

Curriculum Vitae - Andrea Angius

Name Surname: *Andrea Angius*

Degree: *Biological Sciences*

Nationality: *Italian*

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Current Position (February 2021)

Senior Researcher

Department/Institute/Institution: *Department of Biomedicine - Institute of Biomedical and Genetic Research - National Research Council (CNR)* Street Address: *Cittadella Universitaria di Monserrato, S.S. 554 bivio per Sestu Km 4,500* Zip code City: *09042 Monserrato (CA)*

Province/Country: *Cagliari, Italy*

Adjunct Associate Professor at Dep. of Veterinary Medicine

Department/University: *Dep. of Biomedical Sciences and Dep. of Veterinary Medicine – University of Sassari* Street Address: *V.le San Pietro 43/C* Zip code City: *07100 Sassari (SS)*

Province/Country: *Sassari, Italy*

Research areas

Genetics, Medical Genetics, Molecular Diagnostics, Molecular Biology, Molecular Genetics, Population Genetics, Population Biology, Evolutionary Developmental Biology, Evolutionary Genetics, Cancer Biology, Biomarkers, Human Cancer, Biotechnology, Plant Molecular Biology and Genetics

Education and training

1992 - University of Cagliari, Italy: *Master Degree in Biology*

1999 - University of Sassari, Italy: *Ph.D.*

Positions and Employments

2021 – up to present – *Permanent position as Senior Researcher National Research Council (CNR), Institute of Biomedical and Genetic Research, Monserrato, Cagliari (Italy).*

2011 – 2020 – *Permanent position as Researcher National Research Council (CNR), Institute of Biomedical and Genetic Research, Monserrato, Cagliari (Italy).*

2013-to present: *Adjunct Associate Professor in Biotechnologies at Faculty of Veterinary Medicine, University of Sassari, Sassari (Italy).*

2010-2019: *Adjunct Associate Professor in Medical Genetics at Department of Biomedical Science University of Sassari, Sassari (Italy).*

2001 – 2011 – *Researcher National Research Council (CNR), Institute of Population Genetics, Alghero, Sassari (Italy).*

2010 – 2014 – *Head and Scientific Coordinator of the Next Generation Sequencing Core, CRS4, Pula (Cagliari) (Italy).*

2006 – 2010 – *Head and Scientific Coordinator of the Genotyping Laboratory, Sardegna Ricerche “Polaris” Scientific and Technological Park of Sardinia, Pula (Cagliari) (Italy).*

2002 – 2006 – *Lab Manager of the Genotyping laboratory, Soc. SHARDNA Life Sciences, Cagliari, (Italy).*

2000-2001 - *Fixed-term Researcher at the Institute of Molecular Genetics, of the National Council of Research (CNR), Alghero (Italy).*

1996 – 1999 - *Ph. D. Faculty of Biology, University of Sassari, Sassari (Italy).*

1995-1996 - *CNR fellowship. Institute of Research of Mediterranean Anemia and Thalassemia, National Research Council (CNR), Director: Antonio Cao, MD; Cagliari (Italy).*

1992-1994 - *Post-doctoral fellowship. Laboratory of Molecular Pathology, Ospedale regionale per le Microcitemie, Director: Antonio Cao, MD; Cagliari (Italy).*

1990-1992 - *Undergraduate student. Department of Experimental Biology, University of Cagliari, Cagliari (Italy). Supervisor: G. Floris, Ph.D.*

Teaching

2017 – 2026 *Qualified as Associate Professor in Medical Genetics for Italian Universities*

(Abilitazione Scientifica Nazionale alle funzioni di professore di II fascia nel settore concorsuale 06/A1 ai sensi del DD n. 222/2012)

2014 – 2023 *Qualified as Associate Professor in Genetics and Microbiology for Italian Universities*

(Abilitazione Scientifica Nazionale alle funzioni di professore di II fascia nel settore concorsuale 05/I1 ai sensi del DD n. 222/2012)

Academic courses

2013-14 up to present (7 courses/academic years) – Course “*Genotyping Technics and Genetic Expression*”, Degree in Medical and Veterinary Biotechnologies, Faculty of Veterinary, University of Sassari (Italy).

2010-11 to 2019 (9 courses/academic years) – Course “*Medical Genetics*”, Degree in Exp. and Applied Biology, Faculty of Math., Phys. and Nat. Science, University of Sassari (Italy).

2010-11 to 2012-13 (3 courses/academic years) - Course “*Genotyping Technics*”, Degree in Medical and Veterinary Biotechnologies, Faculty of Veterinary, University of Sassari (Italy).

2006-07 to 2010-11 (5 courses/academic years) – Course “*Human Genetics*”, Degree in Biological Sciences, Faculty of Math., Phys. and Nat. Science, University of Sassari (Italy).

2003-2004 (1 course/academic year) – Course “*Basic Element of Genetics and laboratory*”, Degree in Biological Sciences, Faculty of Mathematical, Physical and Natural Science, University of Sassari (Italy).

Masters courses

2012-13 /2013-14 (2 courses/academic years) - Course “*Genetics and Technics of Genetic Characterization*”, International Master in Medical Biotechnology, Hue University of Medicine and Pharmacy, (Vietnam)

2007- Master in R&S in Medical Biotechnology, Ed. 2007, Sardegna Ricerche “*Polaris*” Scientific and Technological Sardinia Park, Pula (CA) (Italy)

Membership of the college of PhD teachers recognized by MIUR

Board of Lecturers of the PhD Course in Biomedical Sciences in Medical Genetics, Metabolic Diseases and Nutrigenomics, certified by MIUR, University of Sassari from XXIX cycle (a.a. 2013-2014) to today.

Board of Lecturers of the PhD Course in Biomolecular and Biotechnological Sciences, certified by MIUR, University of Sassari from the XXVI cycle (a.a. 2010/11) to XXIII (a.a. 2012/13).

Grants and participation in national and international research projects

Dr. Angius has been involved in the drafting and management of the project both as a responsible and collaborator in various regional, national and international research projects eligible for funding on the basis of competitive calls for proposals, as described in detail in the list below.

1-years research grant funded by Fondazione Banco di Sardegna ROL 2019 Bando “*Salute pubblica, medicina preventiva e riabilitativa*” for the project entitled “*Sviluppo di un pannello di marcatori immunoistochimici ed epigenetici a scopo predittivo nella terapia del carcinoma mammario “Triplo Negativo”*” GRANT_NUMBER: 33141 – Role: Principal Investigator

2-years research grant funded by Fondo di Beneficenza, Intesa Sanpaolo S.p.A. ”(PREDICT) PREcision meDicine In ColorecTal cancer: new clinical-genomic network for expanding tailored oncologic care” GRANT_NUMBER: B/2020/0094 – Role: Principal Investigator

2-years research grant funded by Fondazione Banco di Sardegna ROL 2019 Bando “*Salute pubblica, medicina preventiva e riabilitativa*” for the project entitled “*Prevenzione e diagnosi precoce del carcinoma del colon-retto: sviluppo di un panel di marcatori molecolari da biopsia liquida*” –Role: Coordinator of NGS data generation and analysis

2-years research grant funded by Bando competitivo Fondazione di Sardegna – 2017 per progetti di ricerca con revisione tra pari” for the project entitled “*Personalized Care in Colorectal Cancer: building a new clinical-genomic network for tailoring genotype and phenotype*” –Role: Coordinator of NGS data generation and analysis

4-years research grant funded by MIUR for the project entitled “*PATH - Pathology in Automated Traceable Healthcare*” PON04a2_00557 - Decreto n. 428 del 27/02/2017 Role: Key Personell

5-years research grant funded by European Framework Programme for Research and Innovation HORIZON 2020, “*An integrated approach to dissect determinants, risk factors and pathways of ageing*”

of the immune system". European Community Grant –Role: Coordinator of NGS data generation and analysis

2-years research grant funded by Telethon for the project entitled " Post GWAS functional characterization of BCL11A locus toward the development of a treatment for β -thalassemia". Telethon Grant 2013 GGP13246 – Role: Unit Responsible

1-years research grant funded by Telethon for the project entitled "Identification of novel gene(s) associated with Crisponi/Cold Induced Sweating syndrome-like phenotypes by exome sequencing". Telethon Grant 2013 GEP13093 - Role: Coordinator of NGS data generation and analysis

2-year research grant funded by Autonomous Region of Sardinia entitled "Unravelling the genetic causes of syndromic Intellectual Disability in the era of exome sequencing". - Role: Coordinator of NGS data generation and analysis

6-years research grant funded by Ministero dell'Istruzione, dell'Università e della Ricerca. FIRB Laboratori 2003: for the project entitled "Identificazione di geni-malattia mediante genotipizzazione ad alta densità di popolazioni". – Role: Unit Responsible

5-years research grant funded by Ministero dell'Istruzione, dell'Università e della Ricerca. for the project entitled "Identificazione di fattori genetici associati a malattie multifattoriali comuni tramite un originale approccio allo studio di isolati genetici" (Art. 5 del D.M. 593 8 agosto 2000) - Role: Collaborator

3 years research grant funded by Telethon Fondazione Onlus for the project entitled "Sardinian Isolated Population For The Study of Complex Traits". Telethon Grant E 1185 – Role: Collaborator

2 years research grant funded by Regione Autonoma della Sardegna. for the project entitled "Studio della componente genetica e nutrizionale nell'insorgenza di malattie complesse" POR Sardegna 2000-2006, deliberazione Giunta Regionale della Sardegna n.27/30 del 7/08/2001. – Role: Collaborator

2 years research grant funded by Regione Autonoma della Sardegna for the project entitled "Identificazione di geni associati a malattie multifattoriali con alto impatto socio-economico: Ipertensione arteriosa essenziale e Alopecia androgenetica". Determinazione n. 107 del 22/06/2005. – Role: Collaborator

1,5 years research grant funded by Regione Autonoma della Sardegna for the project entitled "Metodologie computazionali ibride per l'analisi statistica del flusso genico in grandi Dataset".PON "Ricerca, Sviluppo Tecnologico ed Alta Formazione" 2000-2006 per le Regioni dell' obiettivo 1 Decreto 192/Ric del 04/12/2007 – Role: Collaborator

2 years research grant funded by Regione Autonoma della Sardegna for the project entitled "Infrastruttura bioinformatica per un approccio "system biology" alle malattie complesse". Programma di R&S "Tecnologie bio-informatiche applicate alla medicina personalizzata" Bando di selezione N. 364.173 A – Role: Collaborator

Events organized

Dr. Angius organized the following courses:

September 16–20, 2019 - 8th Sardinian International Summer School "From Genomic discoveries to therapeutic targets" Pula, Cagliari, <http://www.irgb.cnr.it>

July 9–13, 2018 - 7° Sardinian International Summer School "From genome-wide association studies (GWAS) to Function" Pula, Cagliari, <http://www.irgb.cnr.it>

June 12–16, 2017 - 6° Sardinian International Summer School "From genome-wide association studies (GWAS) to Function" Pula, Cagliari, <http://www.irgb.cnr.it>

June 20–24, 2016 - 5° Sardinian International Summer School "From genome-wide association studies (GWAS) to Function" Pula, Cagliari, <http://www.irgb.cnr.it>

June 22–26, 2015, - 4° Sardinian International Summer School "From genome-wide association studies (GWAS) to Function" Pula, Cagliari, <http://www.irgb.cnr.it>

Non-Institutional Scientific activities (editorial and reviewer activity)

Topic Editor for International Journal of Molecular Sciences Impact Factor 4.556 (2020 JCR)

Editor For BMC Genomics Impact Factor 3.530 (2020 JCR)

Scientific reviewer for the following journals: Journal of Medical Genetics, Journal of Endocrinological Investigation, Biological Conservation, American Journal of Human Genetics, BMC Genomics, International Journal of Genomics, BMC Medical Genetics, Case Report in Genetics, PloS ONE Journal, Nutrition, Expert Opinion on Orphan Drugs, Metabolism & Cardiovascular Diseases, Gene, Chemical Senses, Journal of Biotechnology, IOVS, Clinical Genetics, Annals of Translational

Awards and Honors

Cossu S, Angius A, Oppo M, Onano S, Persico I, Uva P, Cuccuru G, Asunis M, Pruna D, Crisponi L. Fenotipo epilettico in due fratelli con mutazioni nel gene NALCN. 40° Congresso Nazionale Lega Italiana Contro l'Epilessia (LICE), Roma, Italy, June 7-9 2017 (Prize for the best scientific contribution)

Pira G, Angius A, Uva P, Cossu Rocca P, Sanges F, Loi F, Ena S, Murgia L, Carru C, Muroi MR, DeMiglio MR. MiRNA-135b contributes to triple negative breast cancer molecular heterogeneity: different expression profile in basal-like vs non basal-like. 48° Congresso Nazionale Società Italiana di Biochimica Clinica e Biologia Molecolare Clinica (SIBioC). Torino 18-20 ottobre 2016 (Prize Best Poster SIBioC)

Concas A, Cusano R, Orsini M, Costelli C, Cao A, Angius A. Microalgae based technology for biofuels production and CO2 capture: the role of mathematical modeling and genetic engineering. Accademia Nazionale dei Lincei – Fondazione ENI Enrico Mattei, XXXI Giornata dell'ambiente. Convegno Internazionale: The TeraWatt challenge: What research for our future energy? 5 - 6 Novembre 2013 (Prize Best Poster)

List of publications

1. **Angius A**, Scanu MS, Arru C, Muroi MR, Rallo V, Deiana G, Ninniri MC, Carru C, Porcu A, Pira G, Uva P, Cossu-Rocca P, De Miglio MR. Portrait of cancer stem cells on colorectal cancer: molecular biomarkers, signaling pathways and miRNAome. *Int J Mol Sci.* 2021 Feb 5;22(4):1603. doi: 10.3390/ijms22041603.
2. Baragetti A, Severgnini M, Olmastroni E, Conca Dioguardi C, Mattavelli E, **Angius A**, Rotta L, Cibella J, Consolandi C, Grigore L, Pellegatta F, Giavarini F, Caruso D, Norata GD, Catapano AL, Peano C. Gut Microbiota Functional Dysbiosis Relates to Individual Diet in Subclinical Carotid Atherosclerosis. *Nutrients* 2021, 13, 304. <https://doi.org/10.3390/nu13020304>
3. **Angius A**, Cossu-Rocca P, Arru C, Muroi MR, Rallo V, Carru C, Uva P, Pira G, Orr $\sqrt{\pi}$ S, De Miglio MR. Modulatory Role of microRNAs in Triple Negative Breast Cancer with Basal-Like Phenotype. *Cancers (Basel)*. 2020 Nov 7;12(11):3298. doi: 10.3390/cancers12113298
4. Pira G, Uva P; Scanu A, Cossu Rocca P, Murgia L, Uleri E, Piu C, Porcu A, Carru C, Manca A, Ivana Persico I, Muroi MR, Sanges F, Dolei A, Serra C, **Angius A**, De Miglio MR. Landscape of transcriptome variations uncovering known and novel driver events in colorectal carcinoma. *Sci Rep.* 2020 Jan 16;10(1):432. doi: 10.1038/s41598-019-57311-z.
5. Serra R, Floris M, Pinna A, Boscia F, Cucca F, **Angius A**. Novel mutations in c2orf71 causing an early onset form of cone-rod dystrophy: A molecular diagnosis after 20 years of clinical follow-up. *Mol Vis.* 2019; 25:814-820
6. Balzano F, Campesi I, Cruciani S, Garroni G, Bellu E, Dei Giudici S, **Angius A**, Oggiano A, Rallo V, Capobianco G, Dessole S, Ventura C, Montella A, Maioli M. Epigenetics, stem cells and autophagy: exploring a path involving miRNA. *Int J Mol Sci.* 2019 Oct 14;20(20):5091. doi: 10.3390/ijms20205091.
7. **Angius A**, Pira G, Scanu A, Uva P, Sotgiu G, Saderi L, Manca A, Serra C, Uleri E, Piu C, Caocci M, Ibba G, Zinellu A, Cesaraccio MR, Sanges F, Muroi MR, Dolei A, Cossu Rocca P, De Miglio MR. MicroRNA-425-5p Expression affects BRAF/RAS/MAPK pathways in colorectal cancers. *Int. J. Med. Sci.* 2019; 16(11): 1480-1491. doi: 10.7150/ijms.35269
8. **Angius A**, Uva P, Pira G, Muroi MR, Sotgiu G, Saderi L, Uleri E, Caocci M, Ibba G, Cesaraccio MR, Serra C, Carru C, Manca A, Sanges F, Porcu A, Dolei A, Scanu AM, Cossu-Rocca P, De Miglio MR. Integrated analysis of miRNA and mRNA endorses a twenty miRNAs signature for colorectal carcinoma. *Int J Mol Sci.* 2019 Aug 20;20(16). pii: E4067. doi: 10.3390/ijms20164067.
9. Raveane A, Aneli S, Montinaro F, Athanasiadis G, Barlera S, Birolo G, Boncoraglio G, Di Blasio AM, Di Gaetano C, Pagani L, Parolo S, Paschou P, Piazza A, Stamatoyannopoulos G, **Angius A**, Brucato N, Cucca F, Hellenthal G, Mulas A, Peyret-Guzzon M, Zoledziewska M, Baali A, Bycroft C, Cherkaoui M, Chiaroni J, Di Cristofaro J, Dina C, Dugoujon JM, Galan P, Gienza J, Kivisild T, Mazieres S, Melhaoui M, Metspalu M, Myers S, Pereira L, Ricaut FX, Brisighelli F, Cardinali I, Grugni V, Lancioni H, Pascali VL, Torroni A, Semino O, Matullo G, Achilli A, Olivieri A, Capelli C. Population structure of modern-day Italians reveals patterns of ancient and archaic ancestries in Southern Europe. *Sci Adv.* 2019 Sep 4;5(9):eaaw3492. doi: 10.1126/sciadv.aaw3492. eCollection 2019 Sep.
10. **Angius A**, Uva P, Oppo M, Buers I, Persico I, Onano S, Cuccuru G, Van Allen MI, Hulait G, Aubertin G, Muntoni F, Fry AE, Annerén G, Stattin EL, Palomares-Bralo M, Santos-Simarro F, Cucca F, Crisponi G, Rutsch F, Crisponi L. Exome sequencing in Crisponi/cold-induced sweating syndrome-like individuals reveals unpredicted alternative diagnoses. *Clin Genet.* 2019 Mar 12. doi: 10.1111/cge.13532
11. **Angius A**, Uva P, Oppo M, Persico I, Onano S, Olla S, Pes V, Perria C, Cuccuru G, Atzeni R, Serra G, Cucca F, Sotgiu S, Hennekam RC, Crisponi L. Confirmation of a new phenotype in an individual with a variant in the last part of exon 30 of CREBBP. *American Journal of Medical Genetics Part A.*; 2019; doi:10.1002/ajmg.a.61052.
12. Alves RM, Uva P, Veiga MF, Oppo M, Zschaber F, Porcu G, Porto HP, Persico I, Onano S, Cuccuru G, Atzeni R, Vieira L, Pires M, Cucca F, Toralles MB, **Angius A**, Crisponi L. Novel ANKRD11 gene mutation in an individual with a mild phenotype of KBG syndrome associated to a GEFS+ phenotypic spectrum: a case report. *BMC Medical Genetics*; 2019; 20:16. doi: 10.1186/s12881-019-0745-7
13. Pisanu C, Congiu D, Costa M, Chillotti C, Ardaur R, Severino G, **Angius A**, Heilbronner U, Hou L, McMahon FJ, Schulze TG, Del Zompo M, Squassina A. Convergent analysis of genome-wide genotyping and transcriptomic data suggests association of zinc finger genes with lithium response in bipolar disorder. *Am J Med Genet B Neuropsychiatr Genet.* 2018 Oct;177(7):658-664. doi: 10.1002/ajmg.b.32663.

14. Chiang CWK, Marcus JH, Sidore C, Biddanda A, Al-Asadi H, Zoledziewska M, Pitzalis M, Busonero F, Maschio A, Pistis G, Steri M, **Angius A**, Lohmueller KE, Abecasis GR, Schlessinger D, Cucca F, Novembre J. Genomic history of the Sardinian population. *Nat Genet.* 2018 Oct;50(10):1426-1434. doi: 10.1038/s41588-018-0215-8.
15. Egorov ES, Kasatskaya SA, Zubov VN, Izraelson M, Nakonechnaya TO, Staroverov DB, **Angius A**, Cucca F, Mamedov IZ, Rosati E, Franke A, Shugay M, Pogorelyy MV, Chudakov DM, Britanova OV. The changing landscape of naive T cell TCR repertoire with human ageing. *Front Immunol.* 2018 Jul 24; 9:1618. doi: 10.3389/fimmu.2018.01618. eCollection 2018.
16. Uva P, Cossu-Rocca P, Loi F, Pira G, Murgia L, Orrù S, Floris M, Muroli MR, Sanges F, Carru C, **Angius A**, De Miglio MR. miRNA-135b contributes to Triple Negative Breast Cancer molecular heterogeneity: different expression profile in Basal-like versus non-Basal-like phenotypes. *Int J Med Sci* 2018; 15(6):536-548. doi:10.7150/ijms.23402
17. **Angius A**, Cossu S, Uva P, Oppo M, Onano S, Persico I, Fotia G, Atzeni R, Cuccuru G, Asunis M, Cucca F, Pruna D, Crisponi L. Novel NALCN biallelic truncating mutations in siblings with IHPRF1 syndrome. *Clin Genet.* 2018 Jun;93(6):1245-1247. doi: 10.1111/cge.13162.
18. Cossu S, **Angius A**, Oppo M, Onano S, Persico I, Uva P, Cuccuru G, Asunis M, Pruna D, Crisponi L. Epileptic phenotypes related to the UNC79-UNC80-NALCN protein complex. *Clinical Cases and Reviews in Epilepsy* 2017; 2(1): 54-58. doi: 10.11138/ccre/2017.2.1.054
19. Maccari ME, Scarselli A, Di Cesare S, Floris M, **Angius A**, Deodati A, Chiriaco M, Cambiaso P, Corrente S, Colafati GF, Utzh PJ, Angelini F, Fierabracci A, Aiuti A, Carsetti R, Rosenbergh JM, Cappa M, Rossi P, Bacchetta R, Cancrini C. Severe *Toxoplasma gondii* infection in a member of a NFKB2-deficient family with T and B cell dysfunction. *Clin Immunol.* 2017 Oct; 183:273-277. doi: 10.1016/j.clim.2017.09.011.
20. Pisanu C, Congiu D, Melis C, **Angius A**, Severino G, Ardaù R, Chillotti C, Del Zompo M, Squassina A. Involvement of core clock genes in lithium response. *World J Biol Psychiatry.* 2017 Jul 12:1-2. doi: 10.1080/15622975.2017.1346281
21. Malavasi V, Costelli C, Orsini M, Cusano R, **Angius A**, Cao G. Deep genomic analysis of *Chlorella sorokiniana* SAG 211-8k chloroplast. *European Journal of Phycology.* 2017 52(3): 320-329 doi: 10.1080/09670262.2017.1287959
22. Steri M, Orrù V, Idda ML, Pitzalis M, Pala M, Zara I, Sidore C, Faà V, Floris M, Deiana M, Asunis I, Porcu E, Mulas A, Piras MG, Lobina M, Lai S, Marongiu M, Serra V, Marongiu M, Sole G, Busonero F, Maschio A, Cusano R, Cuccuru G, Deidda F, Poddie F, Farina G, Dei M, Virdis F, Olla S, Satta MA, Pani M, Delitala A, Cocco E, Frau J, Coghe G, Lorefice L, Fenu G, Ferrigno P, Ban M, Barizzone N, Leone M, Guerini FR, Piga M, Firinu D, Kockum I, Lima Bomfim I, Olsson T, Alfredsson L, Suarez A, Carreira PE, Castillo-Palma MJ, Marcus JH, Congia M, **Angius A**, Melis M, Gonzalez A, Alarcón Riquelme ME, da Silva BM, Marchini M, Danieli MG, Del Giacco S, Mathieu A, Pani A, Montgomery SB, Rosati G, Hillert J, Sawcer S, D'Alfonso S, Todd JA, Novembre J, Abecasis GR, Whalen MB, Marrosu MG, Meloni A, Sanna S, Gorospe M, Schlessinger D, Fiorillo E, Zoledziewska M, Cucca F. Overexpression of the Cytokine BAFF and Autoimmunity Risk. *N Engl J Med.* 2017 Apr 27;376(17):1615-1626. doi: 10.1056/NEJMoa1610528.
23. Pala M, Zappala Z, Marongiu M, Li X, Davis JR, Cusano R, Crobu F, Kukurba KR, Gloudemans MJ, Reinier F, Berutti R, Piras MG, Mulas A, Zoledziewska M, Marongiu M, Sorokin EP, Hess GT, Smith KS, Busonero F, Maschio A, Steri M, Sidore C, Sanna S, Fiorillo E, Bassik MC, Sawcer SJ, Battle A, Novembre J, Jones C, **Angius A**, Abecasis GR, Schlessinger D, Cucca F, Montgomery SB. Population and individual-specific regulatory variation in Sardinia. *Nat Genet.* 2017 May;49(5):700-707. doi: 10.1038/ng.3840.
24. Olivieri A, Sidore C, Achilli A, **Angius A**, Posth C, Furtwängler A, Brandini S, MRCapodiferro, Gandini F, Zoledziewska M, Pitzalis M, Maschio A, Busonero F, Lai L, Skeates R, Gradoli MG, Beckett J, Marongiu M, Mazzarello M, Marongiu P, Rubino S, Rito T, Macaulay V, Semino O, Pala M, Abecasis GR, Schlessinger D, Soares P, Richards MB, Cucca F, Torroni A. Mitogenome Diversity in Sardinians: a Genetic Window onto an Island's Past. *Mol Biol Evol.* 2017 May 1;34(5):1230-1239. doi: 10.1093/molbev/msx082
25. Palombo F, Al-Wardy N, Ruscone GA, Oppo M, Kindi MN, **Angius A**, Al Lamki K, Giroto G, Giangregorio T, Benelli M, Magi A, Seri M, Gasparini P, Cucca F, Sazzini M, Al Khabori M, Pippucci T, Romeo G. A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. *J Hum Genet.* 2017 Feb;62(2):259-264. doi: 10.1038/jhg.2016.120.
26. McCarthy S, Das S, Kretschmar W, Delaneau O, Wood AR, Teumer A, Kang HM, Fuchsberger C, Danecek P, Sharp K, Luo Y, Sidore C, Kwong A, Timpson N, Koskinen S, Vrieze S, Scott LJ, Zhang H, Mahajan A, Veldink J, Peters U, Pato C, van Duijn CM, Gillies CE, Gandin I, Mezzavilla M, Gilly A, Cocca M, Traglia M, **Angius A**, Barrett JC, Boomsma D, Branham K, Breen G, Brummett CM, Busonero F, Campbell H, Chan A, Chen S, Chew E, Collins FS, Corbin LJ, Smith GD, Dedoussis G, Dorr M, Farmaki AE, Ferrucci L, Forer L, Fraser RM, Gabriel S, Levy S, Groop L, Harrison T, Hattersley A, Holmen OL, Hveem K, Kretzler M, Lee JC, McGue M, Meitinger T, Melzer D, Min JL, Mohlke KL, Vincent JB, Nauck M, Nickerson D, Palotie A, Pato M, Pirastu N, McInnis M, Richards JB, Sala C, Salomaa V, Schlessinger D, Schoenherr S, Slagboom PE, Small K, Spector T, Stambolian D, Tuke M, Tuomilehto J, Van den Berg LH, Van Rheenen V, Volker U, Wijmenga C, Toniolo D, Zeggini E, Gasparini P, Sampson MG, Wilson JF, Frayling T, de Bakker PI, Swertz MA, McCarroll S, Kooperberg C, Dekker A, Altshuler D, Willer C, Iacono W, Ripatti S, Soranzo N, Walter K, Swaroop A, Cucca F, Anderson CA, Myers RM, Boehnke M, McCarthy MI, Durbin R; Haplotype Reference Consortium. A reference panel of 64,976 haplotypes for genotype imputation. *Nat Genet.* 2016 Oct; 48(10):1279-83. doi: 10.1038/ng.3643.
27. Orsini M, Costelli C, Malavasi V, Cusano R, Concas A, **Angius A**, Cao G. Complete sequence and characterization of mitochondrial and chloroplast genome of *Chlorella variabilis* NC64A. *Mitochondrial DNA A DNA Mapp Seq Anal.* 2016 Sep;27(5):3128-30. doi: 10.3109/19401736.2015.1007297.
28. Greco CM, Kunderfranco P, Rubino M, Larcher V, Carullo P, Anselmo A, Kurz K, Carell T, **Angius A**, Latronico MV, Papait R, Condorelli G. DNA hydroxymethylation controls cardiomyocyte gene expression in development and hypertrophy. *Nat Commun.* 2016 Aug 4; 7:12418. doi: 10.1038/ncomms12418.

29. **Angius A**, Uva P, Buers I, Oppo M, Puddu A, Onano S, Persico I, Loi A, Marcia L, Höhne W, Cuccuru G, Fotia G, Deiana M, Marongiu M, Atalay AT, Inan S, El Assy O, Smit LME, Okur I, Boduroglu K, Utine GE, Kılıç E, Zampino G, Crisponi G, Crisponi L, Rutsch F. Bi-allelic Mutations in KLHL7 cause a Crisponi/CISS1-like phenotype associated with early onset retinitis pigmentosa. *Am J Hum Genet*, July 7, 2016; 99 (1): 236-245. doi: 10.1016/j.ajhg.2016.05.026
30. Marongiu M, Deiana M, Marcia L, Sardellati A, Asunis I, Meloni A, **Angius A**, Cusano R, Loi A, Crobu F, Fotia G, Cucca F, Schlessinger D, Crisponi L. Novel action of FOXL2 as mediator of Col1a2 gene autoregulation. *Dev Biol*. 2016 May 19. pii: S0012-1606(16)30070-7. doi: 10.1016/j.ydbio.2016.05.022.
31. Orsini M, Costelli C, Malavasi V, Cusano R, Concas A, **Angius A**, Cao G. Complete genome sequence of mitochondrial DNA (mtDNA) of *Chlorella sorokiniana*. *Mitochondrial DNA A DNA Mapp Seq Anal*. 2016;27(2):1539-41. doi: 10.3109/19401736.2014.953128.
32. Orsini M, Cusano R, Costelli C, Malavasi V, Concas A, **Angius A**, Cao G. Complete genome sequence of chloroplast DNA (cpDNA) of *Chlorella sorokiniana*. *Mitochondrial DNA A DNA Mapp Seq Anal*. 2016;27(2):838-9. doi: 10.3109/19401736.2014.919466. PubMed PMID: 24865923.
33. Erre G, Piga M, Carru C, **Angius A**, Carcangiu L, Piras M, Sotgia S, Zinellu A, Mathieu A, Passiu G, Pescatori M. Global microRNA profiling of peripheral blood mononuclear cells in patients with Behçet's disease. *Clin Exp Rheumatol*. 2015 Nov-Dec;33(6 Suppl 94):S72-9.
34. Ding J, Sidore C, Butler TJ, Wing MK, Qian Y, Meirelles O, Busonero F, Tsoi LC, Maschio A, **Angius A**, Kang HM, Nagaraja R, Cucca F, Abecasis GR, Schlessinger D. Correction: Assessing Mitochondrial DNA Variation and Copy Number in Lymphocytes of ~2,000 Sardinians Using Tailored Sequencing Analysis Tools. *PLoS Genet*. 2015 Sep 29;11(9):e1005549. doi: 10.1371/journal.pgen.1005549. eCollection 2015 Sep.
35. Sidore C, Busonero F, Maschio A, Porcu E, Naitza S, Zoledziewska M, Mulas A, Pistis G, Steri M, Danjou F, Kwong A, Ortega Del Vecchyo VD, Chiang CW, Bragg-Gresham J, Pitzalis M, Nagaraja R, Tarrier B, Brennan C, Uzzau S, Fuchsberger C, Atzeni R, Reinier F, Berutti R, Huang J, Timpson NJ, Toniolo D, Gasparini P, Malerba G, Dedoussis G, Zeggini E, Soranzo N, Jones C, Lyons R, **Angius A**, Kang HM, Novembre J, Sanna S, Schlessinger D, Cucca F, Abecasis GR. Genome sequencing elucidates Sardinian genetic architecture and augments GWAS findings: the examples of lipids and blood inflammatory markers. *Nat Genet*. 2015 Nov;47(11):1272-81. doi: 10.1038/ng.3368.
36. Danjou F, Zoledziewska M, Sidore C, Steri M, Busonero F, Maschio A, Mulas A, Perseu L, Barella S, Porcu E, Pistis G, Pitzalis M, Pala M, Menzel S, Metrustry S, Spector TD, Leoni L, **Angius A**, Uda M, Moi P, Thein SL, Galanello R, Abecasis GR, Schlessinger D, Sanna S, Cucca F. Whole genome sequencing-based GWAS in Sardinia explicates genetic regulation of hemoglobin levels and clinical consequences. *Nat Genet*. 2015 Nov;47(11):1264-71. doi: 10.1038/ng.3307.
37. Zoledziewska M, Sidore C, Chiang CW, Sanna S, Mulas A, Steri M, Busonero F, Marcus JH, Marongiu M, Maschio A, Del Vecchyo DO, Floris M, Meloni A, Delitala A, Concas MP, Murgia F, Biino G, Vaccargiu S, Nagaraja R, Lohmueller KE; UK10K Consortium, Timpson NJ, Soranzo N, Tachmazidou I, Dedoussis G, Zeggini E; Understanding Society Scientific Group, Uzzau S, Jones C, Lyons R, **Angius A**, Abecasis GR, Novembre J, Schlessinger D, Cucca F. Major height reducing variants and selection for short stature on the island of Sardinia. *Nat Genet*. 2015 Nov;47(11):1352-6. doi: 10.1038/ng.3403.
38. Reinier F, Zoledziewska M, Hanna D, Smith JD, Valentini M, Zara I, Berutti R, Sanna S, Oppo M, Cusano R, Montesu MA, Jones C, Cerimele D, Nickerson DA, **Angius A**, Cucca F, Cottoni F, Crisponi L. Mandibular hypoplasia, deafness, progeroid features and lipodystrophy (MDPL) syndrome: a case report in the context of other inherited lipodystrophies. *Metabolism*. 2015 Nov;64(11):1530-40. doi: 10.1016/j.metabol.2015.07.022.
39. Ding J, Sidore C, Butler TJ, Wing MK, Qian Y, Meirelles O, Busonero F, Tsoi LC, Maschio A, **Angius A**, Kang HM, Nagaraja R, Cucca F, Abecasis GR, Schlessinger D. Assessing mitochondrial DNA variation and copy number in lymphocytes of ~2,000 Sardinians using tailored sequencing analysis tools. *PLOS Genetics*. 2015 Jul; doi: 10.1371/journal.pgen.1005306
40. Costa M, Squassina A, Piras IS, Pisanu C, Congiu D, Niola P, **Angius A**, Chillotti C, Ardaur R, Severino G, Stochino E, Deidda A, Persico AM, Alda M, Del Zompo M. Preliminary Transcriptome Analysis in Lymphoblasts from Cluster Headache and Bipolar Disorder Patients Implicates Dysregulation of Circadian and Serotonergic Genes. *J Mol Neurosci*. 2015 Jul;56(3):688-95. doi: 10.1007/s12031-015-0567-9.
41. Concas A, Costelli C, Malavasi V, Orsini M, Cusano R, Pisu M, **Angius A**, Cao G. The Role of Mathematical Modeling and Genetic Engineering for the Microalgae Based Technology. *Chemical Engineering Transactions* 2015 May 43: 511-16. doi: 10.3303/CET1543086
42. Francalacci P, Sanna D, Useli A, Berutti R, Barbato M, Whalen MB, **Angius A**, Sidore C, Alonso S, Tofanelli S, Cucca F. Detection of phylogenetically informative polymorphisms in the entire euchromatic portion of human Y chromosome from a Sardinian sample. *BMC Res Notes*. 2015 Apr 30;8(1):174.
43. Palagano E, Blair HC, Pangrazio A, Tourkova I, Strina D, **Angius A**, Cuccuru G, Oppo M, Uva P, Van Hul W, Boudin E, Superti-Furga A, Faletra F, Nocerino A, Ferrari MC, Grappiolo G, Monari M, Montanelli A, Vezzoni P, Villa A, Sobacchi C. Buried in the Middle, But Guilty: Intronic Mutations in the TCIRG1 Gene Cause Human Autosomal Recessive Osteopetrosis. *J Bone Miner Res*. 2015 Mar 31. doi: 10.1002/jbmr.2517.
44. Centorame P, Acciari VA, Orsini M, Torresi M, Iannetti L, **Angius A**, Di Giammartino D, Prencipe VA, Migliorati G. Whole Genome Sequence of *Listeria monocytogenes* Serovar 4b strain IZSAM_Lm_hs2008 Isolated from a human infection in Italy. *Genome Announc*. 2015 Mar 5;3(2). pii: e00053-15. doi: 10.1128/genomeA.00053-15.
45. Pistis G, Porcu E, Vrieze SI, Sidore C, Steri M, Danjou F, Busonero F, Mulas A, Zoledziewska M, Maschio A, Brennan C, Lai S, Miller MB, Marcelli M, Urru MF, Pitzalis M, Lyons RH, Kang HM, Jones CM, **Angius A**, Iacono WG, Schlessinger D, McGue M, Cucca F, Abecasis GR, Sanna S. Rare variants genotype imputation with thousands of study-specific whole-genome sequences: Implications for cost-effective study designs. *Eur J Hum Genet*. 2014 Oct 8. doi: 10.1038/ejhg.2014.216.

46. Cuccuru G, Leo S, Lianas L, Muggiri M, Pinna A, Pireddu L, Uva P, **Angius A**, Fotia G, Zanetti G. An Automated Infrastructure to Support High-throughput Bioinformatics. Proc. IEEE International Conference on High Performance Computing & Simulation (HPCS 2014) pages 600-607 Smari, Waleed W. and Zeljkovic, Vesna Eds. - July 2014. Doi: 10.1109/HPCSim.2014.6903742
47. Piras IS, **Angius A**, Andreani M, Testi M, Lucarelli G, Floris M, Markt S, Ciceri F, Nasa GL, Fleischhauer K, Roncarolo MG, Bulfone A, Gregori S, Bacchetta R. BAT2 and BAT3 polymorphisms as novel genetic risk factors for rejection after HLA-related stem cell transplantation. Bone Marrow Transplant. 2014 Nov;49(11):1400-4. doi: 10.1038/bmt.2014.177.
48. Delaneau O, Marchini J, 1000 Genomes Project Consortium (including **A Angius**). Integrating sequence and array data to create an improved 1000 Genomes Project haplotype reference panel. Nature Communications 2014 Jun 13; 5:3934. doi: 10.1038/ncomms4934.
49. Colonna V, Ayub Q, Chen Y, Pagani L, Luisi P, Pybus M, Garrison E, Xue Y, Tyler-Smith C; 1000 Genomes Project Consortium (including **A Angius**), Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA. Human genomic regions with exceptionally high levels of population differentiation identified from 911 whole-genome sequences. Genome Biol. 2014 Jun 30;15(6):R88. doi: 10.1186/gb-2014-15-6-r88.
50. M Sikora, ML Carpenter, A Moreno-Estrada, BM Henn, PA Underhill, F Sánchez-Quinto, I Zara, M Pitzalis, C Sidore, F Busonero, A Maschio, **A Angius**, C Jones, J Mendoza-Revilla, TT Harkinsh, A Kelleri, F Maixner, A Zink, G Abecasis, S Sanna, F Cucca, CD Bustamante. Genomic data from modern Sardinians and ancient Bulgarians provides new insights into the Sardinian ancestry of the Tyrolean Iceman. PLoS Genet. 2014 May 8;10(5):e1004353. doi: 10.1371/journal.pgen.1004353.
51. Piras R, Chiappe F, Torraca IL, Buers I, Usala G, **Angius A**, Akin MA, Basel-Vanagaite L, Benedicenti F, Chiodin E, El Assy O, Feingold-Zadok M, Guibert J, Kamien B, Kasapkara CS, Kiliç E, Boduroğlu K, Kurtoglu S, Manzur AY, Onal EE, Paderi E, Roche CH, Tümer L, Unal S, Utine GE, Zanda G, Zankl A, Zampino G, Crisponi G, Crisponi L, Rutsch F. Expanding the mutational spectrum of CRLF1 in Crisponi/CISS1 Syndrome. Hum Mutat. 2014 Apr;35(4):424-33. doi: 10.1002/humu.22522.
52. Gravel S, Zakharia F, Moreno-Estrada A, Byrnes JK, Muzzio M, Rodriguez-Flores JL, Kenny EE, Gignoux CR, Maples BK, Guiblet W, Dutil J, Via M, Sandoval K, Bedoya G; 1000 Genomes Project (including **A Angius**), Oleksyk TK, Ruiz-Linares A, Burchard EG, Martinez-Cruzado JC, Bustamante CD. Reconstructing Native American migrations from whole-genome and whole-exome data. PLoS Genet. 2013;9(12):e1004023. doi: 10.1371/journal.pgen.1004023.
53. Pippucci T, Parmeggiani A, Palombo F, Maresca A, **Angius A**, Crisponi L, Cucca F, Liguori R, Valentino ML, Seri M, Carelli V. A novel null homozygous mutation confirms CACNA2D2 as a gene mutated in epileptic encephalopathy. PLoS ONE 2013 8(12): e82154. doi:10.1371/journal.pone.0082154
54. Khurana E, Fu Y, Colonna V, Mu XJ, Kang HM, Lappalainen T, Sboner A, Lochovsky L, Chen J, Harmanci A, Das J, Abyzov A, Balasubramanian S, Beal K, Chakravarty D, Challis D, Chen Y, Clarke D, Clarke L, Cunningham F, Evani US, Flicek P, Fragoza R, Garrison E, Gibbs R, Gümüs ZH, Herrero J, Kitabayashi N, Kong Y, Lage K, Liluashvili V, Lipkin SM, MacArthur DG, Marth G, Muzny D, Pers TH, Ritchie GR, Rosenfeld JA, Sisu C, Wei X, Wilson M, Xue Y, Yu F; 1000 Genomes Project Consortium (including **A Angius**), Dermitzakis ET, Yu H, Rubin MA, Tyler-Smith C, Gerstein M. Integrative annotation of variants from 1092 humans: application to cancer genomics. Science. 2013 Oct 4;342(6154):1235587. doi: 10.1126/science.1235587.
55. Orrù V, Steri M, Sole G, Sidore C, Virdis F, Dei M, Lai S, Zoledziewska M, Busonero F, Mulas A, Floris M, Mentzen WI, Urru SA, Olla S, Marongiu M, Piras MG, Lobina M, Maschio A, Pitzalis M, Urru MF, Marcelli M, Cusano R, Deidda F, Serra V, Oppo M, Pilu R, Reinier F, Berutti R, Pireddu L, Zara I, Porcu E, Kwong A, Brennan C, Tarrier B, Lyons R, Kang HM, Uzzau S, Atzeni R, Valentini M, Firinu D, Leoni L, Rotta G, Naitza S, **Angius A**, Congia M, Whalen MB, Jones CM, Schlessinger D, Abecasis GR, Fiorillo E, Sanna S, Cucca F. Genetic Variants Regulating Immune Cell Levels in Health and Disease. Cell. 2013 Sep 26;155(1):242-56. doi: 10.1016/j.cell.2013.08.041.
56. Francalacci P, Morelli L, **Angius A**, Berutti R, Reinier F, Atzeni R, Pilu R, Busonero F, Maschio A, Zara I, Sanna D, Useli A, Urru MF, Marcelli M, Cusano R, Oppo M, Zoledziewska M, Pitzalis M, Deidda F, Porcu E, Poddie F, Kang HM, Lyons R, Tarrier B, Gresham JB, Li B, Tofaneli S, Alonso S, Dei M, Lai S, Mulas A, Whalen MB, Uzzau S, Jones C, Schlessinger D, Abecasis GR, Sanna S, Sidore C, Cucca F. Large-scale low-coverage sequencing from the highly informative Sardinian population permits reconstruction of Y chromosome phylogeny at the European continental level. Science. 2013 Aug 2;341(6145):565-9. doi: 10.1126/science.1237947.
57. Esposito T, Lea RA, Maher BH, Moses D, Cox HC, Magliocca S, **Angius A**, Nyholt DR, Titus T, Kay T, Gray NA, Parnham A, Gianfrancesco F, Griffiths LR. Unique X-linked familial FSGS with co-segregating heart block disorder is associated with a mutation in the NXF5 gene. Hum Mol Genet. 2013 Sep 15;22(18):3654-66. doi: 10.1093/hmg/ddt215.
58. Squassina A, Costa M, Congiu D, Manchia M, **Angius A**, Deiana V, Ardaur R, Chillotti C, Severino G, Calza S, Del Zompo M. Insulin-like growth factor 1 (IGF-1) expression is up-regulated in lymphoblastoid cell lines of lithium responsive bipolar disorder patients. Pharmacol Res. 2013 Jul;73:1-7. doi: 10.1016/j.phrs.2013.04.004.
59. Biino G, Parati G, Concas MP, Adamo M, **Angius A**, Vaccargiu S, Pirastu M. Environmental and genetic contribution to hypertension: prevalence: data from and epidemiological survey on Sardinian genetic isolates. PLoS One. 2013;8(3):e59612. doi: 10.1371/journal.pone.0059612.
60. 1000 Genomes Project Consortium, (including **A Angius**), Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA. An integrated map of genetic variation from 1,092 human genomes. Nature 2012 Nov;491(7422):56-65.
61. Sanna S, Pitzalis M, Zoledziewska M, Zara I, Sidore C, Murru R, Whalen MB, Busonero F, Maschio A, Costa G, Melis MC, Deidda F, Poddie F, Morelli L, Farina G, Li Y, Dei M, Lai S, Mulas A, Cuccuru G, Porcu E, Liang L, Zavattari P, Moi L, Deriu E, Urru MF, Bajorek M, Satta MA, Cocco E, Ferrigno P, Sotgiu S, Pugliatti M, Tracis

- S, **Angius A**, Melis M, Rosati G, Abecasis GR, Uda M, Marrosu MG, Schlessinger D, Cucca F. Variants within the immunoregulatory CBLB gene are associated with multiple sclerosis. *Nat Genet.* 2010 Jun;42(6):495-7.
62. E Mocci, MP Concas, M Fanciulli, N Pirastu, M Adamo, C Valentina, C Fraumene, I Persico, A Sassu, A Picciau, D Prodi, D Serra, G Biino, M Pirastu, **A Angius**. 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. doi: 10.1186/1471-2350-10-81.
 63. Vago L, Perna SK, Zanussi M, Mazzi B, Barlassina C, Stanghellini MT, Perrelli NF, Cosentino C, Torri F, **Angius A**, Forno B, Casucci M, Bernardi M, Peccatori J, Corti C, Bondanza A, Ferrari M, Rossini S, Roncarolo MG, Bordignon C, Bonini C, Ciceri F, Fleischhauer K. Genomic loss of mismatched HLA in leukemia is a major mechanism of in vivo escape from T cell immunosurveillance following haploidentical HSCT. *N Engl J Med.* 2009 Jul 30;361(5):478-88.
 64. G Pistis, I Piras, N Pirastu, I Persico, A Sassu, A Picciau, D Prodi, C Fraumene, E Mocci, MT Manias, R Atzeni, M Cosso, M Pirastu, **A Angius**. 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. doi: 10.1371/journal.pone.0004654.
 65. DA Prodi, N Pirastu, G Maninchedda, A Sassu, A Picciau, MA Palmas, A Mossa, I Persico, M Adamo, **A Angius**, Pirastu M. EDA2R Is Associated with Androgenetic Alopecia. *J Invest Dermatol.* 2008 Apr 3.
 66. S Sotgiu, A Angius, A Embry, G Rosati, S Musumeci. Hygiene hypothesis: innate immunity, malaria and multiple sclerosis. *Med Hypotheses.* 2008;70(4):819-25.
 67. **A Angius**, FCL Hyland, I Persico, N Pirastu, T Woodage, M Pirastu, FM De La Vega 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.
 68. **A Angius**, J Simporè, I Persico, A Sassu, DA Prodi, S Musumeci. Methylenetetrahydrofolate reductase gene polymorphisms in Burkina Faso: impact on plasma fasting homocysteine and after methionine loading test. *Clin Lab* 2007; 53:29-33.
 69. C Fraumene, EMS Belle, L Castri, S Sanna, G Mancosu, M Cosso, F Marras, G Barbuji, M Pirastu, **A Angius**. High resolution analysis and phylogenetic network construction using complete mtDNA sequences in Sardinian genetic isolates. *Mol Biol Evol.*2006; 23: 2101-2111.
 70. M Musumeci, J Simporè, R Barone, **A Angius**, S Musumeci. Synchronic macrophage response and Plasmodium falciparum malaria. *J Vect Borne Dis* 2006, 43: 84–87.
 71. R Chillemi, **A Angius**, I Persico, A Sassu, DA Prodi, S Musumeci. Methylenetetrahydrofolate reductase (MTHFR) from Mediterranean to Sub-Saharan Areas. *J Biol Sciences* 2005: 6 (1): 28-34.
 72. J Simporè, **A Angius**, I Persico, A Sassu, DA Prodi, S Musumeci. Methylenetetrahydrofolate reductase gene polymorphisms in Burkina Faso. *Clin Chim Acta.* 2005; 360(1-2): 199-200.
 73. M Musumeci, J Simporè, R Barone, **A Angius**, L Malaguarnera, S Musumeci. Synchronic macrophage response and Plasmodium falciparum malaria. *Pak J Biol Sci* 2005 8 (7): 954-958.
 74. M Falchi, P Forabosco, E Mocci, C Cappio Borlino, A Picciau, E Viridis, I Persico, D Parracciani, **A Angius**, M Pirastu. Genome-wide search using an original pairwise sampling approach for large genealogies identifies a new locus for total and LDL-cholesterol in two genetically differentiated, isolates of Sardinia. *Am J Hum Genet* 2004; 75(6):1015-31.
 75. L Malaguarnera, R Barone, **A Angius**, S Musumeci. Chitotriosidase, a prematurely orphan enzyme. *Hum Evol*, 2004, 19 (1): 71-75
 76. DA Prodi, D Drayna, P Forabosco, MA Palmas, GB Maestrale, D Piras, M Pirastu, **A Angius**. Bitter taste study in a Sardinian Genetic Isolate supports the association of phenylthiocarbamide sensitivity to the TAS2R38 bitter receptor gene. *Chem Senses* 2004; 29(8):697-702.
 77. A Tenesa, AF Wright, SA Knott, C Carothers, C Hayward, **A Angius**, I Persico, GB Maestrale, ND Hastie, M Pirastu, SS Visscher. Extent of linkage disequilibrium in a Sardinian sub-isolate: sampling and methodological considerations. *Hum Mol Genet* 2004, 13 (1): 25-33.
 78. L Malaguarnera, J Simporè, DA Prodi, **A Angius**, A Sassu, I Persico, R Barone, S Musumeci. A 24-base pair duplication in exon 10 of Human Chitotriosidase gene from the sub-Saharan to the Mediterranean area: role of parasitic diseases and environmental conditions. *Genes Immun* 2003; 4 (8): 570-4.
 79. C Fraumene, E Petretto, **A Angius**, M Pirastu. Striking differentiation of sub-populations within a genetically homogeneous isolate (Ogliastra) in Sardinia as revealed by mtDNA analysis. *Hum Genet* 2003; 114 (1): 1-10.
 80. R Barone, L Malaguarnera, **A Angius**, S Musumeci. Plasma Chitotriosidase activity in patients with beta-thalassemia. *Am J Haemat* 2003; 72 (4): 285-286.
 81. **A Angius**, E Petretto, GB Maestrale, P Forabosco, G Casu, D Piras, M Fanciulli, M Falchi, PM Melis, M Palermo, M Pirastu. A new essential Hypertension susceptibility locus on chromosome 2p24-25 detected by genome-wide search. *Am J Hum Genet.* 2002; 71 (4): 893-905.
 82. **A Angius**, D Bebbere, E Petretto, M Falchi, P Forabosco, GB Maestrale, G Casu, I Persico, PM Melis, M Pirastu. Not all isolates are equal: linkage disequilibrium analysis on Xq13.3 reveals different patterns in Sardinian sub-populations. *Hum Genet.* 2002; 111 (1): 9-15.
 83. E Vitale, C Specchia, M Devoto, **A Angius**, S Rong, M Rocchi, M Schwalb, L Demelas, D Paglietti, S Manca, C Mastropaolo, G Serra. Novel X-Linked mental retardation syndrome with short stature maps to Xq24. *Am J Med Genet* 2001; 103 (1): 1-8.
 84. **A Angius**, PM Melis, L Morelli, E Petretto, G Casu, GB Maestrale, C Fraumene, D Bebbere, P Forabosco, M Pirastu Archival, demographic and genetic studies define a Sardinian sub-isolate as a suitable model for mapping complex traits. *Hum Genet* 2001; 109 (2): 198-209.
 85. Ombra MN, Forabosco P, Casula S, **Angius A**, Maestrale GB, Petretto E, Casu G, Colussi G, Usai E, Melis P, Pirastu M Identification of a new candidate locus for uric acid nephrolithiasis in a genetic isolate. *Am J Hum Genet* 2001; 68: 1119-1129.

86. **A Angius**, P Forabosco, G Maestrale, G Casu, D Piras, A Pala, M Palermo, P Melis, M Pirastu. Genome-wide screen for essential hypertension genes in a deep rooted Sardinian pedigree. *Ann Hum Genet*, 2000; 64 (5): 475-476.
87. **A Angius**, P Spinelli, G Ghilotti, G Casu, G Sole, A Loi, A Totaro, L Zelante, P Gasparini, N Orzalesi, M Pirastu, L Bonomi. TIGR Gln368stop defect and advanced age are two risk factors in a large late onset POAG family. *Arch Ophthalmol* 2000; 118 (5): 674-9.
88. G Palomba, C Rozzo, **A Angius**, N Orzalesi, M Pirastu. A novel spontaneous missense mutation in VMD2 gene is cause of a Best's macular dystrophy (BMD) sporadic case. *Am J Ophthalmol* 2000; 129 (2): 260-2.
89. M Pisano, A Cossu, I Persico, G Palmieri, **A Angius**, G Casu, G Palomba, M G Sarobba, P Cossu Rocca, MF Dedola, N Olmeo, A Pasca, M Budroni, V Marras, A Pisano, A Farris, G Massarelli, M Pirastu, F Tanda. Identification of a founder BRCA2 mutation in Sardinia. *Br J Cancer* 2000; 82 (3): 553-9.
90. G Loudianos, V Dessi, M Lovicu, **A Angius**, M Pirastu, A Cao. Haplotype and mutation analysis in Mediterranean patients with Wilson disease. *J Trace Elem in Exp Med* 1999; 12: 315-319.
91. G Loudianos, V Dessi, M Lovicu, A Angius, B Altuntas, R Giacchino, M Marazzi, M Marcellini, MR Sartorelli, GC Sturniolo, N Kocak, A Yuce, N Akar, M Pirastu, A Cao. Mutation analysis in patients of Mediterranean descent with Wilson disease. Identification of 19 novel mutations. *J Med Genet* 1999; 36 (11): 833-836.
92. G Loudianos, V Dessi, M Lovicu, **A Angius**, AL Figus, F Lilliu, S De Virgiliis, A Nurchi, A Deplano, M Pirastu, A Cao. Molecular characterization of Wilson disease in the Sardinian population. Evidence of a founder effect. *Hum Mutat* 1999; 14 (4): 294-303.
93. **A Angius**, M Pisano, A Manca, G Casu, I Persico, S Pitzalis, E De Gioia, F Grignolo, A Loi, G Sole, A Cao, P Spinelli, G Ghilotti, L Bonomi, M Fossarello, A Serra, S Gandolfi, G Alberti, G Maraini, A Serru, N Orzalesi, M Pirastu. Molecular basis of open-angle glaucoma in Italy. *Acta Ophthalmol Scand Suppl* 1998; (227): 16-7.
94. Loudianos G, Dessi V, Lovicu M, **Angius A**, Kanavakis E, Tzetis M, Kattamis C, Manolaki N, Vassiliki G, Karpathios T, Cao A, Pirastu M. Haplotype and mutation analysis in Greek patients with Wilson disease. *Eur J Hum Genet* 1998; 6: 487-91.
95. C Nobile, A Manca, M Pisano, **A Angius**, R Muresu, IC Gray, NK Spurr. A refined physical and EST map spanning 7.4 Mb of 10q24, a region involved in neurological disorders. *Mamm Genome* 1998; 9 (10): 835-837.
96. G Loudianos, V Dessi, M Lovicu, **A Angius**, S De Virgiliis, A Nurchi, A Solinas, M Marcellini, L Zancan, C Barbera, N Akar, R Yagci, A Vegnente, A Cao, M Pirastu. Further delineation of the molecular pathology of Wilson disease in the Mediterranean population. *Hum Mutat* 1998; 12: 89-94.
97. **A Angius**, E De Gioia, A Loi, M Fossarello, G Sole, N Orzalesi, F Grignolo, A Cao, M Pirastu. A novel mutation in the GLC1A gene causes Juvenile Open Angle Glaucoma in four families from the Italian region of Puglia. *Arch Ophthalmol* 1998; 116: 793-797.
98. G Loudianos, V Dessi, M Lovicu, **A Angius**, A Cao, M Pirastu. The -75 A>C substitution in the 5' UTR of the Wilson disease gene is a sequence polymorphism in the Mediterranean population. *Am J Hum Genet* 1998; 62 (2): 581.
99. **A Angius**, V Dessi, M Lovicu, S De Virgiliis, M Pirastu, A Cao. Early and severe neurological features in a Wilson disease patient compound heterozygous for two frameshift mutations. *Eur J Pediatr* 1998; 157: 128-129.
100. G Loudianos, V Dessi, **A Angius**, M Lovicu, A Loi, M Deiana, N Akar, P Vairo, A Figus, A Cao, M Pirastu. Wilson disease mutations associated with uncommon haplotypes in Mediterranean patients. *Hum Genet* 1996; 98: 640-642.
101. AL Figus, **A Angius**, G Loudianos, C Bertini, V Dessi, A Loi, M Deiana, M Lovicu, N Olla, G Sole, S De Virgiliis, F Lilliu, A Farci, A Nurchi, R Giacchino, A Barabino, MG Marassi, L Zancan, NA Greggio, M Marcellini, A Solinas, A Deplano, C Barbera, M Devoto, S Ozsoylu, N Kocak, N Akar, S Karayalcin, V Mokini, P Cullufi, A Balestrieri, A Cao and M Pirastu. Molecular pathology and haplotype analysis of Wilson's disease in Mediterranean populations. *Am J Hum Genet* 1995; 57: 1318-1324.
102. G Loudianos, AL Figus, A Loi, **A Angius**, V Dessi, M Deiana, S DeVirgiliis, G Monni, A Cao, M Pirastu. Improvement of prenatal diagnosis of Wilson disease using microsatellite markers. *Prenat Diagn* 1994; 14: 999-1002.

Italian Journals

103. Pira G, Murgia L, Sanges F, Uva P, Cossu-Rocca P, Loi F, Orrù S, Muroni MR, Carru C, **Angius A**, De Miglio MR. Espressione del miRNA-135b nella caratterizzazione molecolare del carcinoma della mammella "Triplo Negativo". *Biochimica Clinica* 2017(41(3)): 239-244 ·Doi: 10.19186/BC_2017.035
104. **Angius A**. Genetica del Glaucoma Primario ad Angolo Aperto: variabilità fenotipica nelle mutazioni più comuni del gene Miocillina. *Minerva Oftalmol.* 2002; 44 (2): 61-69.
105. Morelli L, Melis P, **Angius A**, Casu G, Cabras S, Pirastu M. Approccio molecolare e genealogico per la caratterizzazione di linee di discendenza materne e paterne in isolati genetici: applicazione alla genetica delle malattie multifattoriali. *Rivista di Antropologia*, 2000; 78: 219-228.
106. Boggio-Merlo S, Pescetti L, De Gioia E, **Angius A**, Brogliatti B, Grignolo FM. La genetica nel glaucoma primario ad angolo aperto: studio preliminare. *Bollettino di Oculistica* 1998, 77: 49-52.
107. Zedda N, Onnis E, **Angius A**, Balata F, Cherchi PA, Sole G, Olla N, Poddie D, Cao A, Cherchi A, Pirastu M. Esiste una predisposizione genetica per l'espansione infartuale? Valutazione dei polimorfismi genetici del sistema renina-angiotensina. *Cardiologia* 1997; 42 (3): 281-285.
108. Pirastu M, Figus A, Lilliu F, **Angius A**, Dessi V, Argiolas L, Contini I, Nurchi AM, Deplano A, De Virgiliis S. La malattia di Wilson. *Prospettive in pediatria* 1995; 97: 61-74.

Book Chapters

1. R Muzzarelli, N Dahiya, A Giansanti, F Mecozzi, S Musumeci (ed.), H Michelakakis, I Labadaridis, R Boot, D Speijer, AP Bussink, JMFG Aerts, **A Angius**, A Cenarro, F Civeira, J Brunner, S Sotgiu, LF Gonzalez Cuyar, RJ Castellani, XQ Pan, J Johansen, R Barone, M Musumeci, MA Seibold, EG Burchard, J Kzhyshkowska, MG Paoletti

- (ed.), N Lorenzo, E Cozzarini, E Mizoguchi, M Kawada, S Sakuda, F Falcone. Binomium Chitin-Chitinase: Recent Issues. Casa Editrice Nova Science Publishers. Pub. Date: 2009, 1st quarter. ISBN: 978-1-60692-339-9
2. **A Angius**, TS. Bianchi-Marzoli, B. Brogliatti, P. Brusini, R. Carassa, A. Carta, M. Centofanti, R. De Natale, L. Fontana, F. Galassi, S. Gandolfi (ed.), M. Lester, G. Manni, G. Marchini, E. Martini, L. Mastropasqua, S. Miglior, P. Nucci, V. Parisi, A. Perdicchi, M. Pirastu, L. Quaranta, M. Rolando, L. Rossetti, C. Traverso, M. Uva, M. Vetrugno. Il Glaucoma. Casa editrice Mattioli 1885. Anno: 2005. ISBN: 88-89397-11-X.