German Demidov, PhD

Bioinformatician, medical genomics
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Summary

With over a decade of experience in medical bioinformatics, I have contributed to advancements in rare disease diagnostics, tumor genomic profiling and biomarkers discovery, methods and tools development, new disease gene discoveries, epigenetics of aging. I have been an active participant in two major international consortia, PanCancer Analysis of Whole Genomes (PCAWG) and Solve Rare Disease (Solve-RD). My extensive publication record reflects a deep commitment to advancing our understanding of complex genetic diseases and developing innovative computational tools for genomic research.

Technical Skills

Coding: R/Python/bash (daily usage), Java/C++
Analysis: Statistical analysis, Algorithms design, Visualization, HPC, short/long read sequencing data
Developer Tools: Git, Docker
Languages: Russian (native), English (proficient), German (B1), Spanish (A2)

Experience

Postdoc January 2020 – current
Institute of Medical Genetics and Applied Genomics *Tübingen, Germany*

- leading the long variant detection group within the European Rare Diseases Research Alliance (https://erdera.org/).
- worked on Solve-RD consortium (http://solve-rd.eu/) data as a data manager of one of three data analysis hubs, lead of "Consanguinity and relatedness" work group, member of "CNV and SV detection" workgroup. Results published in more than 10 papers.
- member of an extended diagnostic team in NGS-based diagnostics of tumors and rare diseases, side research projects involving statistical support for medical researchers.

Freelance medical data analyst March 2021 - June 2023
Remote

- provided independent consulting services in the field of biomedical data analysis (up to 7 hours per week), in particular, working on cardiovascular disease risks estimation together with a group at the Broad Institute.

Research assistant January 2018 - December 2019
Institute of Medical Genetics and Applied Genomics *Tübingen, Germany*

- adapted the developed method for germline and somatic CNV detection for routine clinical diagnostics, the method is available on https://github.com/imgag/ClinCNV , all the routine NGS-based diagnostics of rare disease/tumors in our clinic and several partner clinics is done using this approach as for now.
- helped in introduction of the developed method for CNV detection into a clinical pipeline (https://github.com/imgag/megSAP).

Research intern November 2016 - May 2017
Immunology Department, Washington University in Saint Louis *Saint Louis, USA*

- developed a pipeline and statistically analyzed the data from methylation (eRRBS) sequencing of 20 old and 20 young donors, results with additions are published in Nature Aging.

PhD Student September 2015 - December 2017
Center for Genomic Regulation *Barcelona, Spain*

- development of a method for NGS-based CNV detection in research settings, application of the method to Pan Cancer Analysis of Whole Genomes germline data.

Bioinformatician July 2014 - July 2015
Parseq Lab *Saint Petersburg, Russia*

- developed of a method for NGS-based detection of copy-number changes for routine neonatal screening using a panel of 3 genes (CFTR, PAH, GALT) and implementation in Python/Java, results are published in BMC Bioinformatics, available on https://github.com/parseq/convector (in line with the industry practices such as code-review, documentation, logging, etc.)

Teaching assistant December 2011 - May 2012

Lomonosov Moscow State University *Moscow, Russia*

- developed a website for English language department of faculty of Mathematics and Mechanics. Typeset a study book (in English, using LaTeX).

Education

Universitat Pompeu Fabra Barcelona, Spain

PhD in Biomedicine *2015 - 2019*

Saint Petersburg Academic University Saint Petersburg, Russia

Master of Science, Algorithmic Bioinformatics *2013 - 2015*

Lomonosov Moscow State University Moscow, Russia

Diploma (BSc+MSc), Number theory and Information security, Faculty of Mechanics and Mathematics *2008 - 2013*

Publications

Full list: Google Scholar (https://scholar.google.de/citations?user=qOtJIWcAAAAJ)

Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses
January 2025, Nature Medicine (second author)

Comprehensive reanalysis for CNVs in ES data from unsolved rare disease cases results in new diagnoses
October 2024, npj Genomic Medicine (first author)

Structural variant calling and clinical interpretation in 6224 unsolved rare disease exomes
May 2024, European Journal of Human Genetics (first author)

Mobile element insertions in rare diseases: a comparative benchmark and reanalysis of 60,000 exome samples
February 2024, European Journal of Human Genetics (co-first author)

Conferences and seminars

- **ESHG**, 2024 (Berlin, Germany) (diagnostic outcome of structural variants calling in 12.500 exomes of rare disease patients)
- **Undiagnosed Network International**, 2023 (Tbilisi, Georgia), 2024 (Seoul, South Korea), oral presentations (diagnostic results of structural variants calling in Solve-RD genomes and exomes)
- **Undiagnosed Hackathon**, 2023 (Stockholm, Sweden), 2024 (Nijmegen, Netherlands)
- **Machine Learning Summer School**, 2019 (London, England)
- PhD project (ClinCNV, tool for detection of CNVs) presented in 2016, **SBB** (Saint Petersburg, Russia), **EACR**, 2017 (Cambridge, England), **Cancer Genomics**, 2017 Heidelberg, Germany, **ECCB**, 2018 (Athens, Greece), **BIATA** (oral presentation), 2018 (Saint Petersburg, Russia)

Organizational and teaching skills

- organizing a Solvathon on SV/CNV interpretation in short read WGS data (Barcelona, 2023), co-organizing Solvathons in RNA-seq (Barcelona, 2023), long read (Nijmegen, 2024) and multi-omics (Barcelona, 2024) data interpretation
- hackathon mentor at four Summer Schools in Bioinformatics (2014, 2018, 2021, Russia, 2019, Poland; teams of approx 10 people, workload of around 3 full days)
- member of NGSchool (ngschool.eu) organising team (each year from 2017 to 2019)
- speaker at various educational events (NGSchool2016, '17, '19, Poland, Summer School in Bioinformatics'17, '18, '21, Russia)
- mentor at introductory statistics courses at Center for Genomic Regulation, Barcelona, Spain (2015-2016)
- semester projects leader for 2 MSc students in Bioinformatics (2014, Russia)