Università degli Studi di Roma "Tor Vergata"

CURRICULUM DIDATTICO-SCIENTIFICO DEL PROF. FABRIZIO BARBETTI

DATI PERSONALI
Nome e Cognome: Fabrizio Barbetti
Luogo e data di nascita: Roma, 29/06/1952

***ATTUALE POSIZIONE*:** Professore Associato
Dipartimento: Medicina Sperimentale e Chirurgia
Indirizzo: Policlinico Tor Vergata, Viale Oxford 81, 1° piano Settore D, stanza 118
Numero studio: 06-2090-0672
E-mail: fabrizio.barbetti@uniroma2.it
Orario ricevimento: il martedì, dalle 14.30 alle 15.30
Settore scientifico-disciplinare: BIO12

ATTIVITA' DIDATTICA - SCIENTIFICA
Titoli accademici e di studio: Laurea in Medicina e Chirurgia, Un. La Sapienza, Roma 1977, 110/110 con lode. Specializzazione in Endocrinologia, Un. La Sapienza, Roma, 1981 70/0 con lode Dottore di Ricerca in Scienze Endocrinologiche e Metaboliche (I ciclo), Un. La Sapienza, 1987
Formazione post-laurea presso istituzioni italiane ed estere ed incarichi professionali (didattici e di ricerca): 1987-1990: Fogarty Fellow, Diabetes Branch, NIDDK, NIH, Bethesda, MD, USA; 1990-1991: Visiting Associate, Diabetes Branch, NIDDK, NIH, Bethesda, MD, USA.
2009-2010: Visiting Associate Professor, Division of Endocrinology, Columbia University, New York, NY, USA
Finanziamenti e premi ricevuti per attività di ricerca:
2014: Ministero della Salute – Ricerca Finalizzata 2011-2012 – PE 2011- 02350284. P.I. Fabrizio Barbetti. Search for new genes a new therapies for Neonatal Diabetes Mellitus (NDM) and Hyperinsulinism and Hypoglycemia (HH). Durata: triennale. Finanziamento: 219.000 €
2011: ESPE Collaborative research project. P.I. Fabrizio Barbetti**.** Finanziamento: 30.000 € **2009: E-RARE** – Progetto/Consorzio: European Network on Genetics, Pathophysiology and Translational Research into Rare Pancreatic Beta-Cell Insufficiency Diseases. Finanziamento: 59.000,00€
2009: Fondazione Roma – Progetto: Molecular mechanisms in the pathogenesis of type 2 diabetes mellitus and its cardiovascular complications. Finanziamento: 180.000 €
2009: TELETHON – Progetto GGP09147. P.I. O. Massa (ricercatrice Barbetti). Finaziamento: 52.000€
2006: **2006**: PRIN. Responsabile Unità Univ. Tor Vergata. Protocollo 2006067105_003 - **2004**: Telethon, Prog. n. GGP04264, P.I. F, Barbetti, biennale. Totale finanziato: 106.500 € **2003**: Ricerca strategica Ministero della Sanità, Coordinatore/P.I.: F. Barbetti
2002: Progetto strategico sul diabete tipo 2, Ministero della Sanità bando 2002, Unità operativa Ospedale Bambino Gesù coordinata da Dr. F. Barbetti (P.I. Prof. G. Pozza, H S Raffaele, Milano).
2001: Progetto FIRB
2001: Progetto **ASI (Agenzia Spaziale Italiana),** Bando **2001**, biennale.
1999: Progetto multicentrico **Telethon n° E.948**, biennale. Coordinatore/P.I. Dr. F. Barbetti. **1998**: Ric. Finalizzata Min. della Sanità, Unità operativa H S Raffaele: Dr. F. Barbetti (P.I.: Dr. V. Trischitta, Osp. S. Giovanni Rotondo, **ICS 160.3/RF98/76**).
1997: **1)** Prog. **Telethon n° E.591**; **2)** Ric. Finalizzata **Min. della Sanità**, Rifer: **RF97.19,** biennale. P.I.: Dr. F. Barbetti; **3)** Fondi istituzionali H S Raffaele: PZ801.
1996: 1) Prog. **Telethon n° E.483**; **2)** Fondi istituzionali H S Raffaele PZ 701
1995: 1) Prog. **Telethon n° E.228**; **2)** Fondi istituzionali H S Raffaele: PZ 601 e 602
1994: 1) Fondo del **Ministero della Sanità**, contratto n° L1660026**; 2)** Prog**. Telethon n. E.098**; **3)**

Prog. Telethon n. E.46, come Unità esterna; Fondi istituzionali H S Raffaele: PZ29510.

Attività di ricerca: 15 pubblicazioni selezionate

1) Taylor SI, …, **Barbetti F**, …, Kadowaki T (**1992**) Mutations in the insulin receptor gene. Endocr Rev 13: 566-595. **(*250 citazioni*)**

2) Njolstad PR, …, **Barbetti F**, …, Bell GI. (**2001**) Neonatal diabetes mellitus due to complete glucokinase deficiency. N Engl J Med 344:1588-1592 **(*296 citazioni*)**

3) Massa O, …, **Barbetti F**. (**2001**) High prevalence of glucokinase mutations in Italian children with MODY. Influence on glucose tolerance, first-phase insulin response, insulin sensitivity and BMI. Diabetologia 44:898-905 **(94 citazioni)**

4) Christesen HBT, …, **Barbetti F.** (**2002**) The second activating glucokinase mutation (A456V): implications for glucose homeostasis and diabetes therapy. Diabetes 51: 1240-1246 **(*116 citazioni*)**

5) Foti D, …, **Barbetti F**, ….,Brunetti A. (**2005**) Lack of the architectural factor HMGA1 causes insulin resistance and diabetes in humans and mice. Nat Med 11: 765-773 **(*135 citazioni*)**

6) Massa O, …, **Barbetti F.** (**2005**) KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes. Hum Mutat 25: 22-27 **(*118 citazioni*)**

7) Koster JC, …, **Barbetti F**. (**2008**) The G53D mutation in Kir6.2 (KCNJ11) is associated with neonatal diabetes and motor dysfunction in adulthood that is improved with sulfonylurea therapy. J Clin Endocrinol Metab 93: 1054-1061. **(*62 citazioni*)**

8) Colombo C, …, **Barbetti F**. (**2008**) Seven mutations in the human insulin gene linked to permanent neonatal/infancy-onset diabetes mellitus. J Clin Invest 118:2148-2156 **(*123 citazioni*)**

9) Bonfanti R,..., **Barbetti F**. (**2009**) Insulin gene mutations as cause of diabetes in children negative for five type 1 diabetes autoantibodies. Diabetes Care 32:123-125 **(44 citazioni)**

10) Loechner KJ, …, **Barbetti F**, …, Nichols CG. (**2011**) Congenital hyperinsulinism and glucose hypersensitivity in homozygous and heterozygous carriers of Kir6.2 (KCNJ11) mutation V290M mutation. KATP channel inactivation mechanism and clinical management. Diabetes 60:209-217 (**15 citazioni**)

11) Iafusco D, …, **Barbetti F** (**2014**) No sign of proliferatve retinopathy in 15 patients with Permanent Neonatal Diabetes Mellitus with a median diabetes duration of 24 years. Diabetes Care, 37:e181-e182 (**20 citazioni**)

12) Delvecchio M, ..., **Barbetti F**. (**2014**) 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 37:e258-e260 (**10 citazioni**)

13) Prudente S, ..., **Barbetti F**, ..., Doria A. (**2015**) Loss-of-function mutations in the *APPL1* gene in familial diabetes mellitus. Am J Hum Genet 97:177-185 (**30 citazioni**)

14) Delvecchio M, ..., **Barbetti F**. (**2017**) Monogenic Diabetes accounts for 6.3% of cases referred to 15 Italian pediatric diabetes Centers during 2007-2012. J Clin Endocrinol Metab 102:1826-1834 (**4 citazioni**)

15) Bowman P, ..., **Barbetti F**, ..., Njolstad PR. (**2018**) Long-term treatment with sulfonylureas is highly effective and safe in neonatal diabetes due to KCNJ11 mutations: an international cohort study. Lancet Diabetes Endocrinol, pubblicato online 04/06/2018 doi.org/10.1016/S2213-8587(18)30106-2 (1 citazione)

Università degli Studi di Roma "Tor Vergata"

ACADEMIC AND SCIENTIFIC CURRICULUM OF PROF. FABRIZIO BARBETTI

PERSONAL DATA

Name and Surname: Fabrizio Barbetti
Place and date of birth: Rome (Italy), 29/06/1952

***CURRENT POSITION*:** Associate Professor of Clinical Biochemistry and Molecular Biology
Department: Experimental Medicine and Surgery
Address: Tor Vergata University Hospital, Viale Oxford 81, 1st floor, Section D, Room 118
Telephone (office): 06-2090-0672; Lab.: 06-2090-2271
E-mail fabrizio.barbetti@uniroma2.it
Office hours: Tuesday, 14.30-15.30 (2.30-3.30 p.m.)

ACADEMIC TRAINING – POSITIONS HELD

1977: M.D. La Sapienza University, Rome, Italy.
1981: Specialty Board in Endocrinology, La Sapienza University, Rome, Italy
1988: Ph.D. La Sapienza Univeristy, Rome, Italy.
Post-graduate education and positions held
1987-1990: Fogarty Fellow NIDDK, NIH, Bethesda, MD, USA;
1990-1991: Visiting Associate, Diabetes Branch, NIDDK, NIH, Bethesda, MD, USA.
2009-2010: Visiting Associate Professor, Division of Endocrinology, Columbia University, New York, NY, USA
2005-to date: Chief, Laboratory of Monogenic Diabetes, Tor Vergata University Hospital **2012-to date**: Chief, Autoimmunity Section, Department of Laboratory Medicine, Tor Vergata University Hospital

SCIENTIFIC ACTIVITY

Grant support:
2014: Ministero della Salute – Ricerca Finalizzata 2011-2012 – PE 2011- 02350284. P.I. Fabrizio Barbetti. Search for new genes a new therapies for Neonatal Diabetes Mellitus (NDM) and Hyperinsulinism and Hypoglycemia (HH). 219.000 €
2011: ESPE Collaborative research project. 30.000 €
2009: E-RARE –European Network on Genetics, Pathophysiology and Translational Research into Rare Pancreatic Beta-Cell Insufficiency Diseases. 59.000 €
2009: Fondazione Roma –Molecular mechanisms in the pathogenesis of type 2 diabetes mellitus and its cardiovascular complications. 180.000 €
2009: TELETHON – Project # GGP09147. P.I. O. Massa (Barbetti's post-doctoral fellow) 52.000 €
2006: PRIN. # 2006067105_003 -
2004: Telethon, Proj. # GGP04264 106.500 €
2003: Ricerca strategica Ministero della Sanità, P.I.: F. Barbetti
2002: **Ministry of Health,** Progetto **strategico** sul diabete tipo 2, Unità operativa Ospedale Bambino Gesù
2001: Progetto **FIRB**
2001: Progetto **ASI (Agenzia Spaziale Italiana),** Bando **2001**, biennale.
1999: **Telethon # E.948**.
1998: **Ministry of Health** , Ric. Finalizzata **ICS 160.3/RF98/76**.
1997: **1) Telethon # E.591**; **2) Ministry of Health** Ric. Finalizzata, **RF97.19,** Barbetti; **3 1996: 1) Telethon # E.483**;
1995: 1) Telethon # E.228;

1994: 1) Ministry of Health, contract # L1660026**; 2) Telethon # E.098**; **3)** Telethon # E.46

Sciantific activity: publications (selection of 15)

1) Taylor SI, …, **Barbetti F**, …, Kadowaki T (**1992**) Mutations in the insulin receptor gene. Endocr Rev 13: 566-595. **(*250 citations*)**

2) Njolstad PR, …, **Barbetti F**, …, Bell GI. (**2001**) Neonatal diabetes mellitus due to complete glucokinase deficiency. N Engl J Med 344:1588-1592 **(*296 citazioni*)**

3) Massa O, …, **Barbetti F**. (**2001**) High prevalence of glucokinase mutations in Italian children with MODY. Influence on glucose tolerance, first-phase insulin response, insulin sensitivity and BMI. Diabetologia 44:898-905 **(94 citations)**

4) Christesen HBT, …, **Barbetti F.** (**2002**) The second activating glucokinase mutation (A456V): implications for glucose homeostasis and diabetes therapy. Diabetes 51: 1240-1246 **(*116 citations*)**

5) Foti D, …, **Barbetti F**, ….,Brunetti A. (**2005**) Lack of the architectural factor HMGA1 causes insulin resistance and diabetes in humans and mice. Nat Med 11: 765-773 **(*135 citations*)**

6) Massa O, …, **Barbetti F.** (**2005**) KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes. Hum Mutat 25: 22-27 **(*118 citations*)**

7) Koster JC, …, **Barbetti F**. (**2008**) The G53D mutation in Kir6.2 (KCNJ11) is associated with neonatal diabetes and motor dysfunction in adulthood that is improved with sulfonylurea therapy. J Clin Endocrinol Metab 93: 1054-1061. **(*62 citations*)**

8) Colombo C, …, **Barbetti F**. (**2008**) Seven mutations in the human insulin gene linked to permanent neonatal/infancy-onset diabetes mellitus. J Clin Invest 118:2148-2156 **(*123 citations*)**

9) Bonfanti R,..., **Barbetti F**. (**2009**) Insulin gene mutations as cause of diabetes in children negative for five type 1 diabetes autoantibodies. Diabetes Care 32:123-125 **(44 citations)**

10) Loechner KJ, …, **Barbetti F**, …, Nichols CG. (**2011**) Congenital hyperinsulinism and glucose hypersensitivity in homozygous and heterozygous carriers of Kir6.2 (KCNJ11) mutation V290M mutation. KATP channel inactivation mechanism and clinical management. Diabetes 60:209-217 (**15 citations**)

11) Iafusco D, …, **Barbetti F** (**2014**) No sign of proliferatve retinopathy in 15 patients with Permanent Neonatal Diabetes Mellitus with a median diabetes duration of 24 years. Diabetes Care, 37:e181-e182 (**20 citations**)

12) Delvecchio M, ..., **Barbetti F**. (**2014**) 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 37:e258-e260 (**10 citations**)

13) Prudente S, ..., **Barbetti F**, ..., Doria A. (**2015**) Loss-of-function mutations in the *APPL1* gene in familial diabetes mellitus. Am J Hum Genet 97:177-185 (**30 citations**)

14) Delvecchio M, ..., **Barbetti F**. (**2017**) Monogenic Diabetes accounts for 6.3% of cases referred to 15 Italian pediatric diabetes Centers during 2007-2012. J Clin Endocrinol Metab 102:1826-1834 (**4 citations**)

15) Bowman P, ..., **Barbetti F**, ..., Njolstad PR. (**2018**) Long-term treatment with sulfonylureas is highly effective and safe in neonatal diabetes due to KCNJ11 mutations: an international cohort study. Lancet Diabetes Endocrinol, pubblicato online 04/06/2018 doi.org/10.1016/S2213-8587(18)30106-2 (**1 citation**)