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| **Project Acronym** | Mut2Dis |
| **Project Code** | PIOF-GA-2009-237225 |
| **Project Title** | New methods to evaluate the impact of single point protein mutation on human health. |
| **Periodic Report** | Returning Phase, Sep 2011 – Aug 2012 (12 months) |

## DISSEMINATION ACTIVITIES

During the returning 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 during the last year, EC published one paper about the results obtained analyzing cancer-causing missense Single Nucleotide Variants (mSNVs) and another paper about the prediction the deleterious effect of mSNVs detected in a family quartet and two reviews about the future perspective in personal genomics and the use of protein structure information for the detection of mSNVs affecting drug response.
In collaboration with other colleagues, EC also submitted 3 posters to meetings and conferences, 2 of which have been orally presented by collaborators. Finally, EC was invited to give 4 seminars where he presented the results of the Mut2Dis research project. EC is also maintaining web page were details of there project are made available. It is expected that other papers and reviews related to this research project currently in preparation will be published during the next few months. More details about the papers, posters and seminars are given below.

### Published Papers

- **Capriotti E**, Altman RB. (2011). A new disease-specific machine learning approach for the prediction of cancer-causing missense variants. Genomics. 98; 310-317. (IF: 3.327)
- Dewey FE, Chen R, Cordero SP, Ormond KE, Caleshu C, Karczewski KJ, Whirl-Carrillo M, Wheeler MT, Dudley JT, Byrnes JK, Cornejo OE, Knowles JW, Woon M, Sangkuhl K, Gong L, Thorn CF, Hebert JM, **Capriotti E**, David SP, Pavlovic A, West A, Thakuria JV, Ball MP, Zaranek AW, Rehm HL, Church GM, West JS, Bustamante CD, Snyder M, Altman RB, Klein TE, Butte AJ, Ashley EA. (2011). Phased whole-genome genetic risk in a family quartet using a major allele reference sequence. PLOS Genetics 7; e1002280. (IF: 9.543)
- **Capriotti E**, Nehrt NL, Kann MG, Bromberg Y. (2012). Bioinformatics for personal genome interpretation. Briefings in Bioinformatics. 13; 495-512.. (IF: 9.283, Google Citation: 1)

- Lahti JL, Tang GW, **Capriotti E**, Liu T, Altman RB. (2012) Bioinformatics and variability in drug response: a protein structural perspective. Journal of Royal Society Interface. 9: 1409-1437. (IF: 4.259).

**Accepted Posters**

- Emidio Capriotti, Piero Fariselli, Pier Luigi Martelli, Rita Casadio (2012). Predicting the effect of single point mutations on protein stability using evolutionary information. XX international conference on Intelligent Systems for Molecular Biology Long Beach, California (USA), July 15-17 2012.

**Poster selected for oral presentation**

- Chet Seligman, Janita Thusberg*, Emidio Capriotti, Biao Li, Jackson Miller, Jim Auer, Michelle Whirl-Carrillo, Teri Klein, Sean Mooney (2012). Predicting pharmacogenetic protein variants. XX international conference on Intelligent Systems for Molecular Biology Long Beach, California (USA), July 15-17 2012.
- Emidio Capriotti, Maria Silvina Fornasari, Juritz Ezequiel, Pier Luigi Martelli, Piero Fariselli, Rita Casadio, Gustavo Parisi* (2012). Improving the prediction of disease-related variants using protein dynamism. SNP-SIG Meeting 2012, XX international conference on Intelligent Systems for Molecular Biology Long Beach, California (USA), July 14 2012.

**Invited seminars**

- Statistics and Genomics Seminar, University of California, Berkeley (California, USA) Computational methods for the prediction of the impact of missense variants. 1 September 2011.
- Institut de Cancerologie Gustave Roussy, Villejuif (France). Computational methods for the detection of deleterious genetic variants, 4 October 2011
- Karlsruhe Institute of Technology, Karlsruhe (Germany). Computational methods for the detection of deleterious genetic variants, 6 October 2011.
- Bologna Winter School 2012, Bologna (Italy). Predicting the effect of protein variants, 17 February 2012.