

# FABIO MARRONI

## BIOGRAPHIC INFORMATION

**Date of Birth:** 01/05/1973.

**Place of birth:** Barga (Lucca)

**Nationality:** Italian.

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## EDUCATION

- **19/07/1999** Degree in Biology, University of Pisa (Italy), 19/07/1999. Mark: 110/110 cum laude
- **08/04/2005** PhD in “Experimental and molecular oncology”, Department of Oncology, University of Pisa. (Tutor: Silvano Presciuttini). Degree topic: “La predizione dello stato di portatore di mutazioni germinali in BRCA1 e BRCA2 - Valutazione dei modelli esistenti, stima delle penetranze, ed elaborazione di un modello adattato alle popolazioni italiane” (“Predicting carrier status for mutations in BRCA1 and BRCA2 - Evaluation of existing models, penetrance estimation, and development of a model tailored for the Italian populations”).

## PUBLICATIONS

### Peer-Reviewed Papers

1. Marroni F., Cipollini G., Peissel B., D’Andrea E., Pensabene M., Radice P., Caligo M.A., Presciuttini S., Bevilacqua G.: **Reconstructing the genealogy of a BRCA1 founder mutation by phylogenetic analysis.** *Annals of Human Genetics* **2008**; 72(Pt 3):310-8.
2. Marroni F, Grazio D, Pattaro C, Devoto M, Pramstaller P. **Estimates of Genetic and Environmental Contribution to 43 Quantitative Traits Support Sharing of a Homogeneous Environment in an Isolated Population from South Tyrol, Italy.** *Hum Hered.* **2008**;65(3):175-182.
3. Pattaro C, Marroni F, Riegler A, Mascalzoni D, Pichler I, Volpato CB, Dal Cero U, De Grandi A, Egger C, Eisendle A, Fuchsberger C, Gogele M, Pedrotti S, Pinggera GK, Stefanov SA, Vogl FD, Wiedermann CJ, Meitinger T, Pramstaller PP: **The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives.** *BMC Med Genet.* **2007** Jun 5;8(1):29
4. Marroni F, Pichler I, De Grandi A, Beu Volpato C, Vogl FD, Pinggera GK, Bailey-Wilson JE and Pramstaller PP: **Population isolates in South Tyrol and their value for genetic dissection of complex diseases.** *Annals of Human Genetics* **2006** 70 (6), 812-821.
5. Pichler I, Marroni F, Beu Volpato C, Gusella JF, Kleine C, Casari G, De Grandi A, Pramstaller PP: **Linkage Analysis Identifies a Novel Locus for Restless Legs Syndrome on Chromosome 2q in a South Tyrolean Population Isolate.** *American Journal of Human Genetics* **2006** 79(4):716-23.
6. Marroni F, Pastrello C, Benatti P, Torrini M, Barana D, Lucci Cordisco D, Viel A, Mareni C, Oliani C, Genuardi M, Bailey-Wilson JE, Ponz De Leon M, Presciuttini S: **A genetic model for determining MSH2 and MLH1 carrier probabilities based on family history and tumor microsatellite instability.** *Clinical Genetics* **2006** 69(3):254-62.
7. Ciampolini R, Cetica V, Ciani E, Mazzanti E, Fosella X, Marroni F, Biagetti M, Sebastiani C, Papa P, Filippini G, Cianci D, Presciuttini S: **Statistical analysis of individual assignment tests among four cattle breeds using fifteen STR loci** *J. Anim Sci.* **2006** 84: 11-19
8. Marangoni R, Marroni F, Gioffre D, Ghetti F, Colombetti G: **Biological Weighting Function of the UV-B induced impairment of phototaxis in the fresh water ciliate Ophryoglena flava.** *Photochemistry and Photobiology* **2004**; 80(3):408-11.

9. Marroni F, Aretini P, D'Andrea E, Caligo MA, Cortesi L, Viel A, Ricevuto E, Montagna M, Cipollini G, Federico M, Santarosa M, Marchetti P, Bailey-Wilson JE, Bevilacqua G, Parmigiani G and Presciuttini S: **Penetrances of breast and ovarian cancer in a large series of families tested for BRCA1/2 mutations.** *European Journal of Human Genetics* **2004**;12(11):899-906
10. Marroni F, Curcio M, Fornaciari S, Lapi S, Mariotti ML, Scatena F, Presciuttini S: **Micro-geographic variation of HLA A, B and DR haplotype frequencies in Tuscany (Italy): implications for recruitment of bone marrow donors.** *Tissue antigens* **2004**; 64(4): 478-85.
11. Marroni F, Aretini P, D'Andrea E, Caligo MA, Cortesi L, Viel A, Ricevuto E, Montagna M, Cipollini G, Ferrari S, Santarosa M, Bisegna R, Bailey-Wilson JE, Bevilacqua G, Parmigiani G, Presciuttini S: **Evaluation of widely used models for predicting BRCA1 and BRCA2 mutations.** *Journal of Medical Genetics* **2004**;41(4):278-85
12. Aretini P, D'Andrea E, Pasini B, Viel A, Costantini RM, Cortesi L, Ricevuto E, Agata S, Bisegna R, Boiocchi M, Caligo MA, Chieco-Bianchi L, Cipollini G, Crucianelli R, D'Amico C, Federico M, Ghimenti C, De Giacomo C, De Nicolo A, Della Puppa L, Ferrari S, Ficorella C, Iandolo D, Manoukian S, Marchetti P, Marroni F, Menin C, Montagna M, Ottini L, Pensotti V, Pierotti M, Radice P, Santarosa M, Silingardi V, Turchetti D, Bevilacqua G, Presciuttini S: **Different Expressivity of BRCA1 and BRCA2: Analysis of 179 Italian Pedigrees with Identified Mutation.** *Breast Cancer Research and Treatment* **2003**, 81: 71-79.
13. Cadetti L., Marroni F., Marangoni R., Kuhlmann H.-W., Gioffré D., Colombetti G.: **Phototaxis in the ciliated protozoan Ophryoglena flava: dose-effect curves and action spectrum determination.** *J. Photochem. Photobiol. B: Biol.* **57(2000)**, 41-50.

### Peer-reviewed conference proceedings

1. Marroni F, Toni C, Pennato B, Tsai YY, Duggal P, Bailey-Wilson JE, Presciuttini S: **Haplotypic Structure of the X-Chromosome in the COGA Population Sample and the Quality of its Reconstruction by Extant Software Packages.** *BMC Genetics* **2005**, 6 (Suppl 1):S77.
2. Fosella X, Marroni F, Manzoni S, Verzeletti A, De Ferrari F, Cerri N, Presciuttini S: **Assigning individuals to ethnic groups based on 13 STR loci.** *Proceedings of the 20th International ISFG Congress, Arcachon, France, 9-13 September 2003, International Congress Series 1261 (2004)* 59-61.
3. Presciuttini S, Toni C, Marroni F, Spinetti I, Bailey-Wilson JE, Domenici R: **The number of STR markers necessary to resolve relationships in deficiency paternity cases.** *Proceedings of the 20th International ISFG Congress, Arcachon, France, 9-13 September 2003, International Congress Series 1261 (2004)* 541-543.

### Popular science

1. Marroni F: **La misura dell'isolamento.** *Academia* N.42 (EURAC's quarterly science magazine), December 2006.
2. Marroni F: **Viaggi genetici? No grazie.** *Academia* N.45 (EURAC's quarterly science magazine), December 2007.

### PROFESSIONAL EXPERIENCE

ARPAT (Regional Agency for Environment Protection), **Via Vittorio Veneto 27, Pisa, Italy** - Post graduate stage, **September 1999- March 2000**

**Tessa Coop, Italy** - Teacher and advisor within "Progetto Ambiente" sponsored by Regione Toscana, a project of Environmental Education in kindergarten, primary and secondary schools, **February-June 2000**

**CNR (National Research Council), Pisa, Italy** - Research activity at the Biophysics Institute, **2000-2001**

**University of Pisa, Italy** - Collaboration to research activity at Biomedicine Department, **June-December 2001**

**University of Pisa, Italy** - Research activity in the context of a PhD program in Experimental and Molecular Oncology, **January 2002-December 2004**

**NHGRI** (National Human Genome Research Institute), **333 Cassell Drive, 21224, Baltimore, MD, USA** – On site collaborator **January 2005-September 2005**

**EURAC research, Bolzano/Bozen, Italy** – Senior researcher. **June 2005-present**

## **SHORT COLLABORATIONS AS VISITING SCIENTIST**

- October 2003: Johns Hopkins University, Department of Biostatistics, Baltimore, MD, USA (prof. Giovanni Parmigiani)
- October 2004: NHGRI/NIH, Inherited Diseases Research Branch, Baltimore, MD, USA (Dr. Joan Bailey-Wilson)
- June 2006: Medical Research Council, Human Genetics Unit, Western General Hospital, Edinburgh, Scotland, UK (Dr. Veronique Vitart).

## **RESEARCH ACTIVITIES**

### ***QUANTITATIVE GENETICS***

- Estimation of heritability of several quantitative traits. **2006-2007**
- QTL mapping by genome wide linkage and association analysis. **2005-2007**

### ***BIODEMOGRAPHY***

- Evaluation of the common biodemographic parameters (inbreeding by pedigree and by isonymy, isonymic relationships) in Alpine settlements in South Tyrol. **2006-2007**

### ***POPULATION GENETICS***

- Population genetics of isolated sub-populations (microisolates). Assessment of linkage disequilibrium on Xq13.3 in populations from South Tyrol, Italy. **2005-2006**
- Studies on X-chromosome. Investigation of haplotypic structure by use of pairwise and multilocus linkage disequilibrium measures. Comparison of the performance of several programs for estimating haplotypic frequencies (SnPHap, Haplotyper, Haploblock, PHASE, Arlequin). **2004**
- Studies on microgeographic variation of HLA genes and haplotype frequency **2003**

### ***GENETIC EPIDEMIOLOGY OF CANCER***

- Validation of the software CRCAPRO **2005-2006**
- Study of *BRCA1* 1499insA founder mutation: reconstruction of shared haplotype, dating of the common progenitor and reconstruction of the genealogy of mutation carriers by means of software for reconstructing phylogenetic trees. **2004-2006**
- Development of a model predicting individual probability of carrying a *MSH2/MLH1* mutation in HNPCC-like families. **2003-2005**
- Segregation analysis of colorectal cancer for the identification of additional colon cancer susceptibility genes. **2005**
- Estimate of cancer penetrance from genetic test results via a Markov Chain Monte Carlo approach **2003**
- Comparison of the accuracy of several models predicting the probability of carrying a *BRCA1/2* germline mutation (BRCAPRO, Myriad, Couch, Shattuck-Eidens). **2002**
- Use of the software program BRCAPRO in predicting individual probability of carrying *BRCA1/2* mutations, and comparison of obtained results with mutation analysis performed in the collaborating centers of the Italian Consortium for Hereditary Breast/Ovarian cancer. **2002**
- Genome wide linkage analysis in an extended pedigree, with an excess of breast and colon cancer cases: two-, three-point linkage analysis. Multipoint linkage analysis performed on restricted subsets of the original pedigree. **2001-2002**

### ***PHOTOBIOLOGY***

- Studies on the effect of ionic composition of the culture medium on motility and photomotility of *Ophryoglena flava*. **2000-2001**

- Degree dissertation “Risposta e fotorisposta di *Ophryoglena flava* a radiazioni ultraviolette” (“*Ophryoglena flava* responsiveness and photoresponsiveness to UV radiation”). **1999**

#### **WATER QUALITY ASSESSMENT**

- Microbiological analysis of sea- and fresh-water. **1999**
- Evaluation of river pollution using Extended Biotic Index (EBI) **1999**

#### **MEETINGS AND COURSES ATTENDED**

- Participation in Biophysics school organized by SIBPA “Biophysics of ionic channels and channelopathies” – Venice **22-26 January, 2001**.
- IGES **2001** - The Tenth Conference of the International Genetic Epidemiology Society - September 2-4, **2001**, Garmisch-Partenkirchen, Germany.
- Participation in the residential course “Le Biotecnologie in Oncologia” (Biotechnologies in Oncology) – October 6-7, **2003**, Pisa (Italy).
- Pancreatic Cancer **2004** - Advances and Challenges, Organized by The Lustgarten Foundation for Pancreatic Cancer Research and American Association for Cancer Research, June 25-26, **2004**, San Francisco, California.
- Genetic Analysis Workshop 14 - September 7-10, **2004**, Noordwijkerhout, Holland.
- 1st Course in Statistical Genetic Analysis Of Complex Phenotypes March 30th – April 2nd, **2005** Bertinoro di Romagna, Italy
- 2nd Course in Statistical Genetic Analysis Of Complex Phenotypes May 3rd – May 5th, **2006** Bertinoro di Romagna, Italy

#### **AD-HOC REVIEWER FOR:**

Human Mutation, Biological Psychiatry, Journal of Medical Genetics.

#### **COMMUNICATIONS IN INTERNATIONAL MEETINGS**

1. Marroni F, Grazio D, Pattaro C, Devoto M, Pramstaller P. Estimates of Genetic and Environmental Contribution to 43 Quantitative Traits Support Sharing of a Homogeneous Environment in an Isolated Population from South Tyrol, Italy. (*Poster Session*) *57<sup>th</sup> Annual Meeting of The American Society of Human Genetics*. October 23-27, **2007**, San Diego, California, USA.
2. De Grandi A., Taibi B., Marroni F., Pichler I., Pedrotti S., Beu Volpato C., Pramstaller P. P.: Genetic characterization of a family with asymptomatic high serum gamma-glutamyltransferase (GGT) concentration. (*Poster Session*) *56<sup>th</sup> Annual Meeting of The American Society of Human Genetics*. October 9-13, **2006**, New Orleans, Louisiana, USA
3. Cipollini G., Marroni F., Bonatti F., Pasini B., Radice P., Montagna M., Contegiacomo A., Bevilacqua G., Caligo M.A., Presciuttini S.: Reconstructing the genealogy of a BRCA1 founder mutation in Italy. *18<sup>th</sup> meeting of the European Association for Cancer Research*. July 3-7, **2004**, Innsbruck, Austria.
4. Marroni F., Aretini P., Bailey-Wilson J.E., Bevilacqua G., Parmigiani G., Presciuttini S., and the Italian Consortium for Hereditary Breast and Ovarian Cancer: Unbiased estimation of breast and ovarian cancer penetrances in BRCA1/2 mutation carriers using genetic-test results (*Poster Session*) *53<sup>rd</sup> Annual Meeting of The American Society of Human Genetics*, November 4-8, **2003**, Los Angeles, California, USA.
5. Marroni F., Aretini P., Bevilacqua G., Bailey-Wilson J.E., Parmigiani G., Presciuttini S., and the Italian Consortium for Hereditary Breast and Ovarian Cancer: Penetrance Estimation Via Maximization Of Retrospective Likelihood Using Genetic Test Results Of Families Screened For Brca1/2 Mutations (*Poster Session*) *12<sup>th</sup> Annual Meeting of the International Genetic Epidemiology Society*, November 3-4, **2003**, Redondo Beach, California, USA
6. Marroni Fabio on behalf of the Italian consortium for Hereditary Breast and Ovarian Cancer: Estimation of breast and ovarian cancer penetrances via a Markov Chain Monte Carlo algorithm in a large series of Italian families screened for BRCA1/2 germline mutations. *14th General Meeting of the Breast Cancer Linkage Consortium (BCLC) and the International Collaborative Group on Familial Breast and Ovarian Cancer (ICG-FBOC)*, June 2nd-4th **2003**, Madrid (Spain).

7. Aretini P., Aceto G., Agata S., Bevilacqua G., Bianchi F., Bisegna R., Boiocchi M., Caligo M.A., Chieco-Bianchi L., Cipollini G., Cortesi L., D'Amico C., D'Andrea E., Federico M., De Giacomo C., De Nicolo A., Della Puppa L., Ferrari S., Ficorella C., Lombardi G., Marchetti P., Mariani Costantini R., Marroni F., Menin C., Montagna M., Ottini L., Porfiri E., Presciuttini S., Ricevuto E., Rosati S., Santarosa M., Sensi E., Silingardi V., Tancredi M., Turchetti D., Viel A.: Regional clustering of identical BRCA1/2 germline mutations in Italy. (*Poster session*) 14th General Meeting of the Breast Cancer Linkage Consortium (BCLC) and the International Collaborative Group on Familial Breast and Ovarian Cancer (ICG-FBOC), June 2nd-4th 2003, Madrid (Spain).
8. Marroni F., Aretini P., Bailey-Wilson J. E., Parmigiani G., Bevilacqua G., Presciuttini S. and the Italian Consortium for Hereditary Breast/Ovarian Cancer: Performance of different models predicting BRCA-mutation carrier status in 570 Italian families. 52nd Annual Meeting of the American Society of Human Genetics, 15-19 October 2002, Baltimore, Maryland (USA).
9. Marroni F., Aretini P., Bevilacqua G., Presciuttini S. And the Italian Consortium for Hereditary Breast and Ovarian Cancer: Predicting the presence of germline mutations in BRCA genes: a comparison of different approaches. Cancer Genetics Group Spring Meeting, 13-14 May 2002 Manchester (UK)- (Poster Session).
10. Gioffré D., Kuhlmann H.-W., Marangoni R., Marroni F., Colombetti G.: Photosensory response to UV-light of a ciliated protozoan, *Ophryoglena flava* - 27th Annual meeting of the American Society for Photobiology, July 10-15, 1999, Washington, DC

## COMMUNICATIONS IN NATIONAL MEETINGS

1. Marroni F., Benatti P., Viel A., Montera M., Barana D., Lucci Cordisco E., Pedroni M., Torrini M., Pastrello C., Roncucci L., Mareni C., Oliani C., Genuardi M., Ponz de Leon M., Presciuttini S.: Predicting the risk of carrying a mutation in MSH2/MLH1: results of an Italian multicenter study. *III Congresso Nazionale AIFEG – Napoli 11-12 November 2004*
2. Aretini P., Marroni F., Barbuti S., Caligo M.A., Cipollini G., Guidugli L., Lombardi G., Pepe C., Sensi E., Tancredi M., Euhus D., Parmigiani G., Presciuttini S. And Bevilacqua G.: A population specific version of Brcapro and CancerGene: predicting the carrier status of BRCA mutations in Italy. (*Oral Communication*) 46<sup>th</sup> Annual Meeting of The Italian Cancer Society – Pisa, October 24-27, 2004
3. Marroni F., Cipollini G., Bonatti F., Radice P., D'Andrea E., Contegiacomo A., Bevilacqua G., Presciuttini S., Caligo M.A.: Reconstructing the genealogy of a BRCA1 founder mutation in Italy. *VII Congresso Nazionale SIGU – Pisa 13-15 October 2004*
4. Marroni F., Benatti P., Viel A., Montera M., Barana D., Lucci Cordisco E., Pedroni M., Torrini M., Pastrello C., Roncucci L., Mareni C., Oliani C., Genuardi M., Ponz de Leon M., Bevilacqua G., Presciuttini S.: Predicting the risk of carrying a mutation in MSH2/MLH1: results of an Italian multicenter study. *VII Congresso Nazionale SIGU – Pisa 13-15 October 2004*
5. Marroni F., Benatti P., Montera M., Pedroni M., Torrini M., Roncucci L., Mareni C., Ponz de Leon M., Bevilacqua G., Presciuttini S.: An accurate genetic model for predicting the carrier status of mmr-gene mutations. *2° Congresso Nazionale AIFEG – Chieti, 19-20 November 2003*
6. Marroni F., Benatti P., Montera M., Pedroni M., Torrini M., Roncucci L., Mareni C., Ponz de Leon M., Bevilacqua G., Presciuttini S.: Development of a genetic model for predicting the carrier status for MMR-gene mutations. *1° Congresso Nazionale AIFEG (Associazione Italiana per lo studio della familiarità ed ereditarietà dei tumori gastrointestinali) – Florence, 15-16 November 2002*
7. Marroni F., Aretini P., Bailey-Wilson J. E., Parmigiani G., Bevilacqua G., Presciuttini S. and the Italian Consortium for Hereditary Breast/Ovarian Cancer: Estimating genetic parameters of BRCA genes appropriate for Italian populations from genetic test results. *Congresso Italiano di Diagnostica e Patologia Molecolare, Pisa 26-28 September 2002 - (Oral Communication)*
8. Marroni F., Lipkin S., Benatti P., Pedroni M., Bailey-Wilson J.E., Collins F., Ponz de Leon M., and Presciuttini S.: Genome-wide linkage scan in a large family with suspected predisposition to cancer - *Problematiche cliniche e molecolari dei tumori ereditari del colon-retto, della mammella e dell'ovaio, Riunione congiunta del Gruppo Italiano per lo Studio dei tumori Colorettali Ereditari e del Consorzio Italiano per i Tumori Ereditari della mammella e dell'ovaio – AIRC: Verona 15-17 November 2001*
9. Marroni F., Presciuttini S.: Genome-wide linkage scan in a large family with suspected predisposition to cancer - *3° Convegno FISV, 21-25 September 2001, Riva del Garda (TN)*
10. Marangoni R., Cadetti L., Gioffré D., Marroni F., Colombetti G.: Fototassi in *Ophryoglena flava*: curve di dose effetto e determinazione dello spettro d'azione - *SIBPA 2000, XV riunione scientifica plenaria, 23-25 Ottobre 2000, Parma*

11. Marroni F., Marangoni R., Ghetti F., Gioffré D., Colombetti G.: Effetto delle radiazioni UV su *Ophryoglena flava*: dipendenza dalla dose e costruzione della Biological Weighting function - *SIBPA 2000, XV riunione scientifica plenaria, 23-25 Ottobre 2000, Parma*

## **SKILLS**

- Experience with methods for linkage and association mapping of quantitative and qualitative traits.
- Good knowledge of the following operating systems: Windows 95/98/2000/XP, UNIX, Linux
- Good knowledge of common windows software (Word, Excel, Access, Power Point, Front Page, Emacs), and software for data analysis (Sigma Plot, MedCalc).
- Good knowledge of bibliography software (EndNote 6.0-9.0, Reference Manager 10).
- Knowledge of R package for statistical computing and R programming instructions.
- Use of linkage analysis software (Fastlink, GeneHunter, Merlin, Mendel, Simwalk2) and of linkage simulation software (Fast-s-link)
- Use of software for QTL mapping (SOLAR, LOKI).
- Use of software for association mapping (FBAT, SOLAR, PLINK).
- Use of software for population genetics and haplotype frequency inference from population data (Arlequin, PHASE, SnpHap, Haplotyper, Haploblock, Gold)
- Use of software for population genetics and phylogeny (PHYLIP, structure)
- Use of software for checking genotypic data in pedigrees (pedcheck, pedstats, prest, altertest)
- Use of software for family based association mapping (QTDT and FBAT).
- Use of software package CaGene (including BRCA1/2 germline mutation).
- Basic knowledge of S.A.G.E. 3.1 (Statistical Analyses for Genetic Epidemiology)
- Test for linkage in the case of interaction between two loci (basic user of T-linkage)
- Knowledge of Progeny software for pedigree management.
- Reviewed papers for publication in Journal of Medical Genetics, Clinical Genetics and Human Molecular Genetics.
- Good knowledge of English language.
- Basic knowledge of German language.

## **INTERESTS AND HOBBIES**

- Actor in several non-professional acting companies, performing, among others, in Macbeth (W. Shakespeare), The Emperor's New Clothes (H.C. Andersen), Carmina Vini (U. Chiti)
- Author of the storyboard of a short video (5 min), "Pena la morte" realized with the contribution of the Municipality of Barga (Lucca, Italy) in the attempt to open a public discussion on Death Row.
- Theater critic (non professional) for the website [www.teatro.org](http://www.teatro.org).
- Writer of several short stories and poems (in Italian).