CV Prof. Luisa Bernardinelli

Prof. Luisa Bernardinelli was born in Casalpusterlengo (Milan, Italy) on 15 February 1955.

Academic degree
Degree in Biological Sciences - University of Pavia (1978)

Other degrees
Post-graduate in Medical Statistics, University of Pavia: 50/50 summa cum laude (1983)

Present position
2003- Full Professor of Medical Statistics - Faculty of Medicine and Surgery, University of Pavia, Pavia (Italy)
2010- Head of Medical and Genomics Statistics Unit, Dept of Brain and Behavioural Sciences, University of Pavia, Pavia (Italy)
2013- Visiting Professor in Biostatistics, University of Manchester, Manchester (UK)
2012- Coordinator of the second-level postgraduate course in Medical Statistics and Genomics, University of Pavia, Pavia (Italy)
2014- : member of Scuola di Alta Formazione Dottorale (SAFD), University of Pavia, Pavia (Italy)

Previous positions
1983-1987: Associate Professor of Medical Statistics at the Specialization School in Sanitary Statistics- University of Pavia.
1988-1993: Associated Professor of Medical Statistics, Faculty of Medicine and Surgery- University of Sassari.
1993-2000: Associated Professor of Medical Statistics, Faculty of Medicine and Surgery- University of Pavia.
2001-2003: Associate Professor of Medical Statistics, Faculty of Medicine and Surgery- University of Pavia
2001-2007: visiting professor at the MRC Biostatistics Unit, Cambridge, UK
2007-2012: visiting professor at the Statistical Laboratory of the Centre of Mathematical Sciences, University of Cambridge, UK

Research Activity
STATISTICAL METHODOLOGY. Bayesian analysis of the geographical variation of the disease risk in space and time; Monte Carlo methods; graphical models; Bayesian estimates and their use in descriptive epidemiology; measurement errors in the covariates in ecological studies; development and application of statistical methods in genetic epidemiology: analysis of the association in presence of measurement error, incomplete data both in familiar studies and in case-control studies. Analysis of microarray, methylation and proteomic data. Analysis of pedigree data. Causal inference, joint analysis of genetic and gene expression data to identify genes causally related to Multiple Sclerosis. Analysis of next generation sequencing data. Bayesian Mendelian Randomization.

EPIDEMIOLOGICAL INVESTIGATION. Cancer epidemiology in Sardinia; epidemiology of mellitus insulin-dependent diabetes in Sardinia; epidemiology of multiple sclerosis in Sardinia; epidemiology of enuresis in schoolchildren. Geographical distribution of HLA using records of bone marrow donors in Lombardy; evaluation of social and welfare needs

in patients suffering from multiple sclerosis. Association studies between candidate polymorphisms, type 2 diabetes and early myocardial infarction. Cancer genetics. Genetic epidemiology of Inflammatory Bowel Disease. The causal direct effect of FTO on susceptibility to myocardial infarction. Identification of susceptibility genes of multiple sclerosis in the Nuoro province. Investigation of the biological function of ACCN1 and multiple sclerosis. Identification of causal biomarkers in multiple sclerosis via a Mendelian Randomization approach. The experience and interest in causal inference emerges from the organization of Workshops and Courses on causal inference to being one of the editor of the book "CAUSALITY: STATISTICAL PERSPECTIVES AND APPLICATIONS. Wiley, 2011". Her scientific interest to mindfulness-based meditation approach has led to the organization of the workshop "Scienza e Meditazione" held in Pavia, 9-10 October 2016. The workshop illustrated the evidence, the scientific methods and the different interpretations of meditation, plus practical sessions conducted by an expert meditator. She also organized for PhD students the intensive course (theory and practice) in Neuroscience of Meditation. She has recently set up a population based study to investigation the causal effect of a meditation based intervention on wellbeing and mental health.

Collaborations with international consortium
-MolPAGE, Molecular Phenotyping to Accelerate Genomic Epidemiology
-IMSGC, International Multiple Sclerosis Genetic Consortium
-TAG, Tobacco and Genetics Consortium
-MIGC, Myocardial Infarction Genetics Consortium
-MIMOmics, Methods for Integrated analysis of Multiple Omics datasets

Past and current International Research Collaborations

-Department of Statistics, University of Oxford, Oxford, UK
-School of Biological Science, University of East Anglia
-Carver College of Medicine, University of Iowa, USA
-Ghent University, Department of Applied Mathematics and Computer Science, Belgium
-LUMC, Leiden, NL
-KTH Royal Institute of Technology Stoccolma, SE
-Human Genetics, The Wellcome Trust SANGER Institute, Hinxton, UK
-European Bioinformatics Institute, The Wellcome Trust SANGER Institute, Hinxton
-University of Miami, Hussman Institute for Human Genomics, US
-Institute of Population Health, University of Manchester

Past and current National Research Collaborations

-ASL Nuoro, Centro di Tipizzazione
-ASL Nuoro, Divisione di Neurologia
-Università degli Studi di Milano
-Neuromed, Istituto Neurologico Mediterraneo
-Istituto Auxologico Italiano
-IEO istituto Europeo di Oncologia

Recent organized Post-graduate courses

- International Master in Genetic Epidemiology of the European School in Molecular Medicine and Genetic Epidemiology of the Istituto Universitario Superiore of Pavia, 1998-2003.
- Specialization School in Medical Statistics – Course in Genetic Epidemiology, 2003-2010

- Design & Analysis of Genetic-based Association Studies, June 23-27, 2008
- MolPAGE Training Program:
a) Statistical Genetics with Mendel, July 4-8, 2005
b) Statistical Analysis of Genetic and Gene Expression, March 20-24, 2006
c) Statistical Analysis of Metabonomic and Proteomic, March 26-30, 2007
d) Causal Inference, May 19-21, 2008
e) Causal Inference: State-of-the-Art, March 16-18, 2009
- 2nd level Master in Molecular and Genetic Epidemiology, 2011, 2012.
- 2nd level Master in Statistics in Medicine and in Genomics, 2013-2015
- 2nd level Master in Statistics in Medicine and in Genomics, 2014-2016
- Workshop "Scienza e Meditazione", Pavia, 9-10 Ottobre 2015
- 2nd level Master in Statistics in Medicine and in Genomics, 2015-2017
- 2nd level Master in Statistics in Medicine and in Genomics, 2018-2020

Publications (last 5 years)

International Multiple Sclerosis Genetics Consortium. Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. **Science. 2019** Sep 27;365(6460). pii: eaav7188. doi: 10.1126/science.aav7188. PubMed PMID: 31604244.

Gentilini D, Oliveri A, Fazia T, Pini A, Marelli S, Bernardinelli L, Di Blasio AM. NGS analysis in Marfan syndrome spectrum: Combination of rare and common genetic variants to improve genotype-phenotype correlation analysis. **PLoS One. 2019** Sep 19;14(9):e0222506. doi: 10.1371/journal.pone.0222506. eCollection 2019. PubMed PMID: 31536524; PubMed Central PMCID: PMC6752800.

International Multiple Sclerosis Genetics Consortium. Electronic address: chris.cotsapas@yale.edu; International Multiple Sclerosis Genetics Consortium. Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. **Cell. 2019** Jun 27;178(1):262. doi: 10.1016/j.cell.2019.06.016. PubMed PMID: 31251915; PubMed Central PMCID: PMC6602362.

International Multiple Sclerosis Genetics Consortium. A systems biology approach uncovers cell-specific gene regulatory effects of genetic associations in multiple sclerosis. Nat Commun. 2019 May 20;10(1):2236. doi: 10.1038/s41467-019-09773-y. Erratum in: **Nat Commun. 2019** Jul 1;10(1):2956. PubMedPMID: 31110181; PubMed Central PMCID: PMC6527683.

Rondanelli M, Castellazzi AM, Riva A, Allegrini P, Faliva MA, Peroni G, Naso M, Nichetti M, Tagliacarne C, Valsecchi C, Fazia T, Perna S, Graziano F, Grassi M, Bernardinelli L. Natural Killer Response and Lipo-Metabolic Profile in Adults with Low HDL-Cholesterol and Mild Hypercholesterolemia: Beneficial Effects of Artichoke Leaf Extract Supplementation. **Evid Based Complement Alternat Med. 2019** Jan 6;2019:2069701. doi: 10.1155/2019/2069701. eCollection 2019. PubMed PMID: 30723511; PubMed Central PMCID: PMC6339758.

Fazia T, Pastorino R, Notartomaso S, Busceti C, Imbriglio T, Cannella M, Gentilini D, Morani G, Ticca A, Bitti P, Berzuini C, Dalmay T, Battaglia G, Bernardinelli L. Acid sensing ion channel 2: A new potential player in the pathophysiology of multiple sclerosis. **Eur J**

Neurosci. 2018 Dec 14. doi: 10.1111/ejn.14302. [Epub ahead of print] PubMed PMID: 30549327.

International Multiple Sclerosis Genetics Consortium. Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. **Cell. 2018** Nov 29;175(6):1679-1687.e7. doi: 10.1016/j.cell.2018.09.049. Epub 2018 Oct 18. PubMed PMID: 30343897; PubMed Central PMCID: PMC6269166.

Berzuini C, Guo H, Burgess S, Bernardinelli L. A Bayesian approach to Mendelian randomization with multiple pleiotropic variants. **Biostatistics. 2018 Aug 1**. doi: 10.1093/biostatistics/kxy027. [Epub ahead of print] PubMed PMID: 30084873.

Conde S, Xu X, Guo H, Perola M, Fazia T, Bernardinelli L, Berzuini C. Mendelian randomisation analysis of clustered causal effects of body mass on cardiometabolic biomarkers. **BMC Bioinformatics. 2018** Jul 9;19(Suppl 7):195. doi: 10.1186/s12859-018-2178-2. PubMed PMID: 30066639; PubMed Central PMCID: PMC6069804.

Scribante A, Montasser MA, Radwan ES, Bernardinelli L, Alcozer R, Gandini P, Sfondrini MF. Reliability of Orthodontic Miniscrews: Bending and Maximum Load of Different Ti-6Al-4V Titanium and Stainless Steel Temporary Anchorage Devices (TADs). **Materials** (Basel). **2018** Jul 5;11(7). pii: E1138. doi: 10.3390/ma11071138. PubMed PMID: 29976856; PubMed Central PMCID: PMC6073155.

Gentilini D, Somigliana E, Pagliardini L, Rabellotti E, Garagnani P, Bernardinelli L, Papaleo E, Candiani M, Di Blasio AM, Viganò P. Multifactorial analysis of the stochastic epigenetic variability in cord blood confirmed an impact of common behavioral and environmental factors but not of in vitro conception. **Clin Epigenetics. 2018** Jun 8;10:77. doi: 10.1186/s13148-018-0510-3. eCollection 2018. PubMed PMID: 29930742; PubMed Central PMCID: PMC5994106.

Fazia T, Pastorino R, Foco L, Han L, Abney M, Beecham A, Hadjixenofontos A, Guo H, Gentilini D, Papachristou C, Bitti PP, Ticca A, Berzuini C, McCauley JL, Bernardinelli L. Investigating multiple sclerosis genetic susceptibility on the founder population of east-central Sardinia via association and linkage analysis of immune-related loci. **MULTIPLE SCLEROSIS**. 2017 Sep 1:1352458517732841. doi: 10.1177/1352458517732841. [Epub ahead of print] PubMed PMID: 28933650

Chiesa A, Fazia T, Bernardinelli L, Morandi G. Citation patterns and trends of systematic reviews about mindfulness. **COMPLEMENTARY THERAPIES IN CLINICAL PRACTICE**. 2017 Aug;28:26-37. doi: 10.1016/j.ctcp.2017.04.006. Epub 2017 Apr 28. PubMed PMID: 28779934

International Multiple Sclerosis Genetics Consortium. Electronic address: cotsapas@broadinstitute.org; International Multiple Sclerosis Genetics Consortium. NR1H3 p.Arg415Gln Is Not Associated to Multiple Sclerosis Risk. **Neuron**. 2016 Oct 19;92(2):333-335. doi: 10.1016/j.neuron.2016.09.052. PubMed PMID: 27764667; PubMed Central PMCID: PMC5641967.

Hadjixenofontos A, Gourraud Pa, Bakthavachalam V, Foco L, Ticca A, Bitti P, Pastorino R, Bernardinelli L, Mccauley Jl (2015). Enrichment for Northern European-derived multiple sclerosis risk alleles in Sardinia. **MULTIPLE SCLEROSIS**, vol. 21, p. 1396-1403, ISSN: 1352-4585, doi: 10.1177/1352458515581872

Price MP, Gong H, Parsons MG, Kundert JR, Reznikov LR, Bernardinelli L, Chaloner K, Buchanan GF, Wemmie JA, Richerson GB, Cassell MD, Welsh MJ (2014). Localization and behaviors in null mice suggest that ASIC1 and ASIC2 modulate responses to aversive

stimuli. **GENES BRAIN AND BEHAVIOR**, vol. 13, p. 174-194, ISSN: 1601-1848, doi: 10.1111/gbb.12108

International Multiple Sclerosis Genetics Consortium (IMSGC), Beecham AH, Patsopoulos NA, Xifara DK, Davis MF, Kemppinen A, Cotsapas C, Shah TS, Spencer C, Booth D, Goris A, Oturai A, Saarela J, Fontaine B, Hemmer B, Martin C, Zipp F, D'Alfonso S, Martinelli-Boneschi F, Taylor B, Harbo HF, Kockum I, Hillert J, Olsson T, Ban M, Oksenberg JR, Hintzen R, Barcellos LF, Wellcome Trust Case Control Consortium 2 (WTCCC2), International IBD Genetics Consortium (IIBDGC), Agliardi C, Alfredsson L, Alizadeh M, Anderson C, Andrews R, Søndergaard HB, Baker A, Band G, Baranzini SE, Barizzone N, Barrett J, Bellenguez C, Bergamaschi L, Bernardinelli L, Berthele A, Biberacher V, Binder TM, Blackburn H, Bomfim IL, Brambilla P, Broadley S, Brochet B, Brundin L, Buck D, Butzkueven H, Caillier SJ, Camu W, Carpentier W, Cavalla P, Celius EG, Coman I, Comi G, Corrado L, Cosemans L, Cournu-Rebeix I, Cree BA, Cusi D, Damotte V, Defer G, Delgado SR, Deloukas P, di Sapio A, Dilthey AT, Donnelly P, Dubois B, Duddy M, Edkins S, Elovaara I, Esposito F, Evangelou N, Fiddes B, Field J, Franke A, Freeman C, Frohlich IY, Galimberti D, Gieger C, Gourraud PA, Graetz C, Graham A, Grummel V, Guaschino C, Hadjixenofontos A, Hakonarson H, Halfpenny C, Hall G, Hall P, Hamsten A, Harley J, Harrower T, Hawkins C, Hellenthal G, Hillier C, Hobart J, Hoshi M, Hunt SE, Jagodic M, Jelčić I, Jochim A, Kendall B, Kermode A, Kilpatrick T, Koivisto K, Konidari I, Korn T, Kronsbein H, Langford C, Larsson M, Lathrop M, Lebrun-Frenay C, Lechner-Scott J, Lee MH, Leone MA, Leppä V, Liberatore G, Lie BA, Lill CM, Lindén M, Link J, Luessi F, Lycke J, Macciardi F, Männistö S, Manrique CP, Martin R, Martinelli V, Mason D, Mazibrada G, McCabe C, Mero IL, Mescheriakova J, Moutsianas L, Myhr KM, Nagels G, Nicholas R, Nilsson P, Piehl F, Pirinen M, Price SE, Quach H, Reunanen M, Robberecht W, Robertson NP, Rodegher M, Rog D, Salvetti M, Schnetz-Boutaud NC, Sellebjerg F, Selter RC, Schaefer C, Shaunak S, Shen L, Shields S, Siffrin V, Slee M, Sorensen PS, Sorosina M, Sospedra M, Spurkland A, Strange A, Sundqvist E, Thijs V, Thorpe J, Ticca A, Tienari P, van Duijn C, Visser EM, Vucic S, Westerlind H, Wiley JS, Wilkins A, Wilson JF, Winkelmann J, Zajicek J, Zindler E, Haines JL, Pericak-Vance MA, Ivinson AJ, Stewart G, Hafler D, Hauser SL, Compston A, McVean G, De Jager P, Sawcer SJ, McCauley JL. Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. **NATURE GENETICS**. 2013 Nov, 45(11):1353-60. doi: 10.1038/ng.2770. Epub 2013 Sep 29. PubMed PMID: 24076602, PubMed Central PMCID: PMC3832895