Alex Handler Wagner, Ph.D. – Curriculum Vitae

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Date of Birth	16^{th} June 1985	**Email**	Alex.Wagner@nationwidechildrens.org

Personal Profile

My ongoing work is to integrate genomic evidence into knowledge used for interpreting genomic variation. My research program is defined by the development of several precision medicine web tools, for aggregating and disseminating genomic knowledge particularly in the cancer domain. These include the Drug-Gene Interaction Database, the database of Clinical Interpretations of Variants in Cancer (CIViC) knowledgebase, and the VICC meta-knowledgebase. My community science research program requires active engagement in professional consortia, and I currently hold advisory or leadership roles in several organizations, including the Variant Interpretation for Cancer Consortium (VICC) (as Director), the Global Alliance for Genomics and Health (GA4GH) (steering committee), the Atlas for Variant Effects Alliance (CVI and DCD committees), and ClinGen (Somatic Cancer, Cancer Variant Interpretation, and Data Platform work groups). I also co-led the development of the GA4GH Variation Representation Specification, and led the 2021 manuscript first describing that work. Ongoing support for this research is provided by a Genomic Innovator Award from the National Human Genome Research Institute.

Research Experience

Aug 2020 - Present Institute for Genomic Medicine, Nationwide Children's Hospital
Principal Investigator

Aug 2020 - Present Departments of Pediatrics and Biomedical Informatics, the Ohio State University
Assistant Professor

Jun 2019 - Jul 2020 Division of Oncology, Washington University in St. Louis
Instructor
Advisors: Obi L. Griffith, Ph.D.; Timothy J. Ley, M.D.

Jan 2015 - May 2019 McDonnell Genome Institute, Washington University in St. Louis
Postdoctoral Research Scholar
Advisors: Obi L. Griffith, Ph.D.; Ramaswamy Govindan, M.D.

Aug 2010 - Dec 2014 Coordinated Laboratory for Computational Genomics, University of Iowa
Graduate Research Assistant
Advisors: Terry A. Braun, Ph.D.; Edwin M. Stone, M.D., Ph.D.

Education

2010-2014 Ph.D, Computational Genetics - University of Iowa
GPA: 3.90

2010-2013 Graduate Certificate, Bioinformatics - University of Iowa
GPA: 3.96

2010-2013 BS, Biology (Minor: Mathematics) - Iowa State University
GPA: 3.51 - *Cum Laude*

Peer-reviewed Publications

*Denotes Co-First Authorship

1. M Cannon, J Steveonson, K Stahl, R Basu, ..., **AH Wagner** (2024) "DGIdb 5.0: rebuilding the drug-gene interaction database for precision medicine and drug discovery platforms". *Nucleic Acids Research*.
2. M Cannon, J Stevenson, K Kuzma, S Kiwala, JL Warner, ..., **AH Wagner** (2023) "Normalization of drug and therapeutic concepts with Thera-Py". *JAMIA Open*.
3. D Danis, JOB Jacobsen, **AH Wagner**, T Groza, ..., PN Robinson (2023) "Phenopacket-tools: Building and validating GA4GH Phenopackets". *PloS one*.
4. MS Ladewig, JOB Jacobsen, **AH Wagner**, D Danis, ..., PN Robinson (2023) "GA4GH Phenopackets: A Practical Introduction". *Advanced Genetics*.
5. K Krysiak, ..., **AH Wagner**, M Griffith, OL Griffith (2023) "CIViCdb 2022: evolution of an open-access cancer variant interpretation knowledgebase". *Nucleic Acids Research*.
6. KP Dalton, ..., **AH Wagner** (2022) "Accessing clinical-grade genomic classification data through the ClinGen Data Platform". *Pacific Symposium on Biocomputing*.
7. W Goar, ..., **AH Wagner** (2022) "Development and application of a computable genotype model in the GA4GH Variation Representation Specification". *Pacific Symposium on Biocomputing*.
8. Y Zhao, M Brush, C Wang, **AH Wagner**, H Liu, RR Freimuth (2022) "Leveraging a pharmacogenomics knowledgebase to formulate a drug response phenotype terminology for genomic medicine". *Bioinformatics*.
9. O Neumann, ..., **AH Wagner**, A Vogel, P Schirmacher, A Stenzinger, D Kazdal (2022) "Genomic architecture of FGFR2 fusions in cholangiocarcinoma and its implication for molecular testing". *British Journal of Cancer*.
10. JOB Jacobsen, ..., **AH Wagner**, JL Warner, C Weiland, MA Haendel, PN Robinson (2022) "The GA4GH Phenopacket schema defines a computable representation of clinical data". *Nature Biotechnology*.
11. K Krysiak, ..., **AH Wagner**, OL Griffith, M Griffith (2022) "A community approach to the cancer-variant-interpretation bottleneck". *Nature Cancer*.
12. N Matentzoglu, ..., **AH Wagner**, CJ Mungall (2022) "A Simple Standard for Sharing Ontological Mappings (SSSOM)". *Database*.
13. P Horak, et al. (2022) "Standards for the classification of pathogenicity of somatic variants in cancer (oncogenicity): Joint recommendations of Clinical Genome Resource (ClinGen), Cancer Genomics Consortium (CGC), and Variant Interpretation for Cancer Consortium (VICC)". *Genetics in Medicine*.
14. S LaHaye, ..., **Alex H Wagner**, AK Eisfeld, K Mrózek, V Magrini, CE Cottrell, ER Mardis, RK Wilson, P White (2021) "Discovery of clinically relevant fusions in pediatric cancer". *BMC Genomics*.
15. **AH Wagner**, Y Akkari, M Li, A Roy, K Tsuchiya, G Raca (2021) "Recommendations for future extensions to the HGNC gene fusion nomenclature". *Leukemia*.
16. HL Rehm, et al. (2021) "GA4GH: International policies and standards for data sharing across genomic research and healthcare". *Cell Genomics*.
17. **AH Wagner**, L Babb, ..., RK Hart (2021) "The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification". *Cell Genomics*.
18. A Stenzinger, ..., **AH Wagner**, F Tacke, D Capper, KR Müller, F Klauschen (2021) "Artificial intelligence and pathology: From principles to practice and future applications in histomorphology and molecular profiling". *Seminars in cancer biology*.
19. SL Freshour*, S Kiwala*, KC Cotto*, ..., **AH Wagner** (2021) "Integration of the Drug–Gene Interaction Database (DGIdb 4.0) with open crowdsource efforts". *Nucleic Acids Research*.

20. **AH Wagner**, B Walsh, G Mayfield, ..., M Griffith, OL Griffith, A Margolin (2020) "A harmonized meta-knowledgebase of clinical interpretations of somatic cancer genomic variants". *Nature Genetics*.

21. S Rao, B Pitel, **AH Wagner**, ..., M Griffith, OL Griffith, S Madhavan (2020) "Collaborative, Multidisciplinary Evaluation of Cancer Variants Through Virtual Molecular Tumor Boards Informs Local Clinical Practices". *JCO Clinical Cancer Informatics*.

22. **AH Wagner***, S Kiwala*, AC Coffman, JF McMichael, KC Cotto, TB Mooney, EK Barnell, K Krysiak, AM Danos, OL Griffith, M Griffith (2020) "CIViCpy: a Python Software Development and Analysis Toolkit for the CIViC knowledgebase". *JCO Clinical Cancer Informatics*.

23. G Alterovitz, B Heale, J Jones, D Kreda, F Lin, L Liu, X Liu, KD Mandl, DW Poloway, R Ramoni, **AH Wagner**, JL Warner (2020) "FHIR Genomics: enabling standardization for precision medicine use cases". *npj Genom Med*.

24. AM Danos*, K Krysiak*, EK Barnell*, ..., **AH Wagner**, S Madhavan, M Griffith, OL Griffith (2019) "Standard operating procedure for curation and clinical interpretation of variants in cancer". *Genome Med*.

25. EK Barnell, P Ronning, KM Campbell, K Krysiak, BJ Ainscough, C Ramirez, N Spies, J Kunisaki, ZL Skidmore, F Gomez, L Trani, M Matlock, **AH Wagner**, SJ Swamidass, M Grifith, OL Griffith (2019) "Standard operating procedure for somatic variant refinement of tumor sequencing data". *Genetics in Medicine*.

26. **AH Wagner***, S Devarakonda*, ZL Skidmore, K Krysiak, A Ramu, L Trani, J Kunisaki, ..., M Griffith, OL Griffith, R Govindan (2018) "Recurrent WNT Pathway Alterations are Frequent in Relapsed Small Cell Lung Cancer". *Nature Communications*.

27. BJ Ainscough*, EK Barnell*, KM Campbell, **AH Wagner**, TE Rohan, R Govindan, M Griffith, ER Mardis, SJ Swamidass, OL Griffith (2018) "A deep learning approach to automate refinement of somatic variant calling from cancer sequencing data". *Nature Genetics*.

28. AM Danos*, DI Ritter*, **AH Wagner**, K Krysiak, ..., S Kulkarni, M Griffith, S Madhavan, OL Griffith (2018) "Adapting crowdsourced clinical cancer curation in CIViC to the ClinGen minimum variant level data community-driven standards". *Human Mutation*.

29. K Cotto*, **AH Wagner***, YY Feng, S Kiwala, AC Coffman, G Spies, A Wollam, NC Spies, OL Griffith, M Griffith (2017) "DGIdb 3.0: a redesign and expansion of the drug-gene interaction database". *Nucleic Acids Research*.

30. M Griffith, NC Spies, K Krysiak, JF McMichael, AC Coffman, AM Danos, BJ Ainscough, CA Ramirez, DT Rieke, L Kujan, EK Barnell, **AH Wagner**, ..., OL Griffith (2017) "CIViC is a community knowledgebase for expert crowdsourcing the clinical interpretation of variants in cancer". *Nature Genetics*.

31. BJ Ainscough, M Griffith, AC Coffman, **AH Wagner**, J Kunisaki, MNK Choudhary, JF McMichael, RS Fulton, RK Wilson, OL Griffith, ER Mardis (2016) "DoCM: a database of curated mutations in cancer". *Nature methods*.

32. M Griffith, OL Griffith, K Krysiak, ZL Skidmore, MJ Christopher, JM Klco, A Ramu, TL Lamprecht, **AH Wagner**, ..., TJ Ley (2016) "Comprehensive genomic analysis reveals FLT3 activation and a therapeutic strategy for a patient with relapsed adult B-lymphoblastic leukemia". *Experimental hematology*.

33. ZL Skidmore, **AH Wagner**, R Lesurf, KM Campbell, J Kunisaki, OL Griffith, M Griffith (2016) "GenVisR: Genomic Visualizations in R". *Bioinformatics*.

34. **AH Wagner**, AC Coffman, BJ Ainscough, NC Spies, ZL Skidmore, KM Campbell, K Krysiak, D Pan, JF McMichael, JM Eldred, JR Walker, RK Wilson, ER Mardis, M Griffith, OL Griffith (2016) "DGIdb 2.0: mining clinically relevant drug/gene interactions". *Nucleic Acids Research*.

35. SS Whitmore, **AH Wagner**, AP DeLuca, AV Drack, EM Stone, BA Tucker, S Zeng, TA Braun, RF Mullins, TE Scheetz (2014) "Transcriptomic analysis across nasal, temporal, and macular regions of human neural retina and RPE/choroid by RNA-Seq". *Experimental Eye Research*.

36. TP Sharma, CM McDowell, Y Liu, **AH Wagner**, D Thole, BP Faga, RJ Workinger, TA Braun, AF Clark (2014) "Optic nerve crush induces spatial and temporal gene expression patterns in retina and optic nerve of BALB/cJ mice". *Molecular Neurodegeneration*.

37. TA Braun, RF Mullins, **AH Wagner**, J Andorf, R Johnston, B Bakall, AP DeLuca, G Fisherman, R Weleber, A Cideciyan, S Jacobson, V Sheffield, B Tucker, EM Stone (2013) "Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease". *Human Molecular Genetics*.

38. **AH Wagner**, KR Taylor, AP DeLuca, TL Casavant, RF Mullins, EM Stone, TE Scheetz, TA Braun (2013) "Prioritization of Retinal Disease Genes: An Integrative Approach". *Human Mutation*.

39. **AH Wagner**, VN Anand, W Wang, JE Chatterton, D Sun, AR Shepard, N Jacobson, L Pang, AP DeLuca, TL Casavant, TE Scheetz, RF Mullins, TA Braun, AF Clark (2013) "Exon-level expression profiling of ocular tissues". *Experimental Eye Research*.

40. AP DeLuca, **AH Wagner**, KR Taylor, B Faga, D Thole, VC Sheffield, ..., TA Braun (2011) "Sequencing and disease variation detection tools and techniques". *IEEE/ACS International Conference on Computer Systems and Applications (AICCSA)*.

Other Published Works

1. JA Arbesfeld, EY Da, ..., **AH Wagner** (2023) "Mapping MAVE data for use in human genomics applications". *bioRxiv*. **[Manuscript Preprint]**

2. **AH Wagner**, et al. (2019) "ga4gh/vr-spec: 1.0 GA4GH Approved". *Global Alliance for Genomics and Health*. **[Technical Standard]**

3. **AH Wagner**, K Krysiak, KM Campbell, EK Barnell (2019) "Cancer Genomics for the Clinician: Tumor Heterogeneity". *Springer Publishing Group*. **[Book Chapter]**

Funding: Research and Fellowship Grant Awards

2024 - Present — **NIH NHGRI U24 Competitive Subaward - GREGoR Consortium**
U24HG011746 - *Role: Subaward PI*

2023 - Present — **NIH NCI U24 - ClinGen Somatic Biomedical Knowledgebase**
U24CA275783 - *Role: Co-I*

2022 - Present — **NIH OD U54 - Bridge2AI BRIDGE Center (Standards)**
U54HG012513 - *Role: Standards Core co-Lead*

2022 - 2023 — **NIH NHGRI R35 Supplement - AI-Ready gnomAD**
R35HG011949 - *Role: PI*

2021 - Present — **NIH NHGRI R35 - Genomic Innovator**
R35HG011949 - *Role: PI*

2019 - Present — **NIH NHGRI K99/R00 - Knowledge Harmonization**
R35HG011949 - *Role: PI*

2017 - 2019 — **NIH NCI F32 - Cancer Genomics**
R35HG011949 - *Role: PI*

Funding: Training Awards

2016 - 2017 — **NIH NCI T32 Postdoctoral Training - Cancer Biology**
T32CA009547 - *Role: Trainee*

2013 - 2014 — **NIH NCI T32 Predoctoral Training - Genetics**
T32GM008629 - *Role: Trainee*

2011 - 2013 — **NIH NCI T32 Predoctoral Training - Bioinformatics**
T32GM082729 - *Role: Trainee*

Research Recognition Awards

2021 Genomic Innovator Award
National Human Genome Research Institute, National Institutes of Health

National recognition awarded for early career success in consortia-driven scientific research.

2018 ICTS Precision Medicine Abstract Award
Washington University in St. Louis

The most outstanding research was selected from more than 75 applicants at the Washington University Precision Medicine Symposium to present to Eric Green, the director of the National Human Genome Research Institute (NHGRI).

2015 D.C. Spriestersbach Dissertation Prize Nominee
University of Iowa, Genetics Program Nomination

This biennial award recognizes excellence in doctoral research. Each of the twenty biological/life sciences programs at the University of Iowa nominates one dissertation submitted between July 1, 2013 and June 30, 2015 to compete for the award.

2013 Outstanding Student Research Award - Bioinformatics and Computational Biology
University of Iowa, College of Engineering

This annual award recognizes a single student in the College of Engineering for exemplary research in the fields of bioinformatics and computational biology.

Travel and Trainee Awards

2019 **Trainee Abstract Award**
Curating the Clinical Genome Annual Conference - *Washington D.C.*

2018 **1st Place Student/Trainee Travel Award**
Cancer Genomics Consortium Annual Conference - *Nashville, TN*

2018 **AACR-Bristol Myers Squibb Scholar-in-Training Award**
American Association for Cancer Research - *Chicago, IL*

2013 **NSF Travel Grant**
Intelligent Systems for Molecular Biology Annual Conference - *Berlin, Germany*

2013 **Graduate Student Senate Travel Grant**
Association for Research in Vision and Ophthalmology Annual Conference - *Seattle, WA*

Presentations

†Oral Presentation; ‡Invited Speaker; *Upcoming

Mar 2024 ‡*Advancing AI-readiness of the gnomAD database with GA4GH genomic knowledge standards*
NIH ODSS AI Supplement PI Meeting - Virtual

Nov 2023 ‡*Application of the VICC Oncogenicity Framework in a Pediatric Setting* ***(Declined; medical)***
AMP Annual Conference - Salt Lake City, UT

Nov 2023 ‡*Variation Representation* ***(Declined; medical)***
AMP Annual Conference - Salt Lake City, UT

Jun 2023 ‡*Genomic Knowledge Standards*
EMQN Seminar Series - Virtual

Jan 2023 †*Accessing clinical-grade genomic classification data through the ClinGen Data Platform*
Pacific Symposium on Biocomputing - Waimea, HI

Nov 2022 ‡ *Gene (Fusion) Product Nomenclature*
AMP Annual Conference - Phoenix, AZ

Sep 2022 ‡ *A starter kit for interoperable genomic knowledge*
GA4GH Plenary - Barcelona, Spain

Sep 2022 ‡ *Decoding the standards for genomic data sharing*
NIH BISTI Seminar - Virtual

Aug 2022 † *A Unified Framework for Gene Fusion Representation*
Cancer Genomics Consortium Annual Conference - St. Louis, MO

Jul 2022 ‡ *The GA4GH Variation Representation Specification*
International Graph Genome Symposium 2022 Conference - Ascona, Switzerland

Jun 2022 † *Cross-consortia Recommendations for a Unifying Framework Supporting Curation and Representation of Gene Fusions*
ClinGen Curating the Clinical Genome - Virtual

Apr 2022 *The GA4GH Variation Representation Specification*
American Association for Cancer Research Annual Conference - New Orleans, LA

Jan 2022 ‡ *Pitfalls of Contemporary Genomic Variation Representation (TERI talk)*
Pacific Symposium on Biocomputing 2022 - Waimea, HI

Nov 2021 † *The GA4GH Variation Representation Specification (VRS): a Computational Framework for the Precise Representation of Molecular Variation*
AMIA Annual Conference - San Diego, CA

Oct 2021 ‡ *Genomic Knowledge Standards*
GA4GH 9th Plenary Connect - Virtual

Sep 2021 ‡ *Phenopackets VRS Integration*
GA4GH 9th Plenary - Virtual

Aug 2021 † *Multi-consortia initiative to standardize the representation and curation of oncogenic fusions*
Cancer Genomics Consortium Annual Conference - Virtual

Jun 2021 † *The ClinGen Data Platform is Driving Interoperability of Expert Evidence through Standards for Computable Genomic Knowledge*
ClinGen Retreat - Virtual

May 2021 † *Interoperability of ClinGen Expert Evidence through Standards for Computable Genomic Knowledge*
ClinGen Curating the Clinical Genome - Virtual

Apr 2021 *Advancing Genomic Medicine through Standards for Computable Genomic Knowledge*
NHGRI Research Conference - Virtual

Apr 2021 *A standard operating procedure for the curation of gene fusions*
AACR Annual Conference - Virtual

Mar 2021 ‡ *Clinical Interoperability of Variant Evidence*
Genomics in Health Implementation Forum - Virtual

Mar 2021 ‡ *VRS 1.3 Planning and Implementation Guidance*
GA4GH Implementation Connect - Virtual

Mar 2021 ‡ *VCF / VRS Alignment*
GA4GH Implementation Connect - Virtual

Dec 2020 ‡ *ASH Precision Medicine Working Group: Integrating Variant Interpretations*
ASH Annual Conference - Virtual

Nov 2020 † *Clinical variant harmonization and standards for knowledgebase interoperability (panelist)*
AMIA Annual Conference - Virtual

Nov 2020 † *Clinical variant harmonization and standards for knowledgebase interoperability (panelist)*
AMIA Annual Conference - Virtual

Sep 2020 ‡ *VCF / VRS Alignment*
GA4GH 8th Plenary - Virtual

Sep 2020 ‡ *VA & VR Implementations: Practical Considerations and Emerging Draft Standards*
GA4GH 8th Plenary - Virtual

Aug 2020 † *Integrating Cytogenomic and Sequence Variants*
Cancer Genomics Consortium Annual Conference - Virtual

Apr 2020 † *Data harmonization standards from the Variant Interpretation for Cancer Consortium*
AACR Annual Conference - Virtual

Nov 2019 ‡ *Computational representation of patient data (panelist)*
AMIA Annual Conference - Washington, D.C.

Aug 2019 ‡ *Community resources for clinical variant classification in cancers*
Cancer Genomics Consortium Annual Conference - Nashville, TN

Aug 2019 † *A new somatic variation model enables precise search strategies for clinical interpretations of patient tumors*
Cancer Genomics Consortium Annual Conference - Nashville, TN

May 2019 † *Somatic variant curation standards enable improved identification of relevant clinical interpretations for tumor variants*
Curating the Clinical Genome - Washington, D.C.

Apr 2019 ‡ *Variant Interpretation for Cancer Consortium: Workstream Goals 2019*
GA4GH Implementation Connect - Hinxton, United Kingdom

Mar 2019 *Cancer genome interpretation with CIViCpy*
AACR Annual Conference - Atlanta, GA

Mar 2019 ‡ *The Variant Interpretation for Cancer Consortium, a Genomic Knowledge Workstream Driver Project of the GA4GH*
GA4GH-AMED Symposium - Sendai, Japan

Oct 2018 ‡ *The Variant Interpretation for Cancer Consortium*
GA4GH 6th Annual Plenary - Basel, Switzerland

Aug 2018 † *Coordinating variant interpretation knowledgebases improves clinical interpretation of genomic variants in cancers*
Cancer Genomics Consortium Annual Conference - Nashville, TN

Apr 2018 *Standardization and coordination of variant interpretation knowledgebases improves clinical genome actionability*
American Association for Cancer Research - Chicago, IL

Jun 2016 *The Drug Gene Interaction Database*
Curating the Clinical Genome - Hinxton, United Kingdom

Feb 2016 *The Drug Gene Interaction Database*
AGBT Annual Conference - Orlando, FL

Jul 2013 *Positive and Unlabeled Learning for Prioritization (PULP)*
ISMB Annual Conference - Berlin, Germany

May 2013 *Positive and Unlabeled Learning for Prioritizing Candidate Variants in Retinal Degenerative Diseases*
ARVO Annual Conference - Seattle, WA

May 2013 *Positive and Unlabeled Learning for Prioritizing Candidate Variants in Retinal Degenerative Diseases*
BICB Industry Symposium - Minneapolis, MN

May 2012 *RNA Sequencing for Identification of Genetic Factors in Retinal Disease*
ARVO Annual Conference - Ft. Lauderdale, FL

Aug 2011 *Using RNA Sequencing To Identify And Isolate Causative Genetic Factors In Retinal Disease*
ISU / UIowa Joint Bioinformatics Retreat - Ames, IA

Referees

Name	Bill Lumbergh	**Name**	Michael ”Big Mike” Tucker
Company	Initech Inc.	**Company**	Burbank Buy More
Position	Vice President	**Position**	Store Manager
Contact	bill@initech.com	**Contact**	mike@buymore.com