CURRICULUM VITAE

Nicola Pirastu

POSTS:	2016-present	Chancellor's Fellow (Assistant professor) at the Usher Institute of the University of Edinburgh, Scotland, UK.
	2009-2015	Post doctoral fellow at the University of Trieste, Italy
	2005-2009	Contract researcher Shardna spa., Italy
DEGREES:	2006-2008	PhD in animal, molecular and human biology, Thesis title: "Statistical approaches to complex diseases: Identification of genetic variants assiociated to Androgenetic Alopecia", University of Cagliari. 2005-2009
	1997-2005	Degree in Biological Sciences (5 years), 110/110 with honours, University of Cagliari, 1997-2005

Participation in research projects

"Italian Taste" – Project of the Italian Society of Sensorial Sciences with the support of the "Louis Bonduelle Foundation". Role: Responsible for the genetic section of the project.

"Friuli Venezia Giulia Genetic Park" - Regione Friuli Venezia Giulia, Legge 26/2005, Art. 23 Role: Genetic data analyst

"From Nutrigenetics to Nutriceutics: development of synergic and integrated actions for the creation of tests, diets and products to improve pubblic health and prevent common food related disorders. - Nutrition – " Role: Principal genetic and epidemiological data analyst for WP1; Organizer and manager of sampling campains.

"Cardiovascular diseases: from genetic and enviromental risk factors to prevention through innovative nutritional components" - REGIONE Friuli Venezia Giulia Role: Principal genetic and epidemiological data analyst.

"INGI: Identification of the genetic and enviromental risk factors for quantitative complex traits (with particular focus on cardiovascular diseases) through the use of isolated genetic isolates." Role: Genetic and epidemiological data analyst

"On Marco Polo's Track: genes, taste and their health implications along the silk road" Ricerca corrente IRCCS Burlo Garofolo Linea 2 35/09 Role: Genetic and epidemiological data analyst; On field phenotype acquisition.

External Consultancies

“Nutrigenomics and coffee consumption: physiological effects, taste genetics and plant genetics.” Consultant for Illy spa.

“Obesity prevention between genetics and physical exercise”. Consultant “Maniago Nuoto s.s.d. spa”.

“Genetic analysis of food liking differences in cats” – Consultant for Diana Pet Food.

Scientific Interests and Experience

Since the beginning of my scientific career in 2005 my research has been focused on understanding the genetic bases of complex traits, in particular focusing on isolated populations.
During my PhD my research has been focused on understading the genetic bases of androgenetic alopecia which led to the identification of EDA2R as being responsible for the predisposition to this disease leading to a patent application to predict male pattern baldness from genetic variants. I was also involved in numerous other projects both on the first genome-wide association studies and linkage studies in large extended pedigrees.
After moving to Trieste in 2009, apart handling numerous international collaborations within genetic association study consortia, my research has been focused on understanding the genetic bases of food choices looking with a particular angle on food liking as a marker of long-term consumption and to effectively change people’s dietary behaviour. This has led to several important studies linking food liking to taste receptors and to the first large scale GWAS on food liking.

During this period, I have also managed several research projects focused on studying the role of genetics on complex traits in the isolated populations of Friuli Venezia Giulia in the north of Italy and part of the Italian Network of Genetic isolates (INGI). I was responsible for analysing the genetic data, organising structure of the data, organising sampling campaigns, the field logistics and carrying out part of the data collection.

In 2016 I became Chancellor’s fellow at the Usher Institute of the Univesity of Edinburgh where I continued my research on food liking genetics with the idea of understanding the biology behind food choices and to devise better interventions or food products. Also, I aim at exploiting the genetic variants associated with food choices to improve our understanding of the consequences of food on health.

This has led my work on two main parallel lines; the first was to create the largest collection of food liking data ever known. For this I first created a standardised food liking questionnaire and then worked to have it administered on cohorts where genetic data was already available. This work was extremely successful, and I was able in collaboration with the participating cohorts to collect close to 200,000 samples mostly available through UK Biobank. By combining GWAS and Genomic SEM we have created a multi-layered comprehensive map of food preferences highilighting two main and independent food liking dimensions: one which includes easy palate caloric dense foods while the other all the foods that have an acquired taste. We have also identified more than 1400 SNP-trait association within 171 loci giving new insight in the biology of several known loci while identifying several new ones. This work has recently been submitted to bioarchives as a pre-print and it is currently under review at Nature Comunications.

The second was focused on improving the statistical methods used to study food consumption measured through questionnaires, looking at Food Frequency Questionnaires in UK Biobank. I have thus conducted a large GWAS on food consumption looking at single foods and foods patterns identifying several novel loci. I have also developed a new statistical method which removes the very well-known biases present in nutritional epidemiology data by exploting Mendellian Randomization to distinguish between the loci directly influencing food consumption from those which are mediated through other traits. The selected variants were then used for Mendellian randomization of food and food patterns on health-related traits including blood metabolites confirming several known relationships while identifying several new ones. I am at the moment working on expanding this work by creating an international consortium to expand the number of analysed food items, include ancestries different from European and increase the number of identified loci.

Apart from these two main lines of research I have other ongoing parallel projects, for example focused on creating a method to objectively measures food consumption starting from blood metabolites or on creating polygenic scores predictive of biological pathway functionality in a tissue specific manner.

Through out my career I have organised and carried out several sampling campaings: 2008 in a shopping mall in Cagliari to study male pattern baldness, 2010-2012 Marcopolo project sample collection in central Asian countries, 2014 resampling campaign within the Friuli Venezia Giulia Genetic Isolates.

I have also developed several methods and statistical approaches: R package for gene-based test, methods for case control studies in isolated populations and application to Random Forest, separation of genetic direct effects from mediated ones both within a multivariable mendellian randomization framework or using GenomicSEM for latent variable methods, correction for sample selection bias in genome-wide association studies.

Technical Competences

Parametric and non-parametric linkage analysis in extended pedigrees using software Simwalk, Merlin.
Genome-wide association analysis using a wide variety of softwares such as GenABEL, Plink, fastGWAS.
R scripting and package development. Shiny app scripting and development: https://npirastu.shinyapps.io/Food_MR/, https://npirastu.shinyapps.io/rg_plotter_2/.
Unix, awk scripting. Pipeline development.
Statistical methods development: case control association within isolated populations; Gene-wide association analysis; direct vs indirect snp effect measurement; conditioned analysis within genetic structural equation modellin.

Active ongoing scientific collaborations

Prof Christina Dahm and Dr Daniel Borch Ibsen: “The genetics of compliance to diet”

Prof. James Wilson, Prof Nick Timposn, Prof Eco de Geuss, Dr. Cristina Menni, Dr. Maria Veldhuinzen, Dr.Nana Matoba: “The genetic determinants of food liking”

Prof. James Wilson, Prof Eco de Geuss, DR. Yann Kilimentidis, Dr. Cristina Menni: “The genetic determinants of liking of physical activity”

Prof. John Perry, Dr. Felix Day: “Estimating the genetic bases of generalised food liking and its biology”

Dr. Maria Veldhuinzen: “Imputing sensory brain network functionality from resting phase fMRI data in order to study its genetics in large datasets”

SCALLOP Consortium: “Using eQTL and pQTL datas to create pathways specific polygenic risk scores”

Prof Monica Laureati, Prof Nick Timpson, Dr Kathiln Wade “Measuring food liking development in the CoCo90 cohort”

Oral and invited communications at international meetings:

“Unraveling the genetics of complex diseases through random forest: application to genome wide Association of asthma in a genetic isolate of Ogliastra”. 4th International Meeting on Genetics of complex diseases and isolated populations. Trieste 20-23 June 2009.

“A genome-wide association study identifies new loci that underlie food preferences in Italian isolated populations.” European Chemoreception Research Organization XXI Congress, 7-10 September 2011, Manchester, UK.

“Genetics of taste and food preferences” V International meeting on complex traits and genetic isolates Trieste 15-16 March 2012.

“Isolated populations give new insight on food preferences genetics.” 10th Pangborn Sensory Science Symposium 11-15 August 2013 Rio de Janeiro, Brazil

“The Italian network of genetic isolates: a resource to establish a personalized medicine framework in Italy” XVI meeting of the Italian Society of Human Genetics 25-28 September 2013 Rome

“Food likings and isolated populations give new insight on the genetics of food choice.” 4th International Research Conference “Postgenomic methods of analysis in biology, laboratory and clinical medicine” 28 October – 1 November 2014 Kazan, Russian federation

“Food preferences genetics: toward a new approach to understand the relationship between human nutrition and health.” GIGA day 27 January 2015 Liège Belgium

“Novel associations for male pattern baldness provide new insights into aetiology and genetic correlations” Platform, American Society of Human Genetics 66th Annual Meeting October 18–22, 2016 Vancouver, Canada

“Food preferences genetics: toward a new approach to understanding nutrition.”Edinburgh Alliance for Complex Trait Genetics 11th meeting – Friday 28th October, 2016

“The genetics of food preferences” Congresso Associazione Genetica Italiana 7-9 Settembre 2017 Cortona (Arezzo)

“The exposural landscape of Coronary Artery Disease gives new insight in its aetiology and missing heritability.” 46th European Mathematical Genetics Meeting, Cagliari, Italy 18th to 20th April 2018

"The genetics of food and drink consumption and correlation with health-related traits” EuroSense 2018 2-5 September 2018, Verona, Italy.

“Using genetics to disentangling the complex relationship between food choices and health status” European Mathematical Genetics Meeting, Lausanne, Switzerland 16th-17th April 2020

“Genome-wide association study of food liking in 162,000 people uncovers the genetic bases of food liking.” 14th Pangborn Sensory Science Symposium August 9-12, 2021

“Biases in GWAS and where to find them: detecting and accounting for biases in GWAS studies beyond population structure.” European Human Genetics Conference Virtual Conference August 28–31, 2021

“Genome-wide association study of food liking in 162,000 people uncovers the genetic bases of food liking.” ECRO 2021 Conference, Cascais Portugal, 13-16th September 2021

PhD Student Supervision:

First supervisor Ciara McDonnel (4th year) project title:” Functional characterization of genetic variants influencing human food preferences using bioinformatics and animal models”

First supervisor Eryk Grzeszkowiak (3rd year) project title “Understanding the role of genetics in human food behaviour”

First supervisor Sebastian May-Wilson (4th year) project title: “”Using integrative omics to disentangle causal relationships between tissue-specific pathways and coronary arterydisease”

Co-Supervisor Marisa Mukian (3rd year): “”Tackling fundamental questions in viral epidemiology and vaccination using genomics and data science”

Co-Supervisor Nele Taba (3rd year) from the University of Tartu: ”Dietary patterns, their genetic and socio-demographic background, effects on metabolic profile and health indicators in the Estonian Biobank cohort"

Committee and society memberships.

Member of ASHG and ESHG
Member of the Institute of Food and Science Tecnology (IFST)
Member of the SSG committee of the IFST
Member of the GCCR consortium
Coordinator of activities of the Molecular Epidemiology group within the Usher Institute
GWAS catalog file format committee

Teaching experiences

Lecturer: “PhD course on Nutritional Epidemiology” Copenhagen 28-30/9 2021
Lecturer: Genetic epidemiology course “Master of Pubblic Health” Edinburgh University (years 2017-2020)
Lecturer: Summer School "La genomica al servizio della Nutrizione" (Udine, 3-4 luglio 2013).

Pubblication list

Total citations: 5996
H-Index: 29
First/Last Author publications: 16 (Name highlighted in bold)

69: Yang et al "Genetic landscape of the ACE2 coronavirus receptor" 2021 Under review Circulation.

68: Kilimentidis Y…. **Pirastu N** "Genome-wide association study of liking of physical activity in the UK Biobank" Submitted Nature Human Behaviour 2021.

67: **Pirastu N.** et al "Variation in the TAS2R38 Taste Receptor Gene and Outcomes Following COVID-19 Infections" Accepted Jama network open October 2021.

66: Rannikmae et al. "Physician-confirmed and administrative definitions of stroke in UK Biobank reflect the same underlying genetic trait" medRxiv 2021 doi: https://doi.org/10.1101/2021.09.30.21264348

65: May-Wilson S., …, **Pirastu N**"Large-scale genome-wide association study of food liking reveals genetic determinants and genetic correlations with distinct neurophysiological traits" bioRxiv 2021 **doi:** https://doi.org/10.1101/2021.07.28.454120

64: Klaric et al. "Mendelian randomisation identifies alternative splicing of the FAS death receptor as a mediator of severe COVID-19" medRxiv 2021 doi: https://doi.org/10.1101/2021.04.01.21254789

63: Taba N. … **Pirastu N** "Mendelian randomization identifies the potential causal impact of dietary patterns on circulating blood metabolites" bioRxiv 2020.10.09.332924 Accepted Frontiers Genetics October 2021

63: Gerkin et al "Recent smell loss is the best predictor of COVID-19: a preregistered, cross-sectional study" medRxiv. 2020 doi: 10.1101/2020.07.22.20157263

62: Parma et al "More Than Smell-COVID-19 Is Associated With Severe Impairment of Smell, Taste, and Chemesthesis" Chem Senses. 2020

61: **Pirastu N et al**. "Genetic analyses identify widespread sex-differential participation bias" Nat Genet 53**,** 663–671 (2021)

60: **Pirastu N et al**. "Using genetics to disentangle the complex relationship between food choices and health status" bioRxiv 829952, Under review PLOS Genetics.

59: Kennedy et al. "Coffee Consumption and Kidney Function: A Mendelian Randomization Study" Am J Kidney Dis. 2019

58: Clark et al. "Associations of autozygosity with a broad range of human phenotypes." Nat Commun. 2019

57: Tin et al. "Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels." Nat Genet. 2019

56: Wuttke et al. “A catalog of genetic loci associated with kidney function from analyses of a million individuals.” Nat Genet. 2019

55: Karlsson Linnér et al. “Genome-wide association analyses of risk tolerance and risky behaviors in over 1 million individuals identify hundreds of loci and shared genetic influences.” Nat Genet. 2019

54: Bevilacqua et al. “Genome-wide association study identifies an association between variants in EFCAB4B gene and periodontal disease in an Italian isolated population.” J. Periodontal Res. 2018

53: Lee at al: “Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals.” Nat Genet. 2018 Jul 23

52: **Pirastu N et al.** “Reply to 'Misestimation of heritability and prediction accuracy of male-pattern baldness'.” Nat Commun. 2018 Jun 29

51: **Pirastu N. et al**. “GWAS for male-pattern baldness
identifies 71 susceptibility loci explaining 38% of the risk.” Nat Commun. 2017 Nov 17

50: Joshi PK, **Pirastu N** et al. “Genome-wide meta-analysis associates
HLA-DQA1/DRB1 and LPA and lifestyle factors with human longevity. Nat Commun. 2017 Oct 13

49: Zupin L. et al. “LTF and DEFB1 polymorphisms are associated with susceptibility toward chronic periodontitis development.” Oral Dis. 2017 Oct.

48: Monteleone E at al. “Exploring influences on food choice in a large population sample: The Italian Taste project” Food Quality and Preference July 2017

47: Schumann G et al. “KLB is associated with alcohol drinking, and its gene product β-Klotho is necessary for FGF21 regulation of alcohol preference.” Proc Natl Acad Sci U S A. 2016 Dec 13

46: Navarra CO et al. “Caries and Innate Immunity: DEFB1 Gene Polymorphisms and Caries Susceptibility in Genetic Isolates from North-Eastern Italy.” Caries Res.2016

45: Marioni RE et al. “Genetic variants linked to education predict
longevity.” Proc Natl Acad Sci U S A. 2016 Nov 22

44: **Pirastu N. et al**. “Non-additive genome-wide association scan reveals a new gene associated with habitual coffee consumption.” Sci Rep. 2016 Aug

43: McCarthy S. et al. “A reference panel of 64,976 haplotypes
for genotype imputation.” Nat Genet. 2016 Oct

42:Masi et al. “Caffeine metabolism rate influences coffee perception, preferences and intake” Food Quality and Preference Oct 2016

41: Okbay, A. et al. “Genome-wide association study identifies 74 loci associated with educational attainment” Nature. 2016 May

40: **Pirastu N et al**. “A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings”. Rev Endocr Metab Disord. 2016 Apr 30.

39: Robino A et al. “Understanding the role of personality and alexithymia in food preferences and PROP taste perception.” Physiol Behav. 2016 Apr 1;157:72-8.

38: Day FR et al. “Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair”. Nat Genet. 2015 Nov;47(11):1294-303.

37: Palister T et al. “Food Preference Patterns in a UK Twin Cohort”. Twin Research and Human Genetics 2015 Sep 28:1-13.

36: Robino A, et al. “Polymorphisms in sweet taste genes (TAS1R2 and GLUT2), sweet liking, and dental caries prevalence in an adult Italian population”. Genes Nutr. 2015 Sep;10(5):485.

35: Martin HC et al. “Multicohort analysis of the maternal age effect on recombination.” Nat Commun. 2015 Aug 5;6:7846.

34: **Pirastu N**, Robino A “Uncovering the genetic basis for food preferences: the key to personalized nutrition plans?” Personalized Medicine 2015 12:4:315-317

33: Joshi PK et al. “Directional dominance on stature and cognition in diverse human populations.” Nature. 2015 Jul 1

32: Huffman JE et al. “Modulation of genetic associations with serum urate levels by body-mass-index in humans.” PLoS One. 2015 Mar 26;10(3):e0119752.

31. **Pirastu N et al**. “Genome wide association analysis on 5 isolated populations identifies variants of the HLA-DOA gene associated with white wine liking.” Eur J Hum Genet. 2015 Mar 11.

30. Mezzavilla M et al. “Genetic landscape of populations along the Silk Road: admixture and migration patterns.” BMC Genet. 2014 Dec 5;15(1):131. [

29. Keller MF et al. “Trans-ethnic meta-analysis of white blood cell phenotypes.” Hum Mol Genet. 2014 Dec 20;23(25):6944-60.

28. Lisa E. Wolber et al. “Salt-inducible kinase 3, SIK3, is a new gene associated with hearing.” Hum Mol Genet. 2014 Jul 24.

27. O'Connell J et al “A General Approach for Haplotype Phasing across the Full Spectrum of Relatedness.” PLoS Genet. 2014 Apr 17;10(4):e1004234.

26. **Pirastu N et al**. “Association Analysis of Bitter Receptor Genes in Five Isolated Populations Identifies a Significant Correlation between TAS2R43 Variants and Coffee Liking.” PLoS One. 2014 Mar 19;9(3):e92065.

25. Robino A et al. “A Population-Based Approach to Study the Impact of PROP Perception on Food Liking in Populations along the Silk Road.” PLoS One. 2014 Mar 13;9(3):e91716.

24. Medici M et al. “Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease.” PLoS Genet. 2014 Feb 27;10(2):e1004123.

23. Moura R et al. “Exome analysis of HIV patients submitted to dendritic cells therapeutic vaccine reveals an association of CNOT1 gene with response to the treatment.” J Int AIDS Soc. 2014 Jan 10;17(1):18938.

22. O'Seaghdha CM et al. “Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations.” PLoS Genet. 2013;9(9):e1003796.

21. Pistis G et al. “Genome wide association analysis of a founder population identified TAF3 as a gene for MCHC in humans.” PLoS One. 2013 Jul 31;8(7):e69206.

20. Paul DS et al “Maps of open chromatin highlight cell type-restricted patterns of regulatory sequence variation at hematological trait loci.” Genome Res. 2013 Jul;23(7):1130-41.

19. Nolan LS et al. “Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice.” Neurobiol Aging. 2013 Aug;34(8):2077.e1-9.

18. Biino G et al. ”Age- and sex-related variations in platelet count in Italy: a proposal of reference ranges based on 40987 subjects' data.” PLoS One. 2013;8(1):e54289.

17. Köttgen A et al. “Genome-wide association analyses identify 18 new loci associated with serum urate concentrations.” Nat Genet. 2012 Dec 23

16. van der Harst P et al. “Seventy-five genetic loci influencing the human red blood cell.” Nature. 2012 Dec 20;492(7429):369-75.

15. Nürnberg ST et al. “A GWAS sequence variant for platelet volume marks an alternative DNM3 promoter in megakaryocytes near a MEIS1 binding site. Blood.” 2012 Dec 6;120(24):4859-68.

14. **Pirastu N et al**. “Genetics of Food Preferences: A First View from Silk Road Populations.” J Food Sci. 2012 Dec;77(12):S413-S418.

13. Boraska V et al “Genome-wide meta-analysis of common variant differences between men and women.” Hum Mol Genet. 2012 Nov 1;21(21):4805-15.

12. Okada Y et al “Meta-analysis identifies multiple loci associated with kidney function-related traits in east Asian populations.” Nat Genet. 2012 Jul 15;44(8):904-9

11. McQuillan R et al. ”Evidence of inbreeding depression on human height.” PLoS Genet. 2012;8(7):e1002655.

10. Gieger C et al. “New gene functions in megakaryopoiesis and platelet formation.” Nature. 2011 Nov 30;480(7376):201-8.

9. Girotto, G. et al. “Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road.” Audiological Medicine, 9(4), 135-140. Informa Scandinavian.

8. Wain LV et al “Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure.” Nat Genet. 2011 Sep 11;43(10):1005-11.

7. Cabras S…. **Pirastu N** “A strategy analysis for genetic association studies with known inbreeding.” BMC Genet. 2011 Jul 18;12:63.

6.Nalls MA et al “Multiple loci are associated with white blood cell phenotypes.” PLoS Genet. 2011 Jun;7(6):e1002113.

5. F. Faletra, et al “A novel mutation in the vWFA2 domain of the COCH gene in an Italian DFNA9 family,” Audiological Medicine, vol. 9, no. 1, pp. 4-7, Nov. 2010.

5. Girotto G, **Pirastu N** et al. “Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways.“ J Med Genet. 2011 Jun;48(6):369-74.

4. Mocci E et al. “Microsatellites and SNPs linkage analysis in a Sardinian genetic isolate confirms several essential hypertension loci previously identified in different populations.” BMC Med Genet. 2009 Aug 28;10:81.

3. Pistis G et al. “High differentiation among eight villages in a secluded area of Sardinia revealed by genome-wide high density SNPs analysis.” PLoS One. 2009;4(2):e4654.

2. Prodi DA et al. “EDA2R is associated with androgenetic alopecia.” J Invest Dermatol. 2008 Sep;128(9):2268-70. Epub 2008 Apr 3.

1. Angius A et al. “Patterns of linkage disequilibrium between SNPs in a Sardinian population isolate and the selection of markers for association studies.” Hum Hered. 2008;65(1):9-22. Epub 2007 Jul 25.

Dichiarazione sostitutiva di certificazioni

(Art.46 del D.P.R. 28 dicembre 2000, n. 445)

Dichiarazione sostitutiva dell'atto di notorietà

(Art. 47 del D.P.R. 28 dicembre 2000, n. 445)

Il/La sottoscritto Nicola Pirastu nato a Cagliari il 04/10/1978 , residente in Edimburgo Scozia Regno Unito, a conoscenza di quanto prescritto dall'art. 76 del D.P.R. 28 dicembre 2000, n. 445, sulla responsabilità penale cui può andare incontro in caso di falsità in atti e di dichiarazioni mendaci, ai sensi e per gli effetti del citato D.P.R. n. 445/2000 e sotto la propria personale responsabilità:

D I C H I A R A

che tutto quanto affermato e riportato nel curriculum corrisponde al vero.

Letto, confermato e sottoscritto. IL/LA DICHIARANTE

Edimburgo, li 14/10/2019. Nicola Pirastu