IVAN IOSSIFOV, Ph.D.

1 Bungtown Road / Cold Spring Harbor, NY 11724
Telephone: +1 (516) 367-6947
E-mail: iossifov@cshl.edu

EDUCATION

2004–2008 **Columbia University,** *Ph.D.* Department of Biomedical Informatics
Advisor: Prof. Andrey Rzhetsky GPA: 4.1/4.0 (*with distinction*)
Thesis: Studying human disease with molecular network models derived from scientific literature

1992–1997 **Sofia University**, Bulgaria, *BS* and *MS* in Computer Science
Advisor: Prof. Darina Dicheva GPA: 5.9/6.0
Thesis: A method for agent-based visualization in information systems

WORK EXPERIENCE

2017– Associate Prof., *Cold Spring Harbor Laboratory*, Cold Spring Harbor, NY

2015– Core Member, Assistant Investigator, *New York Genome Center*, NYC, NY

2012–2017 Assistant Prof., *Cold Spring Harbor Laboratory*, Cold Spring Harbor, NY

2008–2012 QB Fellow, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

2006 Summer Internship at Microsoft Research, Redmond, WA

2001–2004 Senior Programmer/Analyst, Department of Biomedical Informatics, Columbia University, New York

2000–2001 Team Leader, *Nemetschek LLC*, Sofia, Bulgaria

1994–1999 Senior Application Developer, *DISY Inc*. Sofia, Bulgaria

REASEARCH INTEREST

genetics of autism ▪ human genetics ▪ human disease ▪ new generation sequencing
computational biology ▪ molecular networks ▪ applied statistical and machine learning
biomedical text-mining ▪ molecular evolution

PRIMARY PUBLICATIONS

1. Munoz, A., Yamrom, B., Lee, Y.-H., Andrews, P., Marks, M., Lin, K. T., Wang, Z., Krainer, A. R., Darnell, B., Wigler, M., & **Iossifov, I.** De novo indels within introns contribute to ASD incidence (2017) bioRxiv; doi:10.1101/137471

2. Buja, A., Volfovsky, N., Kreiger, A., Lord, C., Lash, A., Wigler, M., **Iossifov, I.** Damaging Mutations are Associated with Diminished Motor Skills and IQ in Children on the Autism Spectrum (2017) bioRxiv; doi:10.1101/141200

3. Andrews, P. A., **Iossifov, I.**, Kendall, J., Marks, S., Muthuswamy, L., Wang, Z., Levy, D. & Wigler, M. MUMdex: MUM-based structural variation detection (2016) bioRxiv; doi:10.1101/078261

4. Ye, K., **Iossifov, I.**, Levy, D., Yamrom, B., Buja, A., Krieger, A., & Wigler, M. Measuring shared variants in cohorts of discordant siblings with applications to autism (2017) *PNAS* doi: 10.1073/pnas.1700439114

5. Fang, H., Bergmann, E.. A., Arora, K., Vacic, V., Zody, M. C., **Iossifov, I.**, O'Rawe, J. A., Wu, Y., Jiménez-Barrón, L. T., Rosenbaum, J., Ronemus, M., Lee, Y. H., 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 (2016) *Nat. Prot.* **11** (12) 2529

6. Turner, T. N., Hormozdiari, F., Duyzend, M. H., McClymont, S. A., Hook, P. W., **Iossifov**, **I.,** Raja, A., Baker, C., Hoekzema, K., Stessman, H. A., Zody, M. C., Nelson, B. J., Huddleston, J., Sandstrom, R., Smith, J. D., Hanna, D., Swanson, J. M., Faustman, E. M., Bamshad, M. J., Stamatoyannopoulos, J., Nickerson, D. A., McCallion, A. S., Darnell, R. & Eichler E. E. Genome sequencing of autism families reveals disruption of putative noncoding regulatory DNA. (2016) *Am. J. Hum. Genet*. **98** (1) 58

7. **Iossifov, I.**, Levy, D., Allen, J., Ye, K., Ronemus, M., Lee, Y.-H., Yamrom, B., & Wigler, M. Low load for disruptive mutations in autism genes and their biased transmission. (2015) *PNAS* **112** (41), E5600–7

8. Homsy, J., Zaidi, S., Shen, Y., Ware, J. S., Samocha, K. E., Karczewski, K. J., DePalma, S. R., McKean, D., Wakimoto, H., Gorham, J., Jin, S. C., Deanfield, J., Giardini, A., Porter, G. A., Kim, R., Bilguvar, K., Lopez-Giraldez, F., Tikhonova, I., Mane, S., Romano-Adesman, A., Qi, H., Vardarajan, B., Ma, L., Daly, M., Roberts, A. E., Russell, M. W., Mital, S., Newburger, J. W., Gaynor, J. W., Breitbart, R. E., **Iossifov**, **I.**, Ronemus, M., Sanders, S. J., Kaltman, J. R., Seidman, J. G., Brueckner, M., Gelb, B. D., Goldmuntz, E., Lifton, R. P., Seidman, C. E., & Chung, W. K. De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. (2015) *Science* **350** (6265) 1262–1266

9. Jiménez-Barrón, L. T., O'Rawe, J. A., Wu, Y., Yoon, M., Fang, H., **Iossifov**, **I.** & Lyon, G. J. Genome-wide variant analysis of simplex autism families with an integrative clinical-bioinformatics pipeline. (2015) *Mol. Case Stud* **1**, (1), a000422

10. **Iossifov, I**.*, O'Roak, B.J.*, Sanders, S.J.*, Ronemus, M.*, Krumm, N., Levy, D., Stessman, H.A., Witherspoon, ., Vives, L., Patterson, K.E., Smith, J.D., Paeper, B., Nickerson, D.A., Dea, J., Dong, S., Gonzalez, L.E., Mandell, J.E., Mane, S.M., Murtha,

M.T., Sullivan, C.A., Walker, M.F., Waqar, Z., Wei, L., Willsey, A.J., Yamrom, B., Lee, Y.H., 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 burden of de novo coding mutations in autism spectrum disorder (2014) *Nature* **515** (7526) 216-21 *,+-equal contribution

11. Glessner, J., Bick, A.G., Ito, K., Homsy, J., Rodriguez-Murillo, L., Fromer, M., Mazaika, E.J., Vardarajan, B., Italia, M.J., Leipzig, J., DePalma, S., Golhar, R., Sanders, S.J., Yamrom, B., Ronemus, M., **Iossifov, I**., Willsey, A.J., State, M.W., Kaltman, J.R., White, P.S., Shen, Y., Warburton, D., Brueckner, M., Seidman, C., Goldmuntz, E., Gelb, B.D., Lifton, R., Seidman, J.G., Hakonarson, H., & Chung, W.K. Increased Frequency of De Novo Copy Number Variations in Congenital Heart Disease by Integrative Analysis of SNP Array and Exome Sequence Data. (2014) Circ Res. Epub

12. Narzisi, G., O'Rawe, J.A., **Iossifov, I.**, Fang, H., Lee, Y.H., 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. (2014) *Nat Methods* Epub

13. Yilancioglu, K., Weinstein, Z.B., Meydan, C., Akhmetov, A., Toprak, I., Durmaz, A., **Iossifov, I.**, Kazan, H., Roth, F.P., & Cokol, M. Target-independent prediction of drug synergies using only drug lipophilicity. (2014) *J Chem Inf Model*. **54** (8) 2286

14. Cokol, M., Weinstein, Z.B., Yilancioglu, K., Tasan, M., Doak, A., Cansever, D., Mutlu, B., Li, S., Rodriguez-Esteban, R., Akhmedov, M., Guvenek, A., Cokol, M., Cetiner, S., Giaever, G., **Iossifov, I.**, Nislow, C., Shoichet, B., & Roth, F.P. (2014) Large-scale identification and analysis of suppressive drug interactions. *Chem Biol.* **21** (4) 541

15. Ronemus, M., **Iossifov, I.**, Levy, D., & Wigler, M. (2014) The role of de novo mutations in the genetics of autism spectrum disorders. *Nat Rev Genet*. **15** (2) 133

16. **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, W.R., & Wigler, M. (2012) De Novo Gene Disruptions in Children on the Autistic Spectrum. *Neuron* **74** (2) 285 *-equal contribution

17. Spector, M.S., **Iossifov, I.**, Kritharis, A., He, C., Kolitz, J.E., Lowe, S.W., & Allen, S.L. (2011) Mast-cell leukemia exome sequencing reveals a mutation in the IgE mast-cell receptor β chain and KIT V654A. *Leukemia*. doi:10.1038/leu.2011.354

18. Parla, J.S.*, **Iossifov, I.***, Grabill, I., Spector, M.S., Kramer, M., & McCombie, W.R. (2011) A comparative analysis of exome capture. *Genome Biol.* **12** (9) R97 *-equal contribution

19. Gilman, S.R.*, **Iossifov, I.***, Levy, D., Ronemus, M., Wigler, M., & Vitkup, D. (2011) Rare de novo variants associated with autism implicate a large functional network of genes involved in formation and function of synapses. *Neuron* **70** 898 *-equal

contribution

20. Levy, D., Ronemus, M., Yamrom, B., Lee, Y.H., Leotta, A., Kendall, J., Marks, S., Lakshmi, B., Pai, D., Ye, K., Buja, A., Krieger, A., Yoon, S., Troge, J., Rodgers, L., **Iossifov, I.**, & Wigler, M. (2011) Rare de novo and transmitted copy-number variation in autistic spectrum disorders. *Neuron* **70** 886

21. **Iossifov, I.**, Rodriguez-Esteban, R., Mayzus, I., Millen, K.J., & Rzhetsky, A. (2009) Looking at cerebellar malformations through text-mined interactomes of mice and humans. *PLoS Comput Biol*. **5** (11) e1000559.

22. Rodriguez-Esteban, R. & **Iossifov, I.** (2009) Figure and table mining for biomedical research. *Bioinformatics,* **25** (16) 2082

23. Liu, J., Ghanim, M., Xue, L., Brown, C.D., **Iossifov, I.**, Angeletti, C., Hua, S., Nègre, N., Ludwig, M., Stricker, T., Al-Ahmadie, H.A., Tretiakova, M., Camp, R.L., Perera-Alberto, M., Rimm, D.L., Xu, T., Rzhetsky, A., & White, K.P. (2009) Integrated genomic analysis of the *Drosophila* segmentation network leads to identification of a highly specific biomarker for human kidney cancer. *Science* **323** (5918) 1218

24. **Iossifov, I.**, Zheng, T., Baron, M., Gilliam, T. C., & Rzhetsky, A. (2008) Genetic-linkage mapping of complex hereditary disorders to a whole-genome molecular-interaction network. *Genome Research,* **18** (7) 1150

25. Cokol, M., **Iossifov, I.,** Rodriguez-Esteban, R., & Rzhetsky, A. (2007) Response by Cokol et al *EMBO rep.* **8** (9) 793

26. Cokol, M., **Iossifov, I.,** Rodriguez-Esteban, R., & Rzhetsky, A. (2007) How many papers should be retracted? *EMBO rep.* **8** (9) 793

27. Rodriguez-Esteban, R., **Iossifov, I.**, & Rzhetsky, A. (2006) Imitating manual curation of text-mined facts in biomedicine. *PLoS Comput Biol*, **2** (9) e118

28. Rzhetsky, A., **Iossifov, I.**, Loh, J.M., White, K.P. (2006) Micro-paradigms: Chains of collective reasoning in publications about molecular interactions. *Proc Natl Acad Sci USA*, **103** (13) 4940

29. Cheng, R., Juo, S.H., Loth, J.E., Nee, J., **Iossifov, I.**, Blumenthal, R., Sharpe, L., Kanyas, K., Lerer, B., Lilliston, B., Smith, M., Trautman, K., Gilliam, T.C., Endicott, J. & Baron, M. (2006) Genome-wide linkage scan in a large bipolar disorder sample from the National Institute of Mental Health genetics initiative suggests putative loci for bipolar disorder, psychosis, suicide, and panic disorder. *Mol Psychiatry*. **11** (3) 252

30. Cokol, M., **Iossifov, I.,** Weinreb, C., & Rzhetsky, A. (2005) Emergent behavior of growing knowledge about molecular interactions. *Nature Biotechnology*, **23** (10) 1243.

31. Rzhetsky, A., **Iossifov, I.**, Koike, T., Krauthammer, M., Kra, P., Morris, M., Yu, H., Duboué, P.A., Weng, W., Wilbur, W.J., Hatzivassiloglou, V., & Friedman, C. (2005) GeneWays: A system for extracting, analyzing, visualizing and integrating molecular pathway data. Book chapter in "*Databasing the Brain: From Data to Knowledge (Neuroinformatics)*", Koslow and Subramaniam (Eds).

32. **Iossifov, I.**, Krauthammer, M., Friedman, C., Hatzivassiloglou, V., Bader, J.S., White, K.P., & Rzhetsky, A. (2004) Probabilistic pathway inference from noisy data sources. *Bioinformatics*, **20** (8) 1205.

33. Rzhetsky, A., **Iossifov, I.**, Koike, T., Krauthammer, M., Kra, P., Morris, M., Yu, H., Duboué, P.A., Weng, W., Wilbur, W.J., Hatzivassiloglou, V., & Friedman, C. (2004) GeneWays: A system for extracting, analyzing, visualizing and integrating molecular pathway data. *Journal of Biomedical Informatics*, **37** (1) 43.

34. Krauthammer, M., Kra, P., **Iossifov, I.**, Gomez, S., Hripcsak, G., Hatzivassiloglou, V., Friedman, C., & Rzhetsky, A. (2002) Of truth and pathways: Chasing bits of information through myriads of articles. *Bioinformatics 18 Suppl 1*, S249–S257.

35. Hong, Y., Hatzivassiloglou, V., Friedman, C., **Iossifov, I.,** & Rzhetsky, A. (2002) A rule-based approach for automatically identifying gene and protein names in MEDLINE abstracts. *ISMB 2002*, Poster 240A