

## GIUSEPPE NARZISI PH.D.

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### WORK EXPERIENCE & POSITIONS HELD

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- May 2014 – present **Lead Bioinformatics Scientist, New York Genome Center, New York, NY.**  
I lead the Constitutional Genomics group at NYGC, which is a team of Scientists, Programmers, and Engineers conducting genomics research and methods development at the center.
- Jan 2017 – May 2017 **Adjunct Faculty, NYU-ePoly Online Graduate Program, NYU Tandon School of Engineering.**  
Instructor for the “Next Generation Sequence Analysis” course.
- Oct 2011 – Apr 2014 **Computational Science Analyst, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY.**
- Developed an accurate DNA sequence assembly pipeline to detect genetic mutation in thousands of human exomes for Illumina sequencing technology. Applications include analysis of genetic variations in human genomes to identify the genetic causes of various human diseases (autism and cancer).
  - Achieved superior sensitivity and one order of magnitude more specificity to detect large mutations compared to state-of-the-art pipelines.
  - Analyzed exome-capture data of ~1300 families (~5200 individuals) from the CSHL Simons Simplex Collection. The study highlights a higher rate of *de novo* disrupting mutations in children affected with autism.
- Jun 2011 – Sept 2011 **Assistant Research Scientist, Courant Institute of Mathematical Sciences, New York University.**
- Designed and implemented novel algorithms and software solutions for base-calling and assembling of next-generation DNA sequencing reads.
  - Managed a team of 3 junior researchers to develop and extend our sequence assembly platform using advanced data-structures and algorithms to achieve 10-fold reduction in size while increasing speed.
- Oct 2005 – Aug 2007 **Junior Research Scientist, NYU Bioinformatics Group, Courant Institute of Mathematical Sciences.**
- Team-leader and primary collaborator for the Large-Scale Emergency Readiness (LaSER) project at the NYU Center for Catastrophe Preparedness & Response.
  - Developed and implemented a powerful large-scale computational multi-agent system to simulate the complex dynamics of thousands of agents in urban areas.

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### EDUCATION

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- Sept 2007 - May 2011 **Courant Institute of Mathematical Sciences, New York University, New York, NY.**  
Ph.D. in Computer Science (GPA 3.919/4.0).  
Dissertation titled "*Scoring-and-Unfolding Trimmed Tree Assembler: Algorithms for Assembling Genome Sequences Accurately and Efficiently*".  
Advisor: Prof. Bud Mishra.
- May 2005 - May 2008 **Department of Mathematics & Computer Science, University of Catania, Italy.**  
Ph.D. in Computer Science  
Dissertation titled "*Optimization and Tradeoffs in Protein Structure Prediction*".  
Advisors: Prof. Vincenzo Cutello and Prof. Giuseppe Nicosia.
- Sept 1999 - Jul 2004 **Department of Mathematics and Computer Science, University of Catania, Italy.**  
“Laurea” Degree (Bachelor) in Computer Science (Grade 110/110 cum laude, equivalent GPA 4.0).  
Dissertation titled "*Prediction of Native Protein Structures using Multi-objective Immune Algorithms*".  
Advisors: Prof. Vincenzo Cutello and Prof. Giuseppe Nicosia.

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### SOFTWARE PROJECTS

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- Lancet  
Lancet uses a localized micro-assembly strategy to detect somatic mutation (SNVs and indels) with high sensitivity and accuracy on a tumor/normal pair. Lancet is based on the colored de Bruijn graph assembly paradigm where tumor and normal reads are jointly analyzed within the same graph. On-the-fly repeat composition analysis and self-tuning k-mer strategy are used together to increase specificity in regions characterized by low complexity sequences. [Implemented in C++].  
URL: <https://github.com/nygenome/lancet>
- Scalpel  
Developed an accurate DNA sequence micro-assembly pipeline, Scalpel, for detection of *de novo* mutations (SNPs, insertion, deletions) within whole-exome capture data. The tool has been used to discover candidate disease-linked genetic mutations in thousands of families from the CSHL autism project. [Implemented in C++ and Perl]  
URL: <http://scalpel.sourceforge.net>

FRCurve	Designed and implemented a novel metric to compare and validate DNA sequence assemblers. The metric has been used to rank the performance of different assemblers that participated to the worldwide <a href="#">Assemblathon</a> competitions. [Implemented in C++] URL: <a href="http://sourceforge.net/apps/mediawiki/amos/index.php?title=FRCurve">http://sourceforge.net/apps/mediawiki/amos/index.php?title=FRCurve</a>
SUTTA	Designed and implemented novel algorithms and software solutions for the assembly of next-generation DNA sequencing data using combinatorial optimization techniques (Branch-and-Bound search method) to achieve higher accuracy than state-of-the-art methods. [Implemented in C++] URL: <a href="http://bioinformatics.nyu.edu/wordpress/projects/sutta/">http://bioinformatics.nyu.edu/wordpress/projects/sutta/</a>
PLAN C	Designed and implemented a scalable hybrid-agent simulation tool for modeling, analyzing and planning against catastrophic scenarios in the New York City urban area. The tool enables automatic generation of Pareto optimal plans satisfying multiple objectives. [Implemented in Java and C++] URL: <a href="http://www.nyu.edu/ccpr/laser/plancinfo.html">http://www.nyu.edu/ccpr/laser/plancinfo.html</a>
I-PAES	Developed and implemented a new tool to predict the 3D structure of proteins starting from the sequence of amino acids. I-PAES has been ranked among the best state-of-the-art folding algorithm in a recent Journal of the Royal Society Interface (G. Helles, 2008). [Implemented in C]

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#### ACADEMIC AND TEACHING EXPERIENCE

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Jan 2017 – May 2017	<b>Instructor NYU-ePoly Tandon School of Engineering</b> Held lecture and practical session for the course “Next Generation Sequence Analysis”.
Sep 2016 & Apr 2017	<b>Instructor for the NYGC Sequencing Informatics Workshop</b> Held lecture and practical session on detecting genetic variants through local assembly.
Jan 2009 – May 2009	<b>Teaching Assistant at Courant Institute of Mathematical Sciences, New York University.</b> Held recitation sections and office hours for the course “Data Structures”.
May 2009 – Aug 2009	<b>Teaching Assistant at Courant Institute of Mathematical Sciences, New York University.</b> Held recitation sections and office hours for the course “Programming Languages”.

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#### SPECIALTIES

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Algorithm design and development, Sequence Assembly, Sequence alignment, Variant calling, Next-Generation Sequencing, High Performance Computing, Human Genetics.

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#### COMPUTER SKILLS

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- ▣ Programming Languages: experience with C / C++, Perl and Java.
- ▣ Systems: Unix and Macintosh

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#### AWARDS, FELLOWSHIPS, AND MEMBERSHIPS

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- ▣ Advisory Board Member for the Bioinformatics Master program at the NYU Polytechnic School of Engineering.
- ▣ Review Editor, Editorial Board of Bioinformatics and Computational Biology, a specialty of Frontiers in Bioengineering and Biotechnology and Genetics
- ▣ Associate Faculty Member for F1000 reviews.
- ▣ IBM PhD Fellowship award for the 2010-2011 academic year.
- ▣ Best Paper Award at the Int. Conference on Biosciences – BIOINFO, 2010.
- ▣ “McCracken” award - Guaranteed Financial Support 2007-2011, Ph.D. Program, NYU.
- ▣ “ARCHIMEDE” award from Department of Mathematics and Computer Science, University of Catania, Italy, 2003-2004.

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#### SELECTED PUBLICATIONS

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- Narzisi G**, Corvelo A, Arora K, Bergmann E, Shah M, Musunuri R, Emde AK, Robine N, Vacic V, Zody MC. *Genome-wide somatic variant calling using localized colored de Bruijn graphs*. **Communications Biology**, Nature Research publishing, volume 1, Article number: 20, 2018 (DOI: [10.1038/s42003-018-0023-9](https://doi.org/10.1038/s42003-018-0023-9)). Also available at **CSHL bioRxiv** 196311; (DOI: [10.1101/196311](https://doi.org/10.1101/196311)).
- Fan P.D., **Narzisi G**., Jayaprakash A.D., Venturini E., Robine N., Smibert P., Germer S., Yu H.A., Jordan E.J., Paik P.K., Janjigian Y.Y., Chافت J.E., Wang L., Jungbluth A.A., Middha S., Spraggon L., Qiao H., Lovly C.M., Kris M.G., Riely G.J., Politi K., Varmus H., Ladanyi M. *YES1 amplification is a mechanism of acquired resistance to EGFR inhibitors identified by transposon mutagenesis and clinical genomics*. **Proceedings of the National Academy of Sciences**, Jun 2018, 201717782 (DOI: [10.1073/pnas.1717782115](https://doi.org/10.1073/pnas.1717782115)) Also available at **CSHL bioRxiv** 275974 (DOI: [10.1101/275974](https://doi.org/10.1101/275974)).
- Dolzhenko E., van Vugt J.J.F.A., Shaw R.J., Bekritsky M.A., van Blitterswijk M., **Narzisi G**., Ajay S.S., Rajan V., Kingsbury Z., Humphray S.J., Schellevis R.D., Brands W.J., Baker M., Rademakers R., Kooyman M., Tazelaar G.H.P., van Es M.A., McLaughlin R, Sproviero W., Shatunov A., Jones A., Khleifat A.A., Pittman A., Morgan S., Hardiman O., Al-Chalabi A., Shaw C., Smith B., Neo E.J., Morrison K., Shaw P.J., Reeves C., Winterkorn L., Wexler N.S., The US-Venezuela Collaborative Research Group, Housman D.E., Ng C., Li A., Taft R.J., van den Berg L.H., Bentley D.R., Veldink J.H., Eberle M.A. *Detection of long repeat expansions from PCR-free whole-genome sequence data*. **Genome Research** September 8 2017. (DOI: [10.1101/gr.225672.117](https://doi.org/10.1101/gr.225672.117))

Also available at CSHL bioRxiv (DOI: [10.1101/093831](https://doi.org/10.1101/093831)).

4. Fang H., Bergmann E.A., Arora K., Vacic V., Zody M.C., Iossifov I., O'Rawe J.A., Wu Y., Jimenez Barron L.T., Rosenbaum J., Ronemus M., Lee Y., Wang Z., Dikoglu E., Jobanputra V., Lyon G.J., Wigler M., Schatz M.C. & Narzisi G. *Indel variant analysis of short-read sequencing data with Scalpel*. **Nature Protocols** 11, 2529–2548, 2016. (DOI: [10.1038/nprot.2016.150](https://doi.org/10.1038/nprot.2016.150))
5. Narzisi G. and Schatz M.C. *The challenge of small-scale repeats for indel discovery*. **Front. Bioeng. Biotechnol.** (2015) (DOI: [10.3389/fbioe.2015.00008](https://doi.org/10.3389/fbioe.2015.00008)).
6. Narzisi G., O'Rawe J.A., Iossifov I., Fang H., Lee Y., Wang Z., Wu Y., Lyon G.J., Wigler M., Schatz M.C. *Accurate de novo and transmitted indel detection in exome-capture data using microassembly*. **Nature Methods** 11, 1033–1036 (2014) (DOI: [10.1038/nmeth.3069](https://doi.org/10.1038/nmeth.3069)).
7. Fang H., Narzisi G., O'Rawe J., Wu Y., Rosenbaum J., Ronemus M., Iossifov I., Schatz M.C., Lyon G.J. Reducing INDEL calling errors in whole-genome and exome sequencing data. **Genome Medicine** 2014, 6:89 (DOI: [10.1186/s13073-014-0089-z](https://doi.org/10.1186/s13073-014-0089-z)).
8. Iossifov I., O'Roak B.J., Sanders S.J., Ronemus M., Krumm N., Levy D., Stessman H.A., Witherspoon K.T., Vives L., Patterson K.E., Smith J.D., Paepfer B., Nickerson D.A., Dea J., Dong S., Gonzalez L.E., Mandell J.D., Mane S.M., Murtha M.T., Sullivan C.A., Walker M.F., Waqar Z., Wei L., Willsey A.J., Yamrom B., Lee Y., Grabowska E., Dalkic E., Wang Z., Marks S., Andrews P., Leotta A., Kendall J., Hakker I., Rosenbaum J., Ma B., Rodgers L., Troge J., Narzisi G., Yoon S., Schatz M.C., Ye K., McCombie W.R., Shendure J., Eichler E.E., State M.W. & Wigler M. The contribution of de novo coding mutations to autism spectrum disorder. **Nature** (2014) (DOI: [10.1038/nature13908](https://doi.org/10.1038/nature13908))
9. Iossifov I., Ronemus M., Levy D., Wang Z., Hakker I., Rosenbaum J., Yamrom B., Lee Y., Narzisi G., Leotta A., Kendall J., Grabowska E., Ma B., Marks S., Rodgers L., Stepansky A., Troge J., Andrews P., Bekritsky M., Pradhan K., Ghiban E., Kramer M., Parla J., Demeter R., Fulton L.L., Fulton R.S., Magrini V.J., Ye K., Darnell J.C., Darnell R.B., Mardis E.R., Wilson R.K., Schatz M.C., McCombie R.W., Wigler M. *De Novo Gene Disruptions in Children on the Autistic Spectrum*. **Neuron**, Vol. 74, Issue 2, pp. 285-299, 26 April 2012 (DOI: [10.1016/j.neuron.2012.04.009](https://doi.org/10.1016/j.neuron.2012.04.009)).
10. Narzisi G. and Mishra B.: *Comparing De Novo Genome Assembly: The Long and Short of It*. **PLoS ONE**, 6(4):e19175. April 2011. (DOI: [10.1371/journal.pone.0019175](https://doi.org/10.1371/journal.pone.0019175)).
11. Narzisi G. and Mishra B.: *Scoring-and-Unfolding Trimmed Tree Assembler: Concepts, Constructs and Comparisons*. **Bioinformatics**, Oxford Journals, 2010 (DOI: [10.1093/bioinformatics/btq646](https://doi.org/10.1093/bioinformatics/btq646)).
12. Menges F., Narzisi G. and Mishra B. *TotalReCaller: Improved Accuracy and Performance via Integrated Alignment & Base-Calling*. **Bioinformatics**, Oxford Journals, 2011 (DOI: [10.1093/bioinformatics/btr393](https://doi.org/10.1093/bioinformatics/btr393)).
13. Smith S.W., Portelli I., Narzisi G., Nelson L.S., Menges F., Rekow E.D., Mincer J.S., Mishra B., Goldfrank L.R.: *A Novel Approach to Multi-Hazard Modeling and Simulation*. **Disaster Medicine and Public Health Preparedness (DMPHP), The American Medical Association (AMA)**, 2009 (DOI: [10.1097/DMP.0b013e3181a88899](https://doi.org/10.1097/DMP.0b013e3181a88899)).
14. Cutello V., Narzisi G., Nicosia G.: *A Multi-Objective Evolutionary Approach to the Protein Structure Prediction Problem*. **Journal of the Royal Society Interface**, Royal Society Publications London, vol. 3(6), pp. 139-151, 2006 (DOI: [10.1098/rsif.2005.0083](https://doi.org/10.1098/rsif.2005.0083)).

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#### PATENTS

1. **System, Method, and Computer-Accessible Medium for Providing a Multi-Objective Evolutionary Optimization of Agent-Based Models**. (Co-Inventors: B. Mishra and M.P. Venkatesh) Filed: September 2007.
2. **Methods, Computer-Accessible Medium, and Systems for Score-Driven Whole-Genome Shotgun Sequence Assembly**. (Co-Inventor: B. Mishra) Filed: February 2009.
3. **Methods, Computer-Accessible Medium, and Systems for Base-Calling and Alignment**. (Co-Inventor: B. Mishra) Filed: April 2009.

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#### HOBBIES

▣ Rock Climbing, Snowboarding, Hiking, Biking, Electric Guitar.