

BIOGRAPHICAL SKETCH

Provide the following information for the key personnel and other significant contributors in the order listed on Form Page 2.
Follow this format for each person. **DO NOT EXCEED FOUR PAGES.**

NAME Maffei, Pietro	POSITION TITLE M.D. Internal Medicine
eRA COMMONS USER NAME	Medical Licence n°: 6668

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)*

INSTITUTION AND LOCATION	DEGREE (if applicable)	YEAR(s)	FIELD OF STUDY
Padua University, Italy	M.D.	1989	Medicine
Padua University, Italy	Specialization	1994	Internal Medicine
Padua University, Italy	PhD	1999	Clinical Methodology
Padua University, Italy	Specialization	2002	Cardiology
Azienda Ospedaliera Padova	Certification	2007	Endocrinology

NOTE: The Biographical Sketch may not exceed four pages. Follow the formats and instructions on the attached sample.

- A. **Positions and Honors.** List in chronological order previous positions, concluding with your present position. List any honors. Include present membership on any Federal Government public advisory committee.

Positions and Employment

- 1989-1997 Fellow of Internal Medicine Department – University Hospital - Padua
 1990-1991 MD, Italian Navy
 1991-1997 On-call Emergency Service for outpatients of Veneto Region
 1994-1994 Medical assistance at Hyperbaric room (Venice);
 1995-1999 Medical assistance as volunteer for underserved population (Padua);
 1997- Permanent position - Internal Medicine Department – DIMED, University Hospital - Padua

Professional Memberships

2003. Teaching position – Specialization School of Endocrinology – Padua University
 1999- Member, Italian Society of Endocrinology (SIE)
 2001- Member, Italian Society of Cardiology (SIC)
 2002- Member, Scientific Advisors Committee, Alstrom Syndrome International (ASI)
 2002- Member, Board of Directors, Alstrom Syndrome International (ASI)
 2006- Member, Italian Association of Clinical Endocrinologist (AME)
 2007- Teaching board, PhD Course in Clinical and Experimental Sciences: Clinical Methodology and medicine of physical exercise, diabetic, endocrinological and nephrological sciences– Dep. of Medicine - Padua University

- 2009- Member, European Society of Cardiology
- 2014- President and funder of ASS.A.I (Associazione Sindrome di Alstrom Italia)
- 2014- Member and funder of Alstrom Europe
- 2017- Teaching board, Master in “Communication of Sciences” – Dep. of Physics and Astronomy, Padua University
- 2017- Regional representative for Veneto and Trentino-Alto Adige of the Italian Association of Clinical Endocrinologist (AME)

Other Experience

Clinical research has been centered in the areas of endocrinology and metabolism with particular dedication to acromegaly and pituitary tumours or dysfunctions, thyroid diseases, diabetes, obesity, and relationships between hormones and the cardiovascular system. Attended three different Consensus Conference on Acromegaly (Cortina 1999; Miami 2007; Paris 2009). Expertise in Alstrom syndrome and endocrine-metabolic complications of Thalassemia Major.

- B. **Selected peer-reviewed publications (in chronological order).** Do not include publications submitted or in preparation.

Publications on Alstrom syndrome (PubMed)

- 1) Citton V, **Maffei P**, Marshall JD, Baglione A, Collin GB, Milan G, Vettor R, Naggert JK, Manara R. Pituitary morphovolumetric changes in Alström syndrome. *J Neuroradiol.* 2016 Jun;43(3):195-9.
- 2) Paisey RB, Smith J, Carey C, Barrett T, Campbell F, **Maffei P**, Marshall JD, Paisey C, Steeds RP, Edwards NC, Bunce S, Geberhiwot T. Duration of Diabetes Predicts Aortic Pulse Wave Velocity and Vascular Events in Alström Syndrome. *J Clin Endocrinol Metab.* 2015 Aug;100(8):E1116-24.
- 3) Marshall JD, Muller J, Collin GB, Milan G, Kingsmore SF, Dinwiddie D, Farrow EG, Miller NA, Favaretto F, **Maffei P**, Dollfus H, Vettor R, Naggert JK. Alström Syndrome: Mutation Spectrum of ALMS1. *Hum Mutat.* 2015 Jul;36(7):660-8.
- 4) Manara R, Citton V, **Maffei P**, Marshall JD, Naggert JK, Milan G, Vettor R, Baglione A, Vitale A, Briani C, Di Salle F, Favaro A. Degeneration and plasticità of the optic pathway in Alström syndrome. *AJNR Am J Neuroradiol.* 2015 Jan;36(1):160-5
- 5) Favaretto F, Milan G, Collin GB, Marshall JD, Stasi F, **Maffei P**, Vettor R, Naggert JK. GLUT4 defects in adipose tissue are early signs of metabolic alterations in Alms1^{GT/GT}, a mouse model for obesity and insulin resistance. *PLoS One.* 2014 Oct 9;9(10):e109540.
- 6) Milani D, Cerutti M, Pezzani L, **Maffei P**, Milan G, Esposito S. Syndromic obesity: clinical implications of a correct diagnosis. *Ital J Pediatr.* 2014 Apr 2;40(1):33.
- 7) Farmer A, Aymé S, de Heredia ML, **Maffei P**, McCafferty S, Młynarski W, Nunes V, Parkinson K, Paquis-Flucklinger V, Rohayem J, Sinnott R, Tillmann V, Tranebjaerg L, Barrett TG. EURO-WABB: an EU rare diseases registry for Wolfram syndrome, Alström syndrome and Bardet-Biedl syndrome. *BMC Pediatr.* 2013 Aug 27;13:130
- 8) Marshall JD, **Maffei P**, Beck S, Barrett TG, Paisey R, Naggert JK. Clinical utility gene card for: Alström Syndrome - update 2013. *Eur J Hum Genet.* 2013 Nov;21(11)
- 9) Romano S, **Maffei P**, Bettini V, Milan G, Favaretto F, Gardiman M, Marshall JD, Greggio NA, Pozzan GB, Collin GB, Naggert JK, Bronson R, Vettor R. Alström syndrome is associated with short stature and reduced GH reserve. *Clin Endocrinol (Oxf).* 2013 Oct;79(4):529-36.
- 10) Citton V, Favaro A, Bettini V, Gabrieli J, Milan G, Greggio NA, Marshall JD, Naggert JK, Manara R, **Maffei P**. Brain involvement in Alström syndrome. *Orphanet J Rare Dis.* 2013 Feb 13;8:24

- 11) Collin GB, Marshall JD, King BL, Milan G, **Maffei P**, Jagger DJ, Naggert JK. The Alström syndrome protein, ALMS1, interacts with α -actinin and components of the endosome recycling pathway. *PLoS One*. 2012;7(5):e37925
- 12) Corbetti F, Razzolini R, Bettini V, Marshall JD, Naggert J, Tona F, Milan G, **Maffei P**. Alström syndrome: cardiac magnetic resonance findings. *Int J Cardiol*. 2013 Aug 20;167(4):1257-63.
- 13) Marshall JD, **Maffei P**, Collin GB, Naggert JK. Alström syndrome: genetics and clinical overview. *Curr Genomics*. 2011 May;12(3):225-35.
- 14) Bettini V, **Maffei P**, Pagano C, Romano S, Milan G, Favaretto F, Marshall JD, Paisey R, Scolari F, Greggio NA, Tosetto I, Naggert JK, Siculo N, Vettor R. The progression from obesity to type 2 diabetes in Alström syndrome. *Pediatr Diabetes*. 2012 Feb;13(1):59-67.
- 15) Zulato E, Favaretto F, Veronese C, Campanaro S, Marshall JD, Romano S, Cabrelle A, Collin GB, Zavan B, Belloni AS, Rampazzo E, Naggert JK, Abatangelo G, Siculo N, **Maffei P**, Milan G, Vettor R. ALMS1-deficient fibroblasts over-express extra-cellular matrix components, display cell cycle delay and are resistant to apoptosis. *PLoS One*. 2011 Apr 26;6(4):e19081.
- 16) Marshall JD, **Maffei P**, Beck S, Barrett TG, Paisey RB. Clinical utility gene card for: Alström syndrome. *Eur J Hum Genet*. 2011 Oct;19(10).
- 17) Izzi C, **Maffei P**, Milan G, Tardanico R, Foini P, Marshall J, Marega A, Scolari F. The Case | Familial occurrence of retinitis pigmentosa, deafness, and nephropathy. *Kidney Int*. 2011 Mar;79(6):691-2.
- 18) Vingolo EM, Salvatore S, Grenga PL, **Maffei P**, Milan G, Marshall J. High-resolution spectral domain optical coherence tomography images of Alström syndrome. *J Pediatr Ophthalmol Strabismus*. 2010 May 21;47 Online:e1-3.
- 19) Kocova M, Sukarova-Angelovska E, Kacarska R, **Maffei P**, Milan G, Marshall JD. The unique combination of dermatological and ocular phenotypes in Alström syndrome: severe presentation, early onset and two novel ALMS1 mutations. *Br J Dermatol*. 2011 Apr;164(4):878-80.
- 20) Paisey RB, Paisey RM, Thomson MP, Bower L, **Maffei P**, Shield JP, Barnett S, Marshall JD. Protection from clinical peripheral sensory neuropathy in Alström syndrome in contrast to early-onset type 2 diabetes. *Diabetes Care*. 2009 Mar;32(3):462-4.
- 21) Romano S, Milan G, Veronese C, Collin GB, Marshall JD, Centobene C, Favaretto F, Dal Pra C, Scarda A, Leandri S, Naggert JK, **Maffei P**, Vettor R. Regulation of Alström syndrome gene expression during adipogenesis and its relationship with fat cell insulin sensitivity. *Int J Mol Med*. 2008 Jun;21(6):731-6.
- 22) Marshall JD, Beck S, **Maffei P**, Naggert JK. Alström syndrome. *Eur J Hum Genet*. 2007 Dec;15(12):1193-202.
- 23) Marshall JD, Hinman EG, Collin GB, Beck S, Cerqueira R, **Maffei P**, Milan G, Zhang W, Wilson DI, Hearn T, Tavares P, Vettor R, Veronese C, Martin M, So WV, Nishina PM, Naggert JK. Spectrum of ALMS1 variants and evaluation of genotype-phenotype correlations in Alström syndrome. *Hum Mutat*. 2007 Nov;28(11):1114-23
- 24) **Maffei P**, Boschetti M, Marshall JD, Paisey RB, Beck S, Resmini E, Collin GB, Naggert JK, Milan G, Vettor R, Minuto F, Siculo N, Barreca A. Characterization of the IGF system in 15 patients with Alström syndrome. *Clin Endocrinol (Oxf)*. 2007 Feb;66(2):269-75.
- 25) Marshall JD, Bronson RT, Collin GB, Nordstrom AD, **Maffei P**, Paisey RB, Carey C, Macdermott S, Russell-Eggitt I, Shea SE, Davis J, Beck S, Shatirishvili G, Mihai CM, Hoeltzenbein M, Pozzan GB, Hopkinson I, Siculo N, Naggert JK, Nishina PM. New Alström syndrome phenotypes based on the evaluation of 182 cases. *Arch Intern Med*. 2005 Mar 28;165(6):675-83
- 26) Paisey RB, Carey CM, Bower L, Marshall J, Taylor P, **Maffei P**, Mansell P. Hypertriglyceridaemia in Alström's syndrome: causes and associations in 37 cases. *Clin Endocrinol (Oxf)*. 2004 Feb;60(2):228-31.
- 27) **Maffei P**, Munno V, Marshall JD, Scandellari C, Siculo N. The Alström syndrome: is it a rare or unknown disease? *Ann Ital Med Int*. 2002 Oct-Dec;17(4):221-8.

28) Collin GB, Marshall JD, Ikeda A, So WV, Russell-Eggitt I, **Maffei P**, Beck S, Boerkoel CF, Sicolo N, Martin M, Nishina PM, Naggert JK. Mutations in ALMS1 cause obesity, type 2 diabetes and neurosensory degeneration in Alström syndrome. Nat Genet. 2002 May;31(1):74-8.

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- C. Research Support.** List selected ongoing or completed (during the last three years) research projects (federal and non-federal support). Begin with the projects that are most relevant to the research proposed in this application. Briefly indicate the overall goals of the projects and your role (e.g. PI, Co-Investigator, Consultant) in the research project. Do not list award amounts or percent effort in projects.

Protocollo A6291010; ACROSTUDY; A multicenter, post marketing surveillance study of Somavert therapy in patients with acromegaly in the US and Europe (since 2007).

This observational study investigates safety and efficacy of the GH receptor antagonist in acromegaly.

Role: Principal-Investigator

Executive Agency for Health and Consumer. An EU rare diseases registry for Wolfram syndrome, Alstrom syndrome and Bardet Biedl syndrome (EURO-Rare diabetes); agreement number 2010 12 05; since 03.01.2011, 36 months;

Main Partner and Project leader: University of Birmingham, Prof. T. Barrett; Associate Partners: University of Glasgow (UK); Centre National de la Recherche Scientifique, Valbonne (France); Fundació Institut Investigació Biomèdica de Bellvitge (Spain); Università degli Studi di Padova (Italy); Medical University of Lodz (Polonia); National Institute of Health and Medical Research (INSERM) (Paris, France); University of Tartu (Estonia); Alström Syndrome UK.

PHASE IIA OPEN-LABEL, DOSE ASCENDING STUDY TO INVESTIGATE THE PHARMACOKINETICS, PHARMACODYNAMICS, SAFETY AND TOLERABILITY OF A SINGLE DOSE OF LANREOTIDE PRF IN PATIENTS WITH ACROMEGALY PREVIOUSLY TREATED AND CONTROLLED WITH OCTREOTIDE LAR STUDY PROTOCOL. STUDY Number: 2-55-52030-309 LANREOTIDE PRF SOLUTION FOR INJECTION

Role: Principal-Investigator