Margherita Squillario, PhD

Date of Birth: 15/09/1981
Citizenship: Italian
Mobile:
Email (work):
Email (private):

Links

Linkedin profile:
ResearchGate:
Google Scholar:
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Current Position

Research scholarship – from January 2020 to January 2021, titled "Implementing clinical exome sequencing into the diagnostic workflow of epileptic encephalopathies and exploiting its potential for personalized medicine" (CUP: G56C18000400001)"
U.O.C. Genetics, Gaslini Pediatric Hospital, Genoa

Skill

My major interest concerns the study of complex diseases through the merged analysis of various molecular (e.g., miRNAs or genes expressions, proteins quantifications, DNA variations) and clinical data (e.g., age, survival time, tumor grades, treatment) and the interpretation of the results from both the statistical and biological aspects. I consider a disease complex when it is caused by the alterations of many genes such as in many tumor types and in the neurodegenerative diseases like Alzherimer's and Parkinson's diseases. The methods I mainly use in the context of machine learning analyses are those that enforced the sparsity where the solution of the biological problem it is thought to depend on few variables (e.g., genes, miRNAs, proteins, genetic variables) with respects to the total number of variables. I prefer to solve, whenever it is possible, supervised biological problems where using the labels associated to the samples is allowed and where a robust and possible unbiased statistical model is built after the training, the testing and the validation phase.
I am also able to choose suitable statistical tests to solve easier biological questions in the base statistic scenarios such as in the verification of the statistical significance of the diversity of two or more data distributions.
I usually address the functional interpretation of the results considering the in silico enrichment analysis, using free online tools such as WebGestalt, DAVID, GeneCodis among others. I usually deepen this part of the analysis workflow, using many biological databases (e.g., NCBI, USCS Genome Browser, Uniprot, MGI, GTEx, GO, KEGG, STRING, Phenopedia/Genopedia, BioGrid, miRBase, GnomAD).
I have a good knowledge of Perl and R and a basic knowledge of Python.

Keywords

Bioinformatics, computational biology, high-throughput data analysis (GWAS, RNA-Seq, miRNAs and gene expression), machine learning (mostly supervised) and classic statistical methods, functional interpretation of the results, Alzheimer's Disease.

Work Experience

Research Grant D.R. n. 5075 del 18/10/2018 rectified with D.R. n. 5359 del 31/10/2018 – from January 2019 to December 2019
DIBRIS, University of Genoa

Post-doctoral research fellow – from April 2017 to November 2018
DIBRIS, University of Genoa

Support to the Teaching in the Fundaments of Informatics course (cod. 66052) to the students of Biomedical Engineering in the University of Genoa. A.A. 2017-2018 – I e II semesters.

Research Grant renewal – from December 2016 to November 2017

DIBRIS, University of Genoa

Teaching in the Informatics and Bioinformatics course (cod. 80791-1617) to the bachelor students of Biotechnology in the University of Genoa, A.A. 2016-2017 – I e II semesters.

Research Grant D.R. N. 7355 on 18/9/2015 – from December 2015 to November 2016
DIBRIS, University of Genoa

Co. co. pro – from November 2014 to October 2015
San Martino Hospital , Genoa, Italy.

Research Grant renewal – from October 2013 to September 2014
DIBRIS, University of Genoa

Research Grant D.R. N. 544 on 14/5/2012 – from October 2012 to September 2013
DIBRIS, University of Genoa

Research Grant renewal – from October 2011 to September 2012
DISI (Department of Computer and Information Sciences), University of Genoa

Research Grant D.D.A. N. 581 on 28/6/2010 – from October 2010 to September 2011
DISI, University of Genoa

Research Grant D.R. N. 6855 on 5/10/2009 – from October 2009 to September 2010
University of Verona

Research Fellow at National Instiute of Genetics (NIG) – from April 2006 to May 2009
Laboratory of DNA Data Analysis Center for Information Biology and DNA Data Bank of Japan (DDBJ) in MIshima-shi, Shizuoka-ken, Japan.
During this experience, I worked mainly in these fields: expression pattern analysis (where the expression data come from both qRT-PCR and microarray), comparative analysis (using of Vista Browser), sequence alignments (aminoacidic and nucleotidic by the use of Clustal, Realigner and other alignment programs), transcription factor binding site analysis (TRANSFAC, TESS and others), CpG island analysis and phylogenetic analysis (PHYLIP).

Education

PhD in Informatics at DIBRIS, University of Genoa – from January 2013 to April 2017

Subject of the PhD thesis:
Application of machine learning methods for the analysis of biomedical data and functional characterization of the identified signatures. In particular I strongly used sparse regularization methods. The majority of the published works are focused on the analysis of molecular data in the context of neurodegenerative diseases, such as Alzheimer's and Parkinson's Diseases. The most recent work is focused on the analysis and interpretation of GWAS data in the context of Alzheimer's disease.

Master Degree in Bioinformatics (108/110), University of Milano-Bicocca - from October 2003 to December 2005

Subject of graduation thesis:
Identification and characterization of tissue-specific promoters in Homo Sapiens by Bioinformatics tools (such as artificial neural nets). The aim of this work is to build the most accurate model to predict if a human DNA sequence contains a tissue-specific promoter and the tissue in which the promoter is more active.

University Degree in Biotechnology (106/110), University of Genoa – from October 2000 to September 2003

Title of graduation thesis:
"In silico elaboration and interpretation of experimental microarray data and succeeding analysis of apoptotic process and cell cycle".

Articles

- Cerminara M, Spirito G, Pisciotta L, **Squillario M**, Servetti M, Divizia M.T, Lerone M, Berloco B, Boeri S, Nobili L, Vozzi D, Sanges R, Gustincich S, Puliti A. "Whole Exome Sequencing Revealed Disease-Causing Variants in Two Genes in a Patient With Autism Spectrum Disorder, Intellectual Disability, Hyperactivity, Sleep and Gastrointestinal Disturbances". Provisionally accepted in Front. Genet. doi: 10.3389/fgene.2021.625564.

- Biassoni R, Di Marco E, **Squillario M**, Barla A, Piccolo G, Ugolotti E, Gatti C, Minuto M, Patti G, Maghnie M, d'Annunzio G. "Gut microbiota un newly-diagnosed children and adolescents with type 1 diabetes: machine-learning algorithms to classify microorganisms as disease-linked". J Clin Endocrinol Metab 2020 Sep 1;105(9):dgaa407.

- **Squillario M**, Abate G, Tomasi F, Tozzo V., Barla A, Uberti D "for the Alzheimer's Disease Neuroimaging Initiative". "A telescope GWAS analysis strategy, based on

SNPs-genes-pathways ensamble and on multivariate algorithms, to characterize late onset Alzheimer's disease ", Sci Rep. 2020 Jul 21;10(1):12063.

- Cilloni D, Petiti J, Campia V, Podestà M, **Squillario M**, Montserrat N, Bertaina A, Sabatini F, Carturan S, Berger M, Saglio F, Bandini G, Bonifazi F, Fagioli F, Moretta L, Saglio G, Verri A, Barla A, Locatelli F, Frassoni F. "Transplantation Induces Profound Changes in the Transcriptional Asset of Hematopoietic Stem Cells: Identification of Specific Signatures Using Machine Learning". J Clin Med 2020 Jun 1;9(6):1670

- Colombo M, Bagnara D, Reverberi D, Matis S, Cardillo M, Massara R, Mastracci L, Ravetti J L, Agnelli L, Neri A, Mazzocco M, **Squillario M**, Mazzarello AN, Cutrona G, Agathangelidis A, Stamatopoulos K, Ferrarini M, Fais F. "Tracing CLL-biased stereotyped immunoglobulin gene rearrangements in normal B cell subsets using a high-throughput immunogenetic approach." Mol Med 2020 Mar 10;26(1):25.

- Pesce S, Belagrano V, Greppi M, Carlomagno S, **Squillario M**, Barla A, Della Chiesa M, Di Domenico S, Mavillo D, Moretta L, Candiani S, Sivori S, De Cian F, Marcenaro E." Different Features of Tumor-Associated NK Cells in Patients With Low-Grade or High-Grade Peritoneal Carcinomatosis.", Front Immunol. 2019 Aug 21;10:1963.

- Pesce S, **Squillario M**, Greppi M, Loiacono F, Moretta L, Moretta A, Sivori S, Castagnola P, Barla A, Candiani S, Marcenaro E." New miRNA Signature Heralds Human NK Cell Subsets at Different Maturation Steps: Involvement of miR-146a-5p in the Regulation of KIR Expression.", Front Immunol. 2018 Oct 15;9:2360.

- Guo S, Lai C, Wu C, Cen G; Alzheimer's Disease Neuroimaging Initiative (ADNI)."Conversion Discriminative Analysis on Mild Cognitive Impairment Using Multiple Cortical Features from MR Images", Front Aging Neurosci. 2017 May 18;9:146

- **Squillario M**, Barbieri M, Verri A and Barla A. "Enhancing Interpretability of Gene Signatures with Prior Biological Knowledge", Microarrays, 2016 June8: 5(2).

- Bagnara D, **Squillario M**, Kipling D, Mora T, Walczak AM, Da Silva L, Weller S, Dunn-Walters DK, Weill JC, Reynaud CA. "A Reassessment of IgM Memory Subsets in Humans", J Immunol. 2015 Oct 15; 195(8):3716-24.

- Scala M, Lenarduzzi S, Spagnolo F, Trapasso M, Ottonello C, Muraglia A, Barla A, **Squillario M** and Strada P. "Regenerative Medicine for the Treatment of Teno-desmic Injuries of the Equine. A Series of 150 Horses Treated with Platelet-derived Growth Factors", In vivo (Athens, Greece), 28 (6), pp.1119--1123, 2014.

- Zycinsky G, Barla A, **Squillario M**, Sanavia T, Di Camillo B, Verri A. "Knowledge Driven Variable Selection (KDVS) – a new approach to enrichment analysis of

gene signatures obtained from high-throughput data", Source Code Biol Med., 2013 Jan 9; 8(1).

- Di Camillo B, Sanavia T, Martini M, Jurman G, Sambo F, Barla A, **Squillario M**, Furlanello C, Toffolo G, Cobelli C. "Effect of size and heterogeneity of samples on biomarker discovery: synthetic and real data assessment", PloS One, 2012, 7(3)

-**Squillario M** and Annalisa B. "A computational procedure for functional characterization of a potential marker genes from molecular data: Alzheimer's as a case study", BMC Med. Genomics, 2011 July 5; 4:55.

Papers & Abstract in preparation

- Biassoni R, Di Marco E, **Squillario M**, Ugolotti E, Mosconi M, Faticato MG, Mattioli G, Avanzini S, Pini Prato A."Pathways and microbiome modifications related to surgery and enterocolitis in Hirschsprung disease". [PAPER]

- Cerminara M. Servetti M. Squillario M, Pisciotta L, Spirito G, Divizia M.T, Lerone M, De Grandis E, Boeri S, Nobili L, Vozzi D, Sanges R, Zara F, Gustincich S, Puliti A. "Complex cases with Autism Spectrum Disorder (ASD), developmental delay, hyperactivity and sleep disturbances explained by oligogenic mechanisms". Abstract sent to the European Society of Human Genetics (ESHG) 2021.

- Avenoso D, **Squillario M**, Barla A, Verri A, Carminati E, Nurra C, Todisco E, Gigli F, Bertolini F, Gregato G, Poletti C, Miglino M. "Epigenetic Regulation In Myelodysplastic Syndrome As A Consequence Of A MicroRNA Dysregulation, A Different Perspective With Integration Of Machine Learning For Comprehension Of Pathogenesis And Disease Progression". [PAPER]

Posters

- **Squillario M**, Tomasi F, Tozzo V, Barla A, Uberti D. "Uncovering the genetic hereditable component of late onset Alzheimer's Disease Through the analysis of GWA data". Poster presentation in AD/PD 2019 Lisbon, Portugal, 26 March – 31 March. Proceedings in NeuroDegenerative Diseases will appear soon.

- **Squillario M**, Tomasi F, Uberti D, Barla A. "An integrated approach to uncover in an unbiased way a robust SNP signature characterizing Alzheimer's disease". Presentation to AAT-AD/PD 2018 Turin, Italy, 15-18 March. Proceedings in NeuroDegenerative Diseases 2018.

- **Squillario M**, Barbieri M, Fiorini S, Barla A. "Uncovering Alzheimer's SNP signature with a multi-view machine learning analysis based on SNPs, genes and pathways", Poster presentation in AD/PD 2017 Vienna, Austria, March 29 – April 2. Proceedings in NeuroDegenerative Diseases 2017; 17(suppl 1): 591-1890 – Page 1052.

- Fiorini S, Tomasi F, **Squillario M** and Barla A. "Adenine: a HPC-oriented tool for biological data exploration", CIBB2017 Cagliari, Italy. Short paper in LNBI/LNCS volume 10834.

- Tomasi F, **Squillario M**, Verri A, Bagnara D and Barla A. "ICING: large-scale inference of immunoglobulin clonotypes", CIBB2017 Cagliari, Italy. Short paper in LNBI/LNCS volume 10834.

- **Squillario M**, Zycinski G, Masecchia S, Verri A, Barla A. "Gene signature identification and validation in the analysis of a Parkinson gene expression dataset", AD/PD 2013 Florence, Italy, 5-10 March. Proceedings in NeuroDegenerative Diseases 2014, Vol 13, N 2-3, ISBN: 978-3-318-02511-8, e-ISBN: 978-3-318-02512-5.

- Vaccari C, Musante I, Gimelli G, Tassano E, Lerone M, Divizia M T, Torre M, Romanini M V, Bedeschi M F, Catena N, Senes F M, Calevo M G, Valle M, Acquaviva M, Baban A, Barla A, **Squillario M**, Ravazzolo R, and Puliti A. "Towards the identification of genetic mechanisms underlying Poland Anomaly", European Human Genetics Conference, 2013.

- Masecchia S, Barla A, **Squillario M**, Coco S, Verri A and Tonini G P."Inferring oncogenetic tree-models from aCGH of metastatic neuroblastoma", ECCB European Conference on Computational Biology, Basel, Switzerland, 9-12 September, 2012.

- **Squillario M**, Masecchia S, Zycinski G, Barla A. "Uncovering Candidate Biomarkers for Alzheimer's and Parkison's Diseases with Regularization Methods and Prior Knowledge", AD/PD 2011, Barcelona, Spain, 9-13 March. Proceedings in NeuroDegenerative Diseases 2012, Volume 10, N 1-4, ISBN: 978-3-318-02172-1, e-ISBN: 978-3-318-02173-8.

- Zycinsky G, **Squillario M,** Barla A, Sanavia T, Verri A, Di Camillo B. "Discriminant functional groups identification with machine learning and prior knowledge", ESANN 2012 proceedings, European Symposium on Artificial Neural Networks, Computational Intelligence and Machine Learning. Bruges (Belgium), 25-27 April 2012, i6doc.com publ., ISBN 978-2-87419-049-0.

- **Squillario M**, Zycinski G, Masecchia S, Verri A, Barla A. "Analysis of a Parkinson dataset: comparison between KDVS and the Standard pipeline", European Conference on Computational Biology, ECCB 2012, Basel, Switzerland, 9-12 September.

- **Squillario M**, Malattia C, Basso C, Barla A. "A molecular based subtyping in juvenile idiopathic arthritis (JIA) ",19th Annual ECCB/ISMB 2011- International Conference on Intelligent Systems for Molecular Biology and 10th European Conference on Computational Biology, Vienna, 17-19 July.

- Barla A, Jurman G, Visintainer R, **Squillario M**, Filosi M, Riccadonna S, Furlanello C. "A machine learning pipeline for discriminant pathways

identification". Proceedings in CIBB2011, ISBN online 9788890643705. Gargnano, Italy.

- Barla A, Jurman G, Visintainer R, **Squillario M**, Filosi M, Riccadonna S, Furlanello C. "A machine learning pipeline for discriminant pathways identification", Short paper in LNBI/LNCS, 2011.

- Barla A, Masecchia S, **Squillario M**. "L1-L2 regularization framework for Alzheimer's molecular characterization", ISMB 2010- 18th Annual International Conference on Intelligent Systems for Molecular Biology, Jul 10, 2010 - Jul 14, Boston, USA.

- Di Camillo B, Martini M, Sanavia T, Jurman G, Sambo F, Barla A, **Squillario M**, Furlanello C, Toffolo G, Cobelli C. "Effect of size and heterogeneity of samples on biomarker discovery: synthetic and real data assessment", ECCB 2010, European Conference on Computational Biology, Ghent, Belgium, 26-29 September.

- **Squillario M**, Masecchia S, Barla A. "Functional characterization of Parkinson by high-throughput data analysis with l1-l2 regularization", ECCB 2010, European Conference on Computational Biology, Ghent, Belgium, 26-29 September.

Book chapters

- Barla A, Jurman G, Visintainer R, **Squillario M**, Filosi M, Riccadonna S, Furlanello C "A machine learning pipeline for Identification of Discriminat Pathways", Editor: N. Kasabov. Springer Handbook of Bio- and Neuroinformatics, April 2014.