 1.9 | 13 |

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|---|---|---|---|
| 73 | Diagnosis of Neonatal and Infancy-Onset Diabetes. , 2007, 11, 83-93. | | 12 |
| 74 | A possible role of transglutaminase 2 in the nucleus of INS-1E and of cells of human pancreatic islets. Journal of Proteomics, 2014, 96, 314-327. | 1.2 | 12 |
| 75 | Case Report: When an Induced Illness Looks Like a Rare Disease. Pediatrics, 2015, 136, e1361-e1365. | 1.0 | 12 |
| 76 | Cardiovascular Fitness, Insulin Resistance and Metabolic Syndrome in Severely Obese Prepubertal Italian Children. Hormone Research in Paediatrics, 2008, 70, 349-356. | 0.8 | 9 |
| 77 | Prevalence of elevated 1-h plasma glucose and its associations in obese youth. Diabetes Research and Clinical Practice, 2016, 116, 202-204. | 1.1 | 9 |
| 78 | Hyperglucagonemia in an animal model of insulin- deficient diabetes: what therapy can improve it?. Clinical Diabetes and Endocrinology, 2016, 2, 11. | 1.3 | 9 |
| 79 | The expression of four pyridoxal kinase (PDXK) human variants in Drosophila impacts on genome integrity. Scientific Reports, 2019, 9, 14188. | 1.6 | 9 |
| 80 | 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-e182. | 4.3 | 8 |
| 81 | Growth Hormone Does not Inhibit Its Own Secretion during Prolonged Hypoglycemia in Man. Journal of Clinical Endocrinology and Metabolism, 1990, 70, 1371-1374. | 1.8 | 7 |
| 82 | 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. | 2.7 | 7 |
| 83 | Severe insulin resistance in disguise: A familial case of reactive hypoglycemia associated with a novel heterozygous INSR mutation. Pediatric Diabetes, 2018, 19, 670-674. | 1.2 | 7 |
| 84 | Single-strand conformation polymorphism analysis of the glucose transporter gene GLUT1 in maturity-onset diabetes of the young. Journal of Molecular Medicine, 2001, 79, 270-274. | 1.7 | 6 |
| 85 | Neonatal Diabetes: Permanent Neonatal Diabetes and Transient Neonatal Diabetes. Frontiers in Diabetes, 2017, , 1-25. | 0.4 | 6 |
| 86 | The application of precision medicine in monogenic diabetes. Expert Review of Endocrinology and Metabolism, 2022, 17, 111-129. | 1.2 | 6 |
| 87 | SGLT2i Improves Glycemic Control in Patients With Congenital Severe Insulin Resistance. Pediatrics, 2022, 150, . | 1.0 | 6 |
| 88 | Role of proline 193 in the insulin receptor post-translational processing. Diabetologia, 1999, 42, 435-442. | 2.9 | 5 |
| 89 | Evaluation of new immunological targets in neuromyelitis optica. Journal of Peptide Science, 2013, 19, 25-32. | 0.8 | 5 |
| 90 | Macrosomia, transient neonatal hypoglycemia, and monogenic diabetes in a family with heterozygous mutation R154X of HNF4A gene. Journal of Endocrinological Investigation, 2011, 34, 252-253. | 1.8 | 4 |

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|---|---|---|---|
| 91 | Insulin: still a miracle after all these years. Journal of Clinical Investigation, 2019, 129, 3045-3047. | 3.9 | 4 |
| 92 | Congenital diabetes mellitus. Minerva Pediatrica, 2020, 72, 240-249. | 2.6 | 4 |
| 93 | GCK Mutation in a Child with Maturity Onset Diabetes of the Young, Type 2. Iranian Journal of Pediatrics, 2013, 23, 226-8. | 0.1 | 4 |
| 94 | Contribution of ONECUT1 variants to different forms of non-autoimmune diabetes mellitus in Italian patients. Acta Diabetologica, 2022, 59, 1113-1116. | 1.2 | 4 |
| 95 | Mutations in IAPP and NEUROG3 genes are not a common cause of permanent neonatal/infancy/childhoodâ€onset diabetes. Diabetic Medicine, 2009, 26, 660-661. | 1.2 | 3 |
| 96 | 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, 332-334. | 1.1 | 3 |
| 97 | MEHMO syndrome and the link between brain, pituitary and pancreas. EBioMedicine, 2019, 42, 26-27. | 2.7 | 3 |
| 98 | 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, . | 0.3 | 3 |
| 99 | Type 2 diabetes in pediatrics. Minerva Pediatrics, 2021, , . | 0.2 | 2 |
| 100 | Sulfonylurea-Insensitive Permanent Neonatal Diabetes Caused by a Severe Gain-of-Function Tyr330His Substitution in Kir6.2. Hormone Research in Paediatrics, 2022, 95, 215-223. | 0.8 | 2 |
| 101 | Intrafamilial Variability of Early-Onset Diabetes due to anINSMutation. Case Reports in Genetics, 2011, 2011, 1-5. | 0.1 | 1 |
| 102 | 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. | 2.9 | 1 |
| 103 | 1264-P: Distinguishing between Obese Patients with Type 1 Diabetes (T1DMob) and Type 2 Diabetes in Adolescence (T2DMad) at Presentation. Diabetes, 2020, 69, . | 0.3 | 1 |
| 104 | 1636-P: Transient Neonatal Diabetes: Clinical Differences between Patients Bearing KATP Mutations and 6q24 Defects May Guide Genetic Screening. Diabetes, 2020, 69, 1636-P. | 0.3 | 1 |
| 105 | Pre-diabetes in Italian obese children and youngsters. Journal of Endocrinological Investigation, 2011, 34, e275-80. | 1.8 | 1 |
| 106 | Clinical and molecular evaluation of Italian patients affected by Pelizaeus-Merzbacher disease. Journal of Inherited Metabolic Disease, 1996, 19, 197-200. | 1.7 | 0 |
| 107 | Not Autoimmune Diabetes Mellitus in Paediatrics. , 2017, , 137-146. | | 0 |
| 108 | Very low birth weight newborn with diabetes mellitus due to pancreas agenesis managed with insulin pump reservoir filled with undiluted insulin: 16-month follow-up. Diabetes and Metabolic Syndrome: Clinical Research and Reviews, 2022, 16, 102561. | 1.8 | 0 |