Curriculum Vitae - Andrea Angius

Name: *Andrea Angius*
Date of birth: *31/03/1964* Place: *Trieste (TS) Italy* Nationality: *Italian*
Present Positions: *Researcher at the Italian National Research Council*
Affiliation: *Institute of the Population Genetics, Italian National Research Council,CNR*
Trav. La Crucca, 3 - Reg. Baldinca - 07100 Li Punti - Sassari.

EDUCATION

1992 - University of Cagliari, Italy: *Degree in Biology*
1999 - University of Sassari, Italy: *Ph.D. in Biochemistry*

TEACHING

2003-2004 – *Course "Basic Element of Genetics and laboratory"*
Faculty of Mathematical, Physical and Natural Science, University of Sassari, SS (Italy).
Faculty of Biotechnologies, University of Sassari, SS (Italy).

2006-2007 / 2007-2008 /2008-2009/ 2009-2010– *Course "Human Genetics"*
Faculty of Mathematical, Physical and Natural Science, University of Sassari, SS (Italy).
Faculty of Biotechnologies, University of Sassari, SS (Italy).

RESEARCH EXPERIENCE

1990-1992 - *Undergraduate student.*
Department of Sperimental Biology, University of Cagliari, Cagliari (Italy). Supervisor: G. Floris, Ph.D..
- Anthropometrical characterization and population genetic studies of the Sardinian population.

1992-1994 - *Post-doctoral fellowship.*
Laboratory of Molecular Pathology, Ospedale regionale per le Microcitemie, Director: Antonio Cao, MD; Cagliari (Italy).
- Mapping and cloning of the gene responsible for Wilson disease. Characterization of the molecular bases of Wilson disease in Sardinia: linkage disequilibrium analysis on Wilson disease patients to identify the founder effect.

1995-1996 - *CNR fellowship.*
Institute of Research of Mediterranean Anemia and Thalassemia, National Research Council (CNR), Director: Antonio Cao, MD; Cagliari (Italy).
- Further studies on the molecular bases of Wilson disease in the Mediterranean area.
- Study of the genetic determinants of hypertension and myocardial infarction in the Sardinian population.

1996 – 1999 - *Ph. D.*
Faculty of Biology, University of Sassari, Sassari (Italy).
- Identification of genetic factors in eye disorders: identification of a mutation with founder effect in several families affected by Juvenile Primary Open Angle Glaucoma (POAG) and identification of genetic risk factors for adult-type POAG in a village of northern Italy.
- Identification of the molecular basis of congenital glaucoma in Italy.
- Identification of a founder BRCA2 mutation present in several Sardinian families with breast cancer.

2000 – up to present – *Researcher National Research Council (CNR), Institute of Population Genetics, Director: Mario Pirastu, MD; Alghero, Sassari (Italy).*
2002 – 2006 – *Director of the Genotyping laboratory, Soc. SHARDNA Life Sciences, Cagliari, (Italy).*
2006 – up to present – *Lab Manager and Scientific Director of the core-facility Genotyping Platform laboratory, "Polaris" Scientific and Technological Park of Sardinia, Pula (Cagliari) (Italy).*
- Study of complex disorders in an isolated and genetically homogenous populations in a particular area of Sardinia. Determination of the Ogliastra genetic diversity based on mtDNA data, Y chromosome and autosomal chromosome.
- Analysis of the genetic factors involved in Essential Hypertension and in Nephrolitiases: study of families with the same phenotype and common ancestors.
- Study of common complex diseases and Quantitative Traits (such as lipid profile, blood pressure, etc.) and search for the candidate genes.
- Deep sequencing using the second generation sequencers to evaluate the genetic diversity of the Sardinia population
- Extensive studies using hundreds of SNPs on the Y chromosome structure and MtDNA in whole Sardinia.

Publications

1. G Loudianos, AL Figus, A Loi, **A Angius**, V Dessì, 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.

2. M Pirastu, A Figus, F Lilliu, **A Angius**, V Dessì, L Argiolas, I Contini, AM Nurchi, A Deplano, S De Virgiliis. La malattia di Wilson. Prospettive in pediatria 1995; 97: 61-74.

3. AL Figus, **A Angius**, G Loudianos, C Bertini, V Dessì, 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.

4. G Loudianos, V Dessì, **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.

5. N Zedda, E Onnis, **A Angius**, F Balata, PA Cherchi, G Sole, N Olla, D Poddie, A Cao, A Cherchi, M Pirastu. Esiste una predisposizione genetica per l'espansione infartuale? Valutazione dei polimorfismi genetici del sistema renina-angiotensina. Cardiologia 1997; 42 (3): 281-285.

6. **A Angius**, V Dessì, 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.

7. G Loudianos, V Dessì, 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.

8. **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.

9. G Loudianos, V Dessì, 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.

10. 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.

11. 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.

12. **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 Ghillotti , 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.

13. G Loudianos, V Dessì, 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.

14. 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.

15. 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.

16. 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.

17. 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.

18. **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.

19. **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.

20. 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.

21. **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.

22. 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.

23. **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.

24. **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.

25. **A Angius**. Genetica del Glaucoma Primario ad Angolo Aperto: variabilità fenotipica nelle mutazioni più comuni del gene Miocillina. Minerva Oftalmol. 2002; 44 (2): 61-69.

26. R Barone, L Malaguarnera, **A Angius**, S Musumeci. Plasma Chitotriosidase activity in patients with β-thalassemia. Am J Haemat 2003; 72 (4): 285-286.

27. 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.

28. 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.

29. 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.

30. DA Prodi, D Drayna, F 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.

31. Malaguarnera L, Barone R, **Angius A**, Musumeci S. Chitotriosidase, a prematurely orphan enzyme. Hum Evol, 2004, 19 (1),: 71-75

32. Falchi M, Forabosco P, Mocci E, Cappio Borlino C, Picciau A, Virdis E, Persico I, Parracciani D, **Angius A,** Pirastu M. 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.

33. 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.

34. J Simporè, **A Angius**, I Persico, A Sassu, DA Prodi, S Musumeci. Methylenetatrahydrofolate reductase gene polymorphisms in Burkina Faso. Clin Chim Acta. 2005; 360(1-2): 199-200.

35. R Chillemi, **A Angius**, I Persico, A Sassu, DA Prodi, S Musumeci. Methylenetatrahydrofolate reductase (MTHFR) from Mediterranean to Sub-Saharan Areas. J Biol Sciences 2005: 6 (1): 28-34.

36. M Musumeci, J Simpore, R Barone, **A Angius**, S Musumeci. Synchronic macrophage response and Plasmodium falciparum malaria. J Vect Borne Dis 2006, 43: 84–87.

37. C Fraumene, EMS Belle, L Castrì, S Sanna, G Mancosu, M Cosso, F Marras, G Barbujani, 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.

38. **A Angius**, J Simpore, 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.

39. **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. 2008;65(1):9-22.

40. 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.

41. 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.

42. G Pistis, I Piras, N Pirastu, I Persico, A Sassu, A Picciau, Da Prodi, C Fraumene, E Mocci, T Manias, R Atzeni, M Pirastu, **A Angius**. Extreme differentiation between eight villages in a secluded area of Sardinia revealed by genome-wide high density SNPs analysis. PLoS ONE. 2009;4(2):e4654.

43. L Vago, SK Perna, M Zanussi, B Mazzi, MT Lupo Stanghellini, NF Perelli, F Torri, **A Angius,** B Forno, M Casucci, M Bernardi, J Peccatori, C Corti, M Ferrari, S Rossini, MG Roncarolo, C Bordignon, C Bonin, F Ciceri, K Fleischhauer. 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.

44. 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 Medical Genetics 2009, 10:81.