Joseph D. Szustakowski, Ph.D.

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Biosketch

Joseph D. Szustakowski, Ph.D. is a biopharmaceutical executive leader in data science, currently Vice President of Clinical Development Data Science and AI at Genmab. He has 20+ years of industry experience, passionately focused on the development and deployment of innovative data science methods to advance portfolio projects.

Dr. Szustakowski has built and led talented, global, collaborative data science teams that worked in multiple therapeutic and technical areas (Oncology, Hematology, Cardiovascular, Immunoscience; genomics, genetics, proteomics, image analysis, AI/ML) across all phases of drug discovery and development. His strategic thinking and enterprise mindset catalyze tight alignment between innovative analytics and company objectives.

Joseph joined Genmab in 2024 to build and lead a new data science and AI team, where he is responsible to develop and execute a strategic vision for using modern AI technologies to streamline and optimize clinical development processes and outcomes.

Prior to Genmab, Dr. Szustakowski was actively recruited to BMS in 2015 to reboot the central computational biology function Translational Bioinformatics. He quickly evolved the team's talent, impact, and culture - leading BMS to substantially up-invest in this function, and his rapid promotion to Vice President and head of Translational Bioinformatics.

From 2003 to 2015, Joseph worked as a bioinformatician at Novartis in Cambridge, Massachusetts, where he held various positions of increasing responsibility. Joseph initially worked in high-throughput biology departments focused on target discovery and validation. Mid-way through his tenure, he transitioned to Translational Medicine to build and lead the Bioinformatics team in Biomarker Development - an early industry team focused on analysis of clinical trial exploratory biomarker data.

Joseph has broad experience across the biopharmaceutical lifecycle. He has worked closely with therapeutic areas, technology labs, and biomarker teams - including strategic line management of a genomics experimental lab. Additional collaborations involved program strategies, clinical trial design, medical affairs and communications, and business development assessments.

Throughout his career, he and his team have made numerous high-impact contributions to novel drug targets, the design and execution of precision medicine strategies, and successful regulatory submissions. They have been recognized via health authority approvals, high-profile scientific publications (Nature, Nature Genetics, New England Journal of Medicine, …), prestigious company awards, and - most importantly - positive feedback from patients who benefited from novel therapies.

Dr. Szustakowski received his undergraduate training in Physics and Mathematics (B.S., minor) at the State University of New York at Buffalo Presidential Honors Program. Joseph studied Biomedical Engineering at Boston University (Ph.D.) as a Dean's fellow, where he worked in the emerging field of Bioinformatics. While in graduate school, Joseph served as a member of the Human Genome Project's annotation team, and is a co-author of the 2001 Nature paper describing the first draft of the human genome.

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Professional Profile

Results driven data science executive with 20+ years of experience in the biopharmaceutical industry. Strong track record of building and leading global, matrixed, high performance, collaborative data science teams. Strategic thinker focused on aligning innovative analytical capabilities with enterprise objectives, resulting in concrete impact on pipeline programs, patient benefit, and company revenue. Ability to navigate and manage many types of change (organizational redesigns, strategic refreshes, budget adjustments, etc.).

Key Accomplishments

Deliverables - Select KPIs

- AI innovation and deployment across **many scientific and operational use cases**
- Exploratory analysis of data from **100s of clinical trials**
- Biomarker data analysis included in **10s of Health Authority submissions**
- Critical biomarker findings that led to **2+ precision medicine and favorable label approvals**
- Identified, validated, and progressed **5+ novel targets** to clinical trials
- Managed and mentored **25+ employees** who advanced to Director level+ roles
- Contributed to **10s of business development due diligence assessments**
- Represented the company in **5+ external, industry-wide consortia**

Data Science, AI/ML

- Development and deployment of modern AI/ML methods focused on concrete portfolio impact
  - LLM agentic algorithms to generate draft documents for internal use, regulatory submissions, etc.
  - AI "super-advisors" to optimize asset strategies, clinical trial protocols, etc.
  - Federated exploration of various high content databases
  - Accurate translation of scientific documents across multiple languages
- AI driven Deep Learning algorithms for digital pathology image analysis
  - Replicate human pathologist assessments in more consistent and scale-able manner
  - Generate automated, complex, multi-dimensional signal detections
  - End-to-end deep learning algorithms to predict tumor state, genomic characteristics, patient outcomes, etc.
- Modern machine learning methods to analyze structured, numerical data
  - Assessment of clinical trial site efficiency, prediction of recruitment rates
  - Interpretable techniques to analyze clinical trial exploratory biomarker data

Scientific Leadership

- Identification of novel targets currently in clinical trials (e.g. anti–CCR8 in solid tumors: (trial: NCT04895709))
- Analysis and interpretation of exploratory data from 100s of clinical trials (oncology and non-oncology)
  - Identify and validate predictive biomarkers, enhance disease and drug mechanism understanding
  - deucravacitinib / plaque psoriasis: contributed to FDA approval with a favorable label (Press Release)
  - nivolumab / gastroesophageal adenocarcinoma: EMA approval with a precision medicine label (Press Release)
- Executive sponsor and representative for Real World Data / Human Genetics consortia (UK Biobank Exome Sequencing Consortium (UKB-ESC paper), UK Biobank Pharma Proteomics Project (UKB-PPP paper), etc.)
- Exploration of multi-modal clinical trial and real world patient data in complex diseases

Business Strategy, External Executive Presence

- Contributed to multiple enterprise level strategies
- Due diligence assessments for multiple acquisitions, equity investments, asset licenses, and external partnerships

- Core member of several business partnership Joint Steering/Executive Committees (JSC, JEC)
- Organized, led, and participated in multiple key, high-profile external congresses, councils, and forums

External Executive Presence

- Core member of several business partnership Joint Steering/Executive Committees (JSC, JEC)
- Organized, led, and participated in multiple key, high-profile external congresses, councils, and forums

Team and Talent Management

- Planned and executed multiple successful organization build-ups, reboots, and restructures
- Consistent history of recruiting, retaining, and developing top talent
- Managed, coached, or mentored 25+ colleagues that now hold Director level or above roles

Areas of Expertise

- Computational research, AI, ML across many domains
- High-impact innovation
- Scientific and business strategy
- Leadership and team building
- Talent acquisition and development
- Collaboration - internal and external
- Cross-functional partnerships
- Managing organizational change
- Budget management
- Communication

Professional Experience

Genmab, June 2024 – Present, Princeton, NJ

2024–Present: Vice President, Clinical Development Data Science and AI

Building and leading a new, global team of data scientists, focused on innovative methodologies to streamline and optimize our clinical development processes and outcomes.

Collaborating with cross-functional teams, including Clinical Operations, Medical Writing, Regulatory Affairs, Safety/Pharmacovigilance, CMC/Manufacturing, Programming, etc. to align data science initiatives with broader clinical and business objectives.

Innovation and deployment of modern AI technologies across various use cases including:
- Automated drafting of internal and regulatory documents
- AI advisors trained on 'sources of truth' to assist in clinical trial protocol optimization
- Efficient and accurate quality assessment of documents, protocols, and policies
- Accurate AI translation of scientific and technical documents across multiple languages

Bristol Myers Squibb (BMS), Feb 2015 – May 2024, Princeton, NJ

2019–2024: Vice President, Translational Bioinformatics

2017–2019: Executive Director, Translational Bioinformatics

2015–2017: Group Director, Translational Bioinformatics

Leader of a global team of approximately 90 Ph.D. and M.S. level bioinformaticians, data scientists, statisticians, AI/ML specialists, and genomics bench scientists. Broad global footprint of 10 sites located in 9 cities / 4 countries / 3 continents.

Team Structure
- Portfolio-facing computational biology teams focused on specific disease areas
  - Solid Tumor Oncology
  - Hematology / Oncology
  - Immunoscience, Cardiovascular, and Neuroscience (ICN)

- Core, ”horizontal” capabilities with deep technical expertise that collaborate broadly across the portfolio
  - Computational Genomics
  - Sequencing Research and Genomics (an –omics experimental web lab)
  - Digital Pathology
  - Bioinformatics Methodology

Select Achievements

- Actively recruited to BMS in 2015 to reboot its central bioinformatics function
- Evolved the team culture and stakeholder relationships from service-oriented to collaborative partnership
- Addressed team employee demographic disparities via progressive recruitment tactics
- Raised team and stakeholder expectations on skills, deliverables, and program impact
- Oversaw rapid build-up from apprx. 15 to 90 highly talented computational scientists
- Introduced BMS to the importance of human genetics research for drug discovery
- Led efforts to define and implement quality procedures and reproducible research environment

Novartis Institutes for Biomedical Research (NIBR), July 2003 – Feb 2015, Cambridge, MA

2014–2015: Associate Director, Marker Data Sciences (Biomarker Development)

2009–2014: Associate Director, Bioinformatics (Biomarker Development)

2007–2009: Research Investigator II

2006–2007: Research Investigator I

2003–2006: Investigator

Held roles of rapidly increasing responsibility while at Novartis. From 2003 to 2008 focused on early target discovery and validation projects. In 2008 transitioned to the Biomarker Development group in Translational Medicine to build and lead a new Bioinformatics team. In the last role, led the Biomarker Development Marker Data Sciences team, a group of apprx. 10 bioinformaticians and statisticians focused on applying cutting edge approaches to the analysis and interpretation of various biomarker data sets generated in early clinical trials.

Select Achievements

- Early innovator for pathway-centric analysis of -omics data
- Developed one of the first in industry bioinformatics teams focused on exploratory clinical trial data analysis
- Contributed to multiple HA submissions
- Led multiple human genetics research initiatives, resulting in the identification of causal biology in 10+ historically unsolved inherited conditions
- Innovative advocate for reproducible research standards and technologies

Compaq Cambridge Research Laboratory, 2000, Cambridge, MA

While in graduate school, held a side-gig as a consultant for Compaq Computer's Cambridge Research Laboratory, and contributed to the annotation of the International Human Genome Sequencing project - **leading to co-authorship of the 2001 Nature paper describing the first draft of the human genome**.

Education

Ph.D. Biomedical Engineering, Boston University (Dean's Fellowship), Boston, MA
Dissertation: A Protein Structure Alignment Method and Application to the Discovery of Recurrent Structure Motifs.
Dissertation Committee: Zhiping Weng, Ph.D. (adviser); Charles DeLisi, Ph.D.; Temple Smith, Ph.D.; Simon Kasif, Ph.D.; Scott Mohr, Ph.D.

B.S. Physics; minor Mathematics, State University of New York at Buffalo, Buffalo, NY
Presidential Honors Program, *Magna Cum Laude*

Publications (select, additional in appendix)

"Plasma proteomic associations with genetics and health in the UK Biobank"
Nature, Oct 5, 2023 [Pubmed | DOI]

- *This paper was accompanied by a Research Highlight in Nature Genetics:*
  Wei Li. "UK Biobank pharma proteomics resource."
  Nature Genetics, published online November 8, 20233. [Pubmed | DOI]

"Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank "
Nature Genetics June 28, 2023 [Pubmed | DOI]

- *This paper was accompanied by a Research Highlight in Nature Reviews Genetics:*
  Linda Koch. "The power of large-scale exome sequencing."
  Nature Reviews Genetics, published online July 15, 2021. [Pubmed | DOI]

The International Human Genome Sequencing Consortium.
"Initial sequencing and analysis of the human genome."
Nature, 409(6822):860-921, Feb 15 2001. [Pubmed | DOI]

"Nivolumab plus Ipilimumab in Lung Cancer with a High Tumor Mutational Burden."
The New England Journal of Medicine, online April 16, 2018. [Pubmed | DOI]

"STK11/LKB1 Mutations and PD-1 Inhibitor Resistance in KRAS-Mutant Lung Adenocarcinoma."
Cancer Discovery, May 17, 2018. [Pubmed | DOI]

"STK11 and KEAP1 Mutations as Prognostic Biomarkers in an Observational Real-World Lung Adenocarcinoma Cohort"
ESMO Open Online April 16, 2020 [Pubmed | DOI]

"Genome-Wide Association Analysis Identifies Genetic Correlates of Immune Infiltrates in Solid Tumors."
PLoS ONE, 27 July 2017. [Pubmed | DOI]

"Whole Exome Sequencing Reveals Overlap Between Macrophage Activation Syndrome in Systemic Juvenile Idiopathic Arthritis and Familial Hemophagocytic Lymphohistiocytosis."
Arthritis and Rheumatology, published online Jul 21, 2014. [Pubmed | DOI]

"Less is more: towards an optimal universal description of protein folds."
Bioinformatics, 21(Supplement 2, ECCB):66-71, 2005. [Pubmed | DOI]

Publication Metrics, Web of Science, April 2025

- H-Index: 29
- Publications: 53
- Sum of Times Cited 23,405
- Citing Articles: 22,612
- Sum of Times Cited by Patents: 383
- Citing Patents: 355

Enterprise Level Awards (select, additional in appendix)

BMS, **James B.D. Palmer Award for Excellence in Drug Development**, for Tumor Mutational Burden (TMB) as a predictive biomarker, 2018

Privileged to be part of a team recognized for their scientific contributions to Oncology Precision Medicine *via the **most prestigious Drug Development award granted by BMS***

BMS, **Leveraging Technology (LevTech)**, for Reproducible Research Environment, 2017

*Team was granted the **most prestigious BMS technology award** for their innovative Reproducible Research system*

Novartis, **Global IT Award for Innovation**, Pathway Analysis Toolkit, 2007

Awarded for the systematic application of pathway-centric analysis methods to ALL microarray experiments conducted at NIBR

Appendices

Scientific Research Leadership Experience (select)

Artificial Intelligence and Machine Learning (AI/ML)

- Development and deployment of modern AI/ML approaches with a focus on concrete impact on productivity and pipeline impact
- AI driven Deep Learning algorithms for image analysis, focused on digital pathology
  - Replicate human pathologist assessments in more consistent and scale-able manner (PD-L1 tumor proportion score, HIFs, various IHC readings)
  - Generate accurate readings too complicated for human pathologists to perform in a consistent, scale-able manner (CD8 topology, PD-L1 combined positive score)
  - Develop end-to-end deep learning algorithms to predict tumor state, genomic characteristics, patient outcomes, etc.
- Generative Large Language Models (LLMs)
  - Increase efficiency of scientific programming
  - Federated exploration of various high content databases
  - Highly efficient drafting of regulatory documents, experiment reports
  - Accurate translation of scientific documents across languages
- Modern machine learning methods to analyze structured, numerical data
  - Assessment of clinical trial site efficiency, prediction of recruitment rates
  - Interpretable techniques to analyze clinical trial exploratory biomarker data

Real-World Data

- Contributed to enterprise strategy initiatives to identify and up-invest in external RWD resources.
- BMS executive sponsor and representative for multiple cross industry pre-competitive real world data consortia (UK Biobank - Exome Sequencing Consortium, and Pharma Proteomics Project (UKB-ESC, UKB-PPP), Illumina/Nashville Biosciences Alliance for Genomics Discovery (AGD), AACR GENIE Biopharma Consortium)
- Assessment to replicate, validate, or complement findings identified in smaller internal data sets. Examples include validation of STK11 as a prognostic (not predictive) biomarker for cancer patient outcomes

Human Genetics and Disease (non-Oncology)

- Introduced BMS to the importance of human genetics research for drug discovery, leading to an up-investment in talent, real world data resources, and collaborative partnerships (UKB-ESC, UKB-PPP, AGD)
- Identified the genetic cause of 10+ inherited diseases in partnership with academic collaborators: renal disorders, Keratolytic Winter Erythema (KWE), auto-inflammatory conditions, SJIA/macrophage activation syndrome (MAS)/Hemophagocytic lymphohistiocytosis (HLH)
- Analysis of high-throughput data sets to contribute to critical portfolio life-cycle decisions including target identification and validation, patient and indication selection, disease and therapeutic mechanisms of action, etc.
  - TYK2 / deucravacitinib / multiple auto-immune conditions
  - Myosin / mavacamten / cardiomyopathy
  - IL-1β/ canakinumab / MWS, SJIA, CAPS, FMF, FCAS, ...

Proteomics

- Participated in cross-company collaboration to expand a cutting edge affinity based protein screening platform to include 1000s of analytes
- Early advocate for UKB-PPP consortium, leading to the generation of the largest human plasma proteomics data resource to date
- Cutting edge analysis of proteomics screening data via Mendelian Randomization (forward and reverse), pQTL analysis, phenotypic associations studies, etc. to identify novel drug targets and biomarkers

Next Generation Sequencing

- Evaluation, deployment, and optimization of many NGS technologies
- Assessment, internalization, and robust implementation of reproducible NGS data analysis pipelines
- External partnerships with computational vendors to production-ize and scale pipelines in a cost effective manner
- Responsible for the BMS Sequencing Research and Genomics (SRG) team

  - Experimental, laboratory-based genomics capability
  - Collaborates with Research teams across BMS to develop and apply innovative genomics technologies
  - Established and manages an ecosystem of external partners that perform commoditized experiments in a high quality and cost effective manner

Oncology

- Deployment of novel -omics technologies and innovative analytical methods to assess the tumor microenvironment (bulk RNA-Seq, Whole Exome Seq, scRNA-Seq, scCITE-Seq, scATAC-Seq, IHC, flow cytometry, spatial technologies, etc.)
- Assessment of tumor immunogenicity via various proxies (Tumor Mutational Burden/TMB, neo-antigen predictions, etc.) to understand and predict response to immuno-therapies.
- Mining of large-scale, multi-modal patient data sets to evaluate tumor intrinsic/extrinsic interactions, identity homogeneous patient subsets, propose and validate new targets with precision medicine potential

Historic

- Early adopter of legacy technologies and methods including DNA/RNA Microarray, pathway-analysis methods, protein structural informatics, sequence library searches and analysis,

Publications

Highlights - Genetics / Disease Biology

Benjamin B. Sun, Joshua Chiou, Matthew Traylor, ..., **Joseph D. Szustakowski**, Bradford W. Gibson, Melissa R. Miller, Christopher D. Whelan.
"Plasma proteomic associations with genetics and health in the UK Biobank"
***Nature*, Oct 5, 2023** [Pubmed | DOI]

- *This paper was accompanied by a Research Highlight in Nature Genetics:*
  Wei Li. "UK Biobank pharma proteomics resource."
  Nature Genetics, published online November 8, 20233. [Pubmed | DOI]

Joseph D Szustakowski, Suganthi Balasubramanian, Erika Kvikstad, ..., Aris Baras, Lyndon J Mitnaul, Jeffrey G Reid, UKB-ESC Research Team
"Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank "
***Nature Genetics* June 28, 2021** [Pubmed | DOI]

- *This paper was accompanied by a Research Highlight in Nature Reviews Genetics:*
  Linda Koch. "The power of large-scale exome sequencing."
  Nature Reviews Genetics, published online July 15, 2021. [Pubmed | DOI]

Chen Li, Nicolas De Jay, Supriya Sharma, Katrina A. Catalano, Emily R. Holzinger, Venkatesh Sridharan, Zhaoqing Wang, Lei Zhao, **Joseph D. Szustakowski**, Ching-Pin Chang, Joseph C. Maranville, Erika M. Kvikstad.
"Proteome-wide Mendelian randomization identifies candidate causal proteins for cardiovascular diseases"
***Advanced Genetics*, March 10, 2025** [DOI | Pubmed]

Lander ES, Linton LM, ... , **Szustakowski J**, ...
The International Human Genome Sequencing Consortium.
"Initial sequencing and analysis of the human genome."
***Nature*, 409(6822):860-921, Feb 15 2001.** [Pubmed | DOI]

Thandiswa Ngcungcu, Martin Oti, Bjorn I. Haukanes, Jan C. Sitek, Fan Yang, ..., **Joseph Szustakowski**, Frank Staedtler, Huiqing Zhou, Torunn Fiskerstrand, Michele Ramsay.
"Duplicated enhancer region upstream of the CTSB gene segregates with Keratolytic Winter Erythema in South African and Norwegian families."
***American Journal of Human Genetics*, 100:5, p737-750, 4 May 2017.** [Pubmed | DOI]

Albane A. Bizet, Anita Becker-Heck, Rebecca Ryan, ..., **Joseph D. Szustakowski**, Friedhelm Hildebrandt, Esben Lorentzen, Andreas W. Sailer, Alexandre Benmerah, Pierre Saint-Mezard, Sophie Saunier.
"Mutations in TRAF3IP1/IFT54 reveal a new role for IFT proteins in microtubule stabilization."
***Nature Communications*, 6:8666, October 21, 2015.** [Pubmed | DOI]

Olivia Boyer, Stéphanie Woerner, Fan Yang, Edward J. Oakeley, Bolan Linghu, ..., **Joseph D. Szustakowski**, ..., Marie-Claire Gubler, Keith J. Johnson, Salah-Dine Chibout, Corinne Antignac.
"LMX1B mutations as unexpected causes of hereditary focal segmental glomerulosclerosis without extra-renal involvement."
***JASN*, Jul;24(8):1216-22, 2013.** [Pubmed | DOI]

- *This paper was accompanied by a special editorial in JASN:*
  Jeffrey B. Kopp. "An Expanding Universe of FSGS Genes and Phenotypes: LMX1B Mutations Cause Familial Autosomal Dominant FSGS Lacking Extrarenal Manifestations."
  JASN, published online July 18, 2013. [Pubmed | DOI]

Highlights - Oncology

Matthew D. Hellmann, Tudor-Eliade Ciuleanu, ..., **Joseph Szustakowski**, Prabhu Bhagavatheeswaran, Diane Healey, M.S., Yali Fu, Faith Nathan, and Luis Paz-Ares.
"Nivolumab plus Ipilimumab in Lung Cancer with a High Tumor Mutational Burden."
The New England Journal of Medicine, online April 16, 2018. [Pubmed | DOI]

Ferdinandos Skoulidis, Michael E. Goldberg, Danielle M. Greenawalt, Matthew D. Hellmann, ... **Joseph Szustakowski**, ..., William J. Geese, Lee A. Albacker and John V. Heymach.
"STK11/LKB1 Mutations and PD-1 Inhibitor Resistance in KRAS-Mutant Lung Adenocarcinoma."
Cancer Discovery, May 17, 2018. [Pubmed | DOI]

Nathan O Siemers, James L Holloway, Han Chang, Scott D Chasalow, Petra B Ross-MacDonald, Charles F Voliva, **Joseph D Szustakowski**.
"Genome-Wide Association Analysis Identifies Genetic Correlates of Immune Infiltrates in Solid Tumors."
PLoS ONE, 27 July 2017. [Pubmed | DOI]

Additional Publications

J Transl Med . 2024 Feb 21;22(1):190. doi: 10.1186/s12967-023-04705-3.
Mike Mason, Óscar Lapuente-Santana, Anni S Halkola, ... Justin Guinney, **Joseph D Szustakowski**, Benjamin G Vincent, David P Carbone.
"A community challenge to predict clinical outcomes after immune checkpoint blockade in non-small cell lung cancer"
Journal of Translational Medicine, Feb 21, 2024 [Pubmed | DOI]

Kosmicki JA, Horowitz JE, Banerjee N, ..., **Szustakowski JD**, ..., Regeneron Genetics Center; UKB Exome Sequencing Consortium, ..., Reid JG, Baras A, Abecasis GR, Ferreira MAR.
"Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals."
Am J Hum Genet. 2021 Jul 1;108(7):1350-1355. [Pubmed | DOI]

Benjamin G. Vincent, **Joseph D. Szustakowski**, Parul Doshi, Michael Mason, Justin Guinney, and David P. Carbone.
"Pursuing Better Biomarkers for Immunotherapy Response in Cancer Through a Crowdsourced Data Challenge"
JCO Precision Oncology January 8, 2021 [DOI]

Natallia Kalinava, Abraham Apfel, Robert Cartmell, ..., **Joseph Szustakowski**, Scott D. Chasalow, Ariella Sasson, Stefan Kirov
"Modeling Performance of Sample Collection Sites Using Whole Exome Sequencing Metrics"
BioTechniques Online October 26, 2020 [DOI]

Daniel K. Wells, Marit M. van Buuren, ... , **The Tumor Neoantigen Selection Alliance(*)**, ..., Pia Kvistborg, Nadine A. Defranoux.
"Key Parameters of Tumor Epitope Immunogenicity Revealed Through a Consortium Approach Improve Neoantigen Prediction"
Cell Online October 09, 2020 [Pubmed | DOI]

○ *(*) My team at BMS participated in this consortium and was included as banner authors.*

Simon Papillon-Cavanagh, Parul Doshi, Radu Dobrin, **Joseph Szustakowski**, Alice M Walsh
"STK11 and KEAP1 Mutations as Prognostic Biomarkers in an Observational Real-World Lung Adenocarcinoma Cohort"
ESMO Open Online April 16, 2020 [Pubmed | DOI]

Han Chang, Ariella Sasson, Sujaya Srinivasan, Ryan Golhar, Danielle M. Greenawalt, William J. Geese, George Green, Kim Zerba, Stefan Kirov, **Joseph Szustakowski**
"Bioinformatic Methods and Bridging of Assay Results for Reliable Tumor Mutational Burden Assessment in Non-

Small Cell Lung Cancer."
Molecular Diagnosis and Therapy Online June, 2019 [Pubmed | DOI]

Ready N, Hellmann MD, ..., **Szustakowski JD**, ..., Green G, Chang H, Ramalingam SS.
"First-Line Nivolumab Plus Ipilimumab in Advanced Non-Small-Cell Lung Cancer (CheckMate 568): Outcomes by Programmed Death Ligand 1 and Tumor Mutational Burden as Biomarkers."
J Clin Oncol., Apr 20, 2019 [Pubmed | DOI]

Hellmann MD, Callahan MK, Awad MM, ..., **Szustakowski JD**, Sasson A, Golhar R, Vitazka P, Chang H, Geese WJ, Antonia SJ.
"Tumor Mutational Burden and Efficacy of Nivolumab Monotherapy and in Combination with Ipilimumab in Small-Cell Lung Cancer."
Cancer Cell. 2019 Feb 11, 2019 [Pubmed | DOI]

Matthew D. Hellmann, Margaret K. Callahan, Mark M. Awad, ..., **Joseph D. Szustakowski**, Ariella Sasson, Ryan Golhar, Patrik Vitazka, Han Chang, William J. Geese, Scott J. Antonia.
"Tumor Mutational Burden and Efficacy of Nivolumab Monotherapy and in Combination with Ipilimumab in Small-Cell Lung Cancer."
Cancer Cell, May 14 2018, 33:5. [Pubmed | DOI]

Nicole Hartmann, Evert Luesink, Edward Khokhlovich, **Joseph D Szustakowski**, Lukas Baeriswyl, Joshua Peterson, Andreas Scherer, Nirmala R Nanguneri, Frank Staedtler.
"The use of haplotype-specific transcripts improves sample annotation consistency."
Biomarker Research, 2:17, 2014. [Pubmed | DOI]

Kenneth M. Kaufman, Bolan Linghu, **Joseph D. Szustakowski**, Ammar Husami, Fan Yang, Kejian Zhang, Alexandra Filipovich, Ndate Fall, John B. Harley, N.R. Nirmala, and Alexei A. Grom.
"Whole Exome Sequencing Reveals Overlap Between Macrophage Activation Syndrome in Systemic Juvenile Idiopathic Arthritis and Familial Hemophagocytic Lymphohistiocytosis."
Arthritis and Rheumatology, published online Jul 21, 2014. [Pubmed | DOI]

Edward Khokhlovich, Daniel Wahl, Anthony Masiello, Pierre Parisot, Stefan El-Ghatta, **Joseph D. Szustakowski**, Nanguneri Nirmala, David S. Tuch.
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"DeconRNASeq: A Statistical Framework for Deconvolution of Heterogeneous Tissue Samples Based on mRNA-Seq data."
Bioinformatics, Feb 21 2013. [Pubmed | DOI]
Bioconductor package: http://www.bioconductor.org/packages/release/bioc/html/DeconRNASeq.html

Gong T, Hartmann N, Kohane IS, Brinkmann V, Staedtler F, Letzkus M, Bongiovanni S, **Szustakowski JD**.
"Optimal deconvolution of transcriptional profiling data using quadratic programming with application to complex clinical blood samples."
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Manley K, Anderson J, Yang F, **Szustakowski J**, Oakeley EJ, Compton T, Feire AL.
"Human cytomegalovirus escapes a naturally occurring neutralizing antibody by incorporating it into assembling virions."
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In Keith DiPetrillo (editor), *Methods in Molecular Biology: Cardiovascular Genomics*, 573:259-84, 2009. [Pubmed | DOI]

Scherer A, Gwinner W, Mengel M, Kirsch T, Raulf F, **Szustakowski JD**, Hartmann N, Staedtler F, Engel G, Klupp J, Korn A, Kehren J, Haller H.
"Transcriptome changes in renal allograft protocol biopsies at 3 months precede the onset of interstitial fibrosis/tubular atrophy (IF/TA) at 6 months."
Nephrol Dial Transplant, Apr 27, 2009. [Pubmed | DOI]

Sandipan Chatterjee, **Joseph D. Szustakowski**, N.R. Nirmala, Craig Mickanin, Mark Labow, Axel Nohturfft, Kumlesh Dev, and Rajeev Sivasankaran.
"Identification of Novel Genes and Pathways Regulating Intracellular Cholesterol Homeostasis."
PLOS One, April 21;4(4), 2009. [Pubmed | DOI]

Qicheng Ma, Gung-Wei Chirn, **Joseph D. Szustakowski**, Adel Bakhtiarova, Penelope A Kosinski, Daniel Kemp and Nanguneri Nirmala.
"Uncovering Mechanisms of Transcriptional Regulations by Systematic Mining of Cis Regulatory Elements with Gene Expression Profiles."
BioData Mining, July 17;1(4), 2008. [Pubmed | DOI]

Joseph D Szustakowski, Penelope A Kosinski, Christine A Marrese, Jee-Hyung Lee, Stephen J Elliman, NR Nirmala and Daniel M Kemp.
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Kemp D, Nirmala NR, **Szustakowski JD**.
"Extending the pathway analysis framework with a test for transcriptional variance implicates novel pathway modulation during myogenic differentiation."
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Shamina M Rangwala, Xiaoyan Li, Loren Lindsley, Xiaomei Wang, Stacey Shaughnessy, Thomas G Daniels, **Joseph Szustakowski**, N.R. Nirmala, Zhidan Wu, Susan C Stevenson.
"Estrogen-Related Receptor-α is a Key Mediator of Mitochondrial Gene Expression."
Biochem Biophys Res Commun, May 25;357(1):231-6, 2007. [Pubmed | DOI]

Wilson L, Yang Q, **Szustakowski JD**, Gullicksen PS, Halse R.
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Ma Q, Chirn GW, Cai R, **Szustakowski JD**, Nirmala NR.
"Clustering protein sequences with a novel metric transformed from sequence similarity scores and sequence alignments

with neural networks."
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Zheng Y, **Szustakowski JD**, Fortnow L, Roberts RJ, Kasif S.
"Computational identification of operons in microbial genomes."
Genome Research, 12(8):1221-30, 2002. [Pubmed]

Szustakowski JD and Weng Z.
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Evolutionary Computation in Bioinformatics.
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Resendes BL, Robertson NG, **Szustakowski JD**, Resendes RJ, Weng Z, Morton CC.
"Gene discovery in the auditory system: Characterization of additional cochlear-expressed sequences."
Journal of the Association for Research in Otolaryngology, 3(1):45-53, 2002. [Pubmed | DOI]

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Proteins, 38(4):428-440, Mar 2000. [Pubmed]

Presentations (select)

Szustakowski, Joseph D. Panel: Getting Started with AI. *BioNJ*, Stevens Institute of Technology, Hoboken, NJ, November 2024.

Szustakowski, Joseph D. Panel: Smart Algorithms Smarter Drugs. *HBA Healthcare Businesswomen's Association*, Rutgers University, New Brunswick NJ, Oct 2024.

Szustakowski, Joseph D. Understanding the Impact of Ancestral and Demographic Diversity On -Omics Data is Essential For Biomedical Research: Case Studies. *AAPS*, Orlando, FL, October 2023.

Szustakowski, Joseph D. Enabling Precision Drug Discovery with -Omics Technologies and Data Analytics. *Festival of Genomics*, Boston, MA, October 2023.

Szustakowski, Joseph D. Accelerating Biomarker Discovery for Precision Medicine with Multi-Modal Data and Advanced Analytics. *BIO-IT World Conference and Expo*, Boston, MA, May 2022.

Szustakowski, Joseph D., George Green, Kim Zerba, Patrik Vitazka, Han Chang. Calibration of FoundationOne with Whole Exome Sequencing for Assessment of Tumor Mutation Burden as a Biomarker for Response to Immune Checkpoint Inhibitors. *SITC; Immuno-Oncology Biomarkers*, National Harbor Maryland, Nov 2017.

Szustakowski, Joseph D. Reproducible NGS Research: Practical Approaches and Case Studies. *BIOIT World 2014*, Boston, MA, April 2015.

Szustakowski, Joseph D. Disease and Big (NGS) Data: Searching for Needles in Needlestacks. *Discovery on Target 2014, Big Data Analytics and Solutions*, Boston, MA, October 2014.

Szustakowski, Joseph D. High-Performance Databases to Manage and Analyze NGS Data. *BIOIT World 2014*, Boston, MA, April 2014.

Szustakowski, Joseph D. Next Generation Sequencing in Drug Discovery. *University of Massachusetts Medical School*, Worcester, MA, September 2013.

Szustakowski, Joseph D. Bioinformatics in Industry. *Harvard Stem Cell Institute (HSCI)*, Cambridge, MA, August 2013.

Szustakowski, Joseph D. Next Generation Sequencing in Drug Discovery. *ADAPT*, Washington D.C., September 2012.

Szustakowski, Joseph D. Sequence Everything! Next Generation Sequencing in Drug Discovery. *Second World Pharmacogenetics Summit*, Boston, MA, April 2012.

Szustakowski, Joseph D. From Data to Discovery: I have my NGS data, now what? *BIOIT World 2011*, Boston, MA, April 2011.

Szustakowski, Joseph D. An NGS Survival Guide. *BIOIT World 2011*, Boston, MA, April 2011.

Szustakowski, Joseph D. Integrating High-Throughput Data via Pathway-Centric Analyses. *CHI Molecular Medicine Tri-Conference, Pathway Analysis track*, San Francisco, CA, March 2008.

Conference Abstracts

A. Hukku, E. Kvikstad, S. Vasquez Grinnell, **J. D. Szustakowski**, J. Maranville, E. Holzinger
Integrative analysis of UK Biobank proteomics data with autoimmune disease GWAS reveals complex causal relationships

American Society of Human Genetics (ASHG), Washington DC, November 2023 (oral presentation)

K. Catalano, C. Li1, A. Hukku, E. Kvikstad, S. Vasquez-Grinnell, **J. Szustakowski**, E. Holzinger, J. Maranville
"Sensitivity of MR analyses to pQTL instrumental variable selection contributes to lack of replication in heart failure"
American Society of Human Genetics (ASHG), Washington DC, November 2023 (poster)

Ron Ammar, Michael Basso, ..., **Joseph D. Szustakowski**, ..., Julio A. Chirinos, Thomas P. Cappola
"Proteomic biomarkers of risk in patients with Heart failure and Reduced Ejection Fraction: Insights from the PENN Heart Failure Study (PHFS)"
American Heart Association (AHA) Scientific Sessions 2019, Philadelphia, PA, November 2019 (poster)

Ron Ammar, Michael Basso, ..., **Joseph D. Szustakowski**, ..., Julio A. Chirinos, Thomas P. Cappola
"Penalized regression defines a composite marker for risk stratification in heart failure with reduced ejection fraction in the PENN Heart Failure Study (PHFS)"
American Heart Association (AHA) Scientific Sessions 2019, Philadelphia, PA, November 2019 (poster)

H. Chang, S. Srinivasan, A. Sasson, R. Golhar, D. Greenawalt, S. Kirov, **J.D. Szustakowski**.
"Toward the Standardization of Bioinformatics Methods for the Accurate Assessment of Tumor Mutational Burden (TMB)."
ESMO, 2018, Munich, Germany, October 2018 (poster)

J. D. Szustakowski, G. Green, W. J. Geese, K. Zerba, H. Chang.
"Evaluation of tumor mutation burden as a biomarker for immune checkpoint inhibitor efficacy: A calibration study of whole exome sequencing with FoundationOne."
AACR Annual Meeting, 2018, Chicago, IL, April 2018 (poster)

M. D. Hellmann, T. Ciuleanu, A. Pluzanski, ..., **J. D. Szustakowski**, P. Bhagavatheeswaran, D. Healey, Y. Fu, F. Nathan, L. Paz-Ares.
"Nivolumab (nivo) + ipilimumab (ipi) vs platinum-doublet chemotherapy (PT-DC) as first-line (1L) treatment (tx) for advanced non-small cell lung cancer (NSCLC): initial results from CheckMate 227."
AACR Annual Meeting, 2018, Chicago, IL, April 2018 (oral presentation by M. Hellmann)

S. Ramalingam, M. D. Hellmann, M. Awad, H. Borghaei, J. Gainor, ..., B. Lestini, **J. D. Szustakowski**, H. Chang, N. Ready.
"Tumor mutational burden (TMB) as a biomarker for clinical benefit from dual immune checkpoint blockade with nivolumab (nivo) + ipilimumab (ipi) in first-line (1L) non-small cell lung cancer (NSCLC): identification of TMB cutoff from CheckMate 568."
AACR Annual Meeting, 2018, Chicago, IL, April 2018 (oral presentation by S. Ramalingam)

Thandiswa Ngcungcu, M. Oti, P. Hull, ..., **Joseph Szustakowski**, Frank Staedtler, Jo Zhou, Torunn Fiskerstrand, Michele Ramsay.
"Duplicated enhancer region upstream of the CTSB gene segregates with keratolytic winter erythema in South African and Norwegian families."
European Conference of Human Genetics, Barcelona, Spain, May 2016. (oral presentation by T. Ngcungcu)

B. Linghu, F. Yang, R. Bruccoleri, **J. Szustakowski**.
"A high-performance database framework for fast and easy prioritization of disease related variants from Exome Sequencing data."
Annual Meeting of The American Society of Human Genetics, San Diego, CA, October 2014. (poster)

T. Ngcungcu, B. Linghu, F. Yang, E. Oakeley, F. Staedtler, R. Bruccoleri, N. Nirmala, S. Buechmann-Moller, M. Marc Sultan, **J. Szustakowski**, M. Ramsay.
"Non-coding variants segregate with disease in South African families with keratolytic winter erythema (KWE)."
Annual Meeting of The American Society of Human Genetics, Baltimore, MD, October 2015. (poster)

K.J Johnson, O. Boyer, S. Woerner, F. Yang, E. Oakeley, ...,**J.D. Szustakowski**, F. Heibel, M. Matignon, V. Baudouin F. Chantrel, J. Champigneuelle, L. Martin, P. Nitshke, M-C. Gubler, S-D. Chibout, C. Antignac.
"Autosomal Dominant FSGS without extra-renal symptoms due to mutations in LMX1B gene."
Annual Meeting of The American Society of Human Genetics, Boston, MA, October 2013 (poster)

B. Linghu, F. Yang, R. Bruccoleri, D. Spiewak, **J. Szustakowski**.
"A high-performance database framework for fast and easy prioritization of disease related variants from Next Generation Sequencing data."
17th International Conference on Research in Computational Molecular Biology, Bejing, China, April 2013. (poster)

Gong, T and **Szustakowski, Joseph D.**
"A statistical framework for computational deconvolution of RNA Sequencing data from complex samples."
ISMB 2012. (poster)

E. Khokhlovich, D. Tuch, **J. Szustakowski**.
"RAMP: A tool to search for imaging probes by disease pathways."
SNM 2011 Annual Meeting, San Antonio, TX, June 2011. (oral presentation by E. Khokhlovich)

Gong T, Hartmann N, Kohane IS, Brinkmann V, Linghu B, Staedtler F, Letzkus M, **Joseph D. Szustakowski.**
"Computational Deconvolution of Complex Transcriptomics Data from Clinical Trials."
RECOMB 2011, Vancouver Canada, March 2011. (poster)

Szustakowski JD, Kasif S, Weng Z.
"Less is more: towards an optimal universal description of protein folds."
ECCB/JBI Computational Biology, Madrid Spain, Sept 2005. (oral presentation by Z. Weng)

Szustakowski JD, Lee J-H, Marrese CA, Kosinski PA, Nirmala NR, Kemp D.
"Identification of novel pathway regulation during differentiation of myogenic progenitor cells."
FASEB Conference on Skeletal Muscle Satellite and Stem Cells, Tuscon Arizona, June 2005. (poster)

Zheng Y, **Szustakowski JD**, Roberts R, Kasif S.
"Computational identification of operons in microbial genomes."
The Sixth Annual International Conference on Research in Computational Molecular Biology, Washington D.C., April 2002. (poster)

Szustakowski JD and Weng Z.
"A protein structure alignment algorithm: statistics and application to uncovering similarities among different folds."
Forty-fifth Annual Meeting of the Biophysical Society, Boston, MA, February 2001. (poster)

Resendes BL, Robertson NG, **Szustakowski JD**, Resendes RJ, Weng Z, and Morton CC.
"Characterization of additional cochlear ESTs for gene discovery in the auditory system."
50th Annual Meeting of the American Society of Human Genetics, Philadelphia, PA, October 2000. (poster)

Szustakowski JD and Weng Z.
"Protein structure alignment using a genetic algorithm."
Thirteenth Annual Symposium of the Protein Society, Boston, MA, July 2000. (poster)

Honors and Awards

Enterprise Level Awards

BMS, **James B.D. Palmer Award for Excellence in Drug Development**, for Tumor Mutational Burden (TMB) as a predictive biomarker, 2018

Privileged to be part of a team recognized for their scientific contributions to Oncology Precision Medicine via the ***most prestigious Drug Development award granted by BMS***

BMS, **Leveraging Technology (LevTech)**, for Reproducible Research Environment, 2017

Team was granted the ***most prestigious BMS technology award*** *for their innovative Reproducible Research system*

Novartis, **Global IT Award for Innovation**, Pathway Analysis Toolkit, 2007

Awarded for the systematic application of pathway-centric analysis methods to ALL microarray experiments conducted at NIBR

Additional Awards

BMS, Bravo Ovation Award, for contributions to the 2023 II-ON Symposium
BMS, Bravo Ovation Award, for contributions to Quantum Leap strategic refresh, 2022
BMS, Bravo Celebrate Award, 2017
BMS, Bravo Ovation Award, 2017
BMS, Bravo Well Done Award, 2017
BMS, Bravo Encore Award, 2017
BMS, Bravo Celebrate Award, 2016

Novartis, NRG Spotlight Award, Tech Talk, 2014
Novartis, NRG Spotlight Award, Quality Assurance Audit, 2014
Novartis, NRG Star Award, Mentoring, 2014
Novartis, NRG Star Award, Internal People of Translational Medicine Presentation, 2014
Novartis, NRG Star Award, Diversity and Inclusion, 2014
Novartis, NRG Spotlight Award, Microarray Lab QC Tools, 2013
Novartis, NRG Star Award, Internal Presentation, 2013
Novartis, NRG Spotlight Award, Level 2 Quality Team, 2013
Novartis, NRG Spotlight Award, Collaboration w/Necker Hospital, 2013
Novartis, NRG Fusion Team Award, R Transition, 2013
Novartis, NRG Spotlight Award, Ilaris/SJIA Submission, 2013
Novartis, NRG Star Award, Malaria NGS, 2013
Novartis, NRG Star Award, University of Leeds Collaboration, 2013
Novartis, NRG Fusion Team Award, Electronic Lab Notebook (ELN), 2013
Novartis, NRG Star Award, Unit Offsite Planning and Presentation, 2013
Novartis, NRG Star Award, Internal Presentation, 2013
Novartis, NRG Star Award, LMX1B Manuscript, 2013
Novartis, Team Award Winner, Lumiracoxib, 2008
Novartis, SPOT Award Winner, 2007
Novartis, Catalyst Award Winner, 2006
Novartis, Team Award Winner, Mitochondriogenesis, 2005

Dean's Fellow, Boston University, 1996-1997
Magna Cum Laude, State University of New York at Buffalo, 1995
Presidential Honors Program, State University of New York at Buffalo, 1991-1995