

<b>Project Acronym</b>	Mut2Dis
<b>Project Code</b>	PIOF-GA-2009-237225
<b>Project Title</b>	New methods to evaluate the impact of single point protein mutation on human health.
<b>Periodic Report</b>	Outgoing Phase, Sep 2009 – Aug 2011 (24 months)

## DISSEMINATION ACTIVITIES

During the outgoing period EC dedicated part of the time to disseminate the results of this project in international conferences, workshops and in invited seminars in institutions both in US and Europe. To summarize the dissemination activity performed in the last two years, EC published a paper about the results obtained in this study and a review about the personalized challenges in personalized medicine. He also submitted 10 posters to meetings and conferences, 6 of which have been selected for oral presentation. Finally EC was invited to give 12 seminars where he presented the results of the Mut2Dis research project. EC is also maintaining web page where details of there project are made available. In the returning phase, EC is planning to attend important meetings and conferences to advertise the web server for the prediction of deleterious variants that will be developed during the next year. In addition other papers and review are in preparation and we believe that will be published during the second phase. More details about the papers, posters and seminars are given below.

### Published Papers

- Capriotti E, Altman RB. (2011). Improving the prediction of disease-related variants using protein three-dimensional structure. BMC Bioinformatics. 12 (Suppl 4); S3 (IF: 3.028)
- Fernald GH, Capriotti E, Daneshjou R, Karczewski KJ, Altman RB. (2011). Bioinformatics challenges for personalized medicine. Bioinformatics. 27: 1741-1748. (IF: 4.877)

### Accepted Posters

- Capriotti E, Altman RB. (2010) A new disease-specific machine learning approach for the prediction of cancer-related SNPs. XVII international conference on Intelligent Systems for Molecular Biology (ISMB). Boston (USA), July 11-13 2010.
- Capriotti E, Russ B Altman. (2010) Predicting cancer-causing single amino acid polymorphisms using functional annotation. 4th Comprehensive Cancer Research Training Program (CCRTP) at Stanford University, Palo Alto California (USA), September 13-17 2010.

- Capriotti E Bromberg Y, Altman RB (2011). Improving the detection of deleterious mutations integrating the predictions four well-tested methods. XIX international conference on Intelligent Systems for Molecular Biology (ISMB). Wien (Austria) July 17-19 2011.
- Thusberg J, Capriotti E, Auer J, Mooney S (2011). Functional profiling of pharmacogenetics non-synonymous SNPs. XIX international conference on Intelligent Systems for Molecular Biology (ISMB). Wien (Austria) July 17-19 2011.

### **Poster selected for oral presentation**

- Capriotti E\*, Altman RB. (2010) Predicting disease-related single amino acid polymorphisms using protein structure. 9th International Conference on Computational Systems Bioinformatics (CSB2010). Stanford, Palo Alto, California (USA), August 17-18 2010.
- Capriotti E\*, Altman RB. (2010) Improving the prediction of disease-related variants using protein three-dimensional structure. II Workshop on Annotation, Interpretation and Management of Mutations (AIMM) at the European Conference on Computational Biology (ECCB). Ghent, Belgium, September 26-29 2010.
- Capriotti E\*, Altman RB. (2010) Improving the detection of deleterious single amino acid polymorphisms using protein functional and structural information. Exploring the functional consequences of genomic variation (HGVS meeting). Washington DC (USA), November 2 2010.
- Capriotti E\*, (2010). Predicting deleterious mutation from CAGI. Critical Assessment of Genome Interpretation (CAGI). Berkeley, California (USA), 10 December 2010.
- Capriotti E\*, Altman RB. (2011). Predicting deleterious single amino acid polymorphisms using protein functional and structural information. EMBO Young Scientist Forum. Warsaw (Poland), June 30th – July 1st 2011.
- Capriotti E, Fariselli P, Martell PL, Casadio R (2011). SNPs&GO: predicting the deleterious effect of human mutations using functional annotation. XIX international conference on Intelligent Systems for Molecular Biology (ISMB). Wien (Austria) July 17-19 2011.

### **Invited seminars**

- Buck Institute, Novato (California, USA), Predicting the effect of single amino acid polymorphisms. 27 May 2010.

- Luxembourg Centre for System Biomedicine, Luxemburg University, Luxembourg. Computational methods for molecular biology. 02 Jul 2010.
- Department of Genetics and Bioengineering, Yeditepe University, Istanbul (Turkey), Computational methods for molecular biology. 20 Sep 2010.
- Department of Medicinal Chemistry and Molecular Pharmacology, Purdue University, Lafayette (Indiana, USA). Computational methods for molecular biology. 25 Jan 2011.
- Lawrence Berkeley National Laboratory, Berkeley (California, USA). Predicting the effect of Single Amino Acid Polymorphisms. 18 Feb 2011.
- Department of Computer Sciences, Wayne State University, Detroit (Michigan, USA). Computational methods for molecular biology. 21 Mar 2011.
- J. Craig Venter Institute, San Diego (California, USA). Computational methods for molecular biology. 06 Apr 2011.
- Department of Bioengineering, University of Texas at Dallas (Texas, USA). Computational methods for molecular biology. 22 Apr 2011.
- Department of Pathology, University of Alabama at Birmingham (Alabama, USA). Computational methods for molecular biology. 17 May 2011.
- Department of Computer Science, Université Pierre et Marie Curie, Paris (France). Computational methods for molecular biology. 26 May 2011.
- Instituto Gulbenkian de Ciencia, Oeiras (Portugal). Computational methods for molecular biology. 16 Jun 2011.
- Statistics and Genomics Seminar, University of California, Berkeley (California, USA) Computational methods for the prediction of the impact of missense variants. 01 Sep 2011.