Priv.-Doz. Dr. Barbara Vona

Group Leader

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DESCRIPTION OF MY ACADEMIC CAREER

I completed my Bachelor and Master degrees at the University of Arizona (USA) before arriving in Germany to specialize in the genetics of human hearing loss. I completed my PhD at the University of Würzburg (2014) on the topic “Molecular characterization of genes involved in hearing loss”. During my post doc (University of Würzburg) until 2018, I was recruited as a Junior Group Leader at the Tübingen Hearing Research Center (University of Tübingen). In Tübingen, I contributed my expertise to a diverse team of hearing research experts while absorbing in-depth knowledge in hearing science and mouse models of deafness. In 2019, I completed my Habilitation titled “Identification and characterization of genes in connection with hereditary hearing disorders”. In 2021, I joined the Institute for Auditory Neuroscience at the University Medical Center Göttingen, starting my present group. I came with a vision that shaped and motivated contributions to novel frameworks for exciting science while enhancing patient care. I conceived and established the Otoferlin and *CABP2* Patient Registries. These were particularly impactful, as they are first gene-based registries in the world for isolated forms of hearing loss. In parallel, I founded the “Center for Rare Hearing Disorders” at the Center for Rare Diseases Göttingen, improving multidisciplinary care while contributing structures for research and clinical trials. My vision is to build a national hearing loss registry framework to support genetic stratification for clinical trials and improved care.

I am an editorial board member for *The Journal of the Association for Research in Otolaryngology*, *Scientific Reports* and *Molecular Syndromology* and serve as the sole German-based member for the international ClinGen Hearing Loss Variant and Gene Curation Expert Panels. I am a reviewer for well-known journals, such as *Nature Communications*, *Nucleic Acids Research*, *The New England Journal of Medicine*, and *JAMA Neurology*. I serve the Communications Committee of the *Association for Research in Otolaryngology*. I have given invited talks at the German Society of Oto-Rhino-Laryngology, Head and Neck Surgery, DGA/ADANO, and the European Society of Human Genetics. I have supervised 35 trainees, from bachelor to PhD and medical student level and have extensive experience in training young scientists and medical professionals.

Due to synergistic opportunities, I was recently able to showcase my specialization in auditory human genomics and was accepted into the Heisenberg Programme at the DFG that started in April 2025. In addition to being offered a W2 tenure-track professorship at University Medical Center Göttingen, I have used the opportunity to enrich my skill set in the genomics of hearing loss at Harvard Medical School and MIT, which is from where I submit this application. This will enhance my profile and improve patient-oriented research and care for patients in Germany and worldwide.

RESEARCH STATEMENT

I am interested in characterizing human hereditary disorders using modern molecular methods. My longstanding interest involves identifying novel hearing loss-associated genes by analyzing exome or genome sequencing data from undiagnosed families from Germany as well as via my extended collaborative network throughout the world who have access to populations conducive to novel gene discovery (i.e. large consanguineous families or families with multiple generations of individuals co-segregating hearing loss). My work contributes to the functional characterization of novel genes and variants of uncertain significance that are implicated in hereditary hearing loss using mouse and cell-based approaches to study gene function and variant effects, respectively. Considering emerging gene therapies, this work is imperative to correctly annotate genes and variants involved in the basic molecular physiology of hearing loss and deafness. I am also engaged in patient outreach demonstrated by the creation of patient registries for otoferlin (*OTOF*) and *CABP2*-associated hearing loss to support long term efforts on the Göttingen Campus that include possible future clinical trials.

I am broadening a synergistic line of research that involves developing new high-throughput methods to characterize variant effects on a gene-by-gene basis to overcome conventional low-throughput *in vitro* approaches. This work is expected to make significant strides to resolve variants of uncertain significance throughout entire gene sequences for non-synonymous and putative splicing variants that require laborious studies not generally done in routine diagnostic settings. I am currently generating a preliminary dataset to be used for future assay "benchmarking" to characterize a subset of coding and non-coding and variants in *OTOF* that are predicted to cause aberrant splicing, as well as variants in *CLRN1* that causes Usher syndrome (causing hearing and vision impairment). These projects have been registered with the MAVE (Multiplexed Assays of Variant Effect) Registry to leverage the expertise of the collaborative Atlas of Variant Effects community already engaged in the development and validation of these cutting-edge assays for different target genes. This work is supported by CRC 1690. Longer-term interests involve being a key member of an interdisciplinary translational team for multiple genetic targets throughout the experimental and pre-clinical/clinical pipelines to advance therapies for preventing or treating human hearing loss at the Göttingen Campus.

RESEARCH EXPERIENCE

Visiting Scientist

Since 09/2025 Department of Obstetrics and Gynecology, Brigham and Women's Hospital, Harvard Medical School, Boston, MA, USA

Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, Cambridge, MA, USA

Group Leader

Since 06/2022 Institute for Auditory Neuroscience and InnerEarLab, University Medical Center Göttingen, Göttingen, Germany

09/2021-03/2025 Institute of Human Genetics, University Medical Center Göttingen, Göttingen, Germany

Impact:

- Heisenberg Fellow (DFG) (Since 04/2025)
- Expanded committee and scientific network leadership involvement: Collaborative Network for European Clinical Trials for Children (conect4children) in Ear, Nose, and Throat and Genetics/Cell Therapy; ClinGen AVE Alliance Functional Working Group; Coordinator of the ClinGen Splicing Working Group; Communications Committee, Association for Research in Otolaryngology, Management team of the European Consortium of the Inner Ear (ECIE)
- Founded a new Center for Rare Hearing Disorders at the Center for Rare Diseases Göttingen; Founding meeting: November 2024
- Designed a SPATA patient registry for individuals with *SPATA5* (*AFG2A*) and *SPATA5L1* (*AFG2B*) diagnoses, hosted at the Coordination of Rare Diseases at Sanford (CoRDS). Release date: September 2024
- Launch of the *CABP2* Patient Registry (ClinicalTrials.gov ID: NCT06680934)
- Launch of the Otoferlin Patient Registry (ClinicalTrials.gov ID: NCT05946057)
- Lead and supervise research projects with undergraduate/graduate students
- Participate in teaching of undergraduate/graduate courses
- Joined as a Genomics England PanelApp Reviewer for Auditory Neuropathy and Monogenic Hearing Loss Panels
- Continue to participate in monthly ClinGen Hearing Loss Gene and Variant Curation Expert Panel meetings
- Manage and lead a growing number of international collaborations

Fellow

09/2021-05/2022 Institute for Auditory Neuroscience and InnerEarLab, University Medical Center Göttingen, Göttingen, Germany

Impact:

- Acquired third-party funding: German Research Foundation “Eigene Stelle” for launching my independent research group
- Establishment of a Twitter-based science communication and patient outreach page called @Otoferlin_Group designed to disseminate information for families with otoferlin genetic diagnoses (02/2022 – present)
- Initiated the first steps to establish a new Center for Rare Hearing Disorders at the Center for Rare Diseases Göttingen
- Participated in teaching of undergraduate/graduate courses

Junior Group Leader

08/2018-08/2021 Department of Otolaryngology—Head & Neck Surgery, Tübingen Hearing Research Centre (THRC), Eberhard Karls University Tübingen, Tübingen, Germany

Impact:

- Acquired various third-party funding and independently ran scientific projects
- Study leader for ethics approved study involving humans
- Served as a lead and educator for clinicians in the dissemination of genetic diagnostic results; participated in monthly meetings about patient genetic diagnostic results
- Joined the ClinGen Hearing Loss Gene and Variant Curation Expert Panel
- Participated in teaching of undergraduate/graduate courses

Post-doctoral Fellow

07/2014-08/2018 Human Genetics, University of Würzburg, University of Würzburg, Würzburg, Germany

Impact:

- Lead a team under a mentor who allowed me substantial scientific independence
- Began several successful independent international collaborations with key scientists who continue to serve as keystones to my research
- Served as diagnostic leader for hearing impairment molecular genetic diagnostics
- Filed and maintained ethical approval for experiments involving humans as study leader
- Gained grant writing experience
- Established zebrafish techniques for auditory phenotyping
- Joined as a variant curator for the Leiden Online Variation Database 3.0 (LOVD) for several hearing loss genes
- Participated in teaching of undergraduate/graduate courses

ACADEMIC EDUCATION

2011-2014 PhD (Dr. rer. nat.), Human Genetics, University of Würzburg, Würzburg, Germany
Dissertation: Molecular Characterization of Genes Involved in Hearing Loss
Grade: *Magna cum laude*
Supervisor: Univ. Prof. Dr. med. Thomas Haaf

2006-2008 Master of Science, Pathobiology, University of Arizona, Tucson, AZ, USA
Thesis: Characterization of Genetic Elements of the Antibiotic Resistant *erm*(X) Gene in *Arcanobacterium pyogenes*
Supervisor: Dr. Stephen Billington

2002-2006 Bachelor of Science, Microbiology (Honors), University of Arizona, Tucson, AZ, USA

SCIENTIFIC DEGREES AND ACADEMIC QUALIFICATIONS

2022 Umhabilitation, Human Genetics, University Medical Center Göttingen, Germany

2019 Habilitation, Human Genetics, University of Würzburg, Germany

2014 PhD (Dr. rer. nat.), Human Genetics, University of Würzburg, Germany

2008 Master of Science, Pathobiology, University of Arizona, Tucson, AZ, USA

2006 Bachelor of Science, Microbiology, University of Arizona, Tucson, AZ, USA

SUPERVISING AND MENTORING EXPERIENCE

Dates	University	Level	No. # Trainees[1]
20/08/2025 – present	University of Göttingen	Master Lab Rotation	35
26/05/2025 – 18/07/2025	University of Göttingen	Master Lab Rotation	34
19/05/2025 – present	University of Göttingen	Master Thesis	27
28/04/2025 – 20/06/2025	University of Göttingen	Master Lab Rotation	33
01/04/2025 – present*	University Medical Center Göttingen	PhD Thesis	32
03/03/2025 – 02/05/2025	University of Göttingen	Master Lab Rotation	31
03/03/2025 – present	University of Hannover	Master Lab Rotation	30
02/17/2025 – present	University of Göttingen	Master Thesis	29
07/10/2024 – 01/11/2024	University of Göttingen	Master Lab Rotation	29
12/08/2024 – 13/09/2024	University Medical Center Göttingen	Bachelor Internship	28
15/07/2024 – 31/08/2024	University of Göttingen	Master Lab Rotation	27
04/03/2024 – present	University of Naples Federico II	Master Thesis	26
22/01/2024 – present**	University Medical Center Göttingen	MD Thesis	25
15/01/2024 – 04/11/2024	Anhalt University of Applied Science	Master Thesis	24
05/02/2024 – present**	University Medical Center Göttingen	MD Thesis	23
21/08/2023 – 08/09/2023	University Medical Center Göttingen	MD Thesis Rotation	23
26/07/2023 – 05/09/2023	University Medical Center Göttingen	Bachelor Practical	22
03/04/2023 – present**	University Medical Center Göttingen	MD Thesis	21
15/09/2023 – 27/05/2024	University Medical Center Göttingen	Master Thesis	20
15/02/2023 – 18/04/2023	University Medical Center Göttingen	Master Lab Rotation	20
05/12/2022 – 20/02/2023	University Medical Center Göttingen	Master Lab Rotation	19
01/11/2022 – 12/05/2023	University Medical Center Göttingen	Master Thesis	18
01/06/2022 – present**	University Medical Center Göttingen	PhD Thesis	17
03/03/2021 – 03/31/2021	University of Tübingen	MD Thesis Internship	16
16/11/2020 – 11/02/2021	University of Tübingen	Master Thesis Internship	15
07/05/2020 – 01/05/2024***	Cairo University	PhD Thesis	14
08/01/2020 – 31/07/2020	Hochschule Coburg	Master Thesis	13
01/11/2019 – 24/06/2024**	University of Tübingen	PhD Thesis	12
15/06/2020 – 27/07/2020	University of Tübingen	Master Internship	11
07/10/2019 – 12/03/2020	University of Tübingen	Bachelor Thesis	11
01/10/2017 – 31/07/2020*	Karolinska Institutet	PhD Thesis	10
01/02/2018 – 13/08/2018	University of Würzburg	Master Thesis	9
04/09/2017 – 06/01/2018	University of Würzburg	Internship (F2)	9
10/07/2017 – 10/01/2025	University of Würzburg	MD Thesis	8
17/10/2016 – 28/10/2016	Islamic Azad University	Practical Internship	7
01/06/2017 – 17/02/2022*	University of Würzburg	PhD Thesis	6
22/06/2016 – 01/02/2017	University of Würzburg	Master Thesis	6
01/04/2016 – 21/06/2016	University of Würzburg	Internship (F2)	6
01/12/2015 – 28/07/2016	Hochschule Coburg	Master Thesis	5
23/02/2015 – 08/07/2020	University of Würzburg	PhD Thesis	4
06/10/2014 – 27/10/2014	University of Würzburg	Pharmacy Internship	3
16/09/2013 – 29/07/2019	University of Würzburg	PhD Thesis	2
08/01/2013 – 17/07/2013	University of Würzburg	Master Thesis	2

14/05/2012 – 07/01/2013	University of Würzburg	Internship (F2)	2
29/08/2011 – 07/10/2011	University of Würzburg	Internship (F1)	1

[1]Intended to show students with multiple research stays to satisfy multiple degree requirements
*Co-supervisor and Doctoral Thesis Committee Member
**Supervisor and Doctoral Thesis Committee Member
***Doctoral Thesis Committee Member

DOCTORAL THESIS COMMITTEE REVIEWER

Date	University	Department
03/12/2023	University of Granada	Otology & Neurotology Genomic Medicine
27/09/2022	University of Granada	Otology & Neurotology Genomic Medicine
21/02/2020	University of Granada	Otology & Neurotology Genomic Medicine
25/02/2019	University of Granada	Otology & Neurotology Genomic Medicine

SECOND SUPERVISOR DOCTORAL THESIS COMMITTEE MEMBER

Dates	University	Department
08/2023 – present	University Medical Center Göttingen	Department of Otolaryngology
01/2023 – present	University Medical Center Göttingen	Institute for Auditory Neuroscience

TEACHING EXPERIENCE

Semester	University Medical Center Göttingen
Winter 2021/22, 2022/23, 2023/24	B.MM.014/M.MM.017 Auditory Neuroscience
Summer 2025	B.MM.014/M.MM.017 Auditory Neuroscience
Winter 2022/23, 2023/24, 2024/25	M.Bio 348, M.Bio 369, Human Genetics
Winter 2023/24, 2024/25	B.MM.106, Introduction to Molecular Medicine
Semester	**University of Tübingen**
Winter 2019/20, 2020/21, Summer 2021	Molecular Human Genetics (MSc)
Winter 2019/20	Neurobiological Practical Course – HEARING
Summer 2020	Sensory Systems – Basics and Principles II
Summer 2021	Novel Therapeutic Approaches for Sensory Disorders
Summer 2021	Genetic and Molecular Basis of Neural Diseases II
Summer 2021	Journal Club
Semester	**University of Würzburg**
Winter 2016/17, 2017/18, 2018/19, 2019/20, 2020/21 2021/22	Foundations of Human Genetics (MSc)
Winter 2015/16, 2016/17, 2017/18	F1 Human Genetics Practical Course (MSc)
Summer 2016, 2017, 2018	Student Seminar in Human Genetics (MSc)
Summer 2016, 2017, 2018	Tumor Genetics, Master-level
Semester	**University of Arizona**
Fall 2006	Microbiological Techniques (MIC421b) (BSc)
Spring 2005	General Biology II (MCB182L) (BSc)

MSc, Master-level; BSc, Bachelor-level

I regularly engage in research-oriented teaching by supervising Bachelor and Master students in my group for internships and rotations (e.g. M.Bio 319).

PROFESSIONAL TEACHING DEVELOPMENT COURSES

03/2019	Good Scientific Practice
07/2018	Examining and assessing in English
06/2018	Presenting to an English-speaking audience
05/2018	Presentation training in scientific practice
05/2018	Supporting international students
03/2018	Teaching and learning in the international classroom
03/2018	Supervising bachelor and master students
07/2017	From research to teaching—my role as a lecturer
06/2017	Decoding the disciplines: overcoming bottlenecks in teaching

TEACHING CERTIFICATE

07/2018	Certificate of Higher Education of the Bavarian Universities (Basic level)

GENE AND VARIANT CURATION WORK

Since 08/2022	Genomics England PanelApp Gene Reviewer for Auditory Neuropathy
Since 08/2022	Genomics England PanelApp Gene Reviewer for Hearing Loss
Since 03/2018	Variant curator for the Leiden Online Variation Database 3.0 (LOVD)

SCIENTIFIC NETWORKS, ADVISORY, AND EXPERT COMMITTEES

09/2025 – present	Coordinator and member, ClinGen Splicing Working Group
04/2025 – present	Collaborative Network for European Clinical Trials for Children (conect4children): Core Expert in Ear, Nose, and Throat; Extended Expert in Genetics and Cell Therapy
03/2025 – present	Member, ClinGen AVE Alliance Functional Working Group
04/2024 – present	Member of the Light2Treat Team I, Optogenetic Hearing Restoration at the Else Kröner Fresenius Center for Optogenetic Therapies
04/2024 – present	Member of the Disease Modeling Platform (Platform 3) at the Else Kröner Fresenius Center for Optogenetic Therapies
10/2024 – present	Management team, European Consortium of the Inner Ear
04/2019 – present	Member of the ClinGen Hearing Loss Variant Curation Expert Panel
04/2019 – present	Member of the ClinGen Hearing Loss Gene Curation Expert Panel

PROMOTION APPOINTMENT COMMITTEE REVIEWER

2023	Served as a committee member to review a candidate W3 Professor Clinic for Child and Adolescent Psychiatry and Psychotherapy at the University Medical Center Göttingen

2022 – 2023 Served as a committee member to review a candidate to be promoted from a W1-Professor (assistant professor) position to a tenure track W2-Professor (associate professor) position at the University Medical Center Göttingen

DIAGNOSTIC SERVICES

2014 – 2018 Leader of hearing loss molecular genetic diagnostic services
Institute of Human Genetics, University of Würzburg, Germany

PATIENT REGISTRY DEVELOPMENT

2024 – present Co-developed and launched the *CABP2* Registry in collaboration with members at the University Medical Center Göttingen, registered in ClinicalTrials.gov under ID NCT06680934

2024 – present Co-developed and launched the SPATA Registry for patients with a diagnosis of *SPATA5* (*AFG2A*) or *SPATA5L1* (*AFG2B*) in collaboration with The SPATA Foundation

2023 – present Co-developed and launched the Otoferlin (*OTOF*) Registry in collaboration with members at the University Medical Center Göttingen, registered in ClinicalTrials.gov under ID NCT05946057

RESEARCH APPROVAL

2024 – present Role: Project Leader
Ethics Commission approval for the study “Assessment of Current and Future Approaches to Address Variants of Uncertain Significance in Hearing and Ocular Genomics Domains”, University Medical Center Göttingen, Study ID: 12/8/24 An

2024 – present Role: Co-Project Leader
Ethics Commission approval for expansion of the “Hereditary Hearing Impairment Patient Registry,” for the *CABP2* Registry, University Medical Center Göttingen, Study ID: 17/8/22 v2, registered in ClinicalTrials.gov under ID NCT06680934

2023 – present Role: Co-Project Leader
Ethics Commission approved protocol for “Hereditary Hearing Impairment Patient Registry,” for the Otoferlin Registry, University Medical Center Göttingen, Study ID: 17/8/22 v1, registered in ClinicalTrials.gov under ID NCT05946057

2019 – 2021 Role: Project Leader
Ethics Commission protocol approved for “Clarification of the genetic causes of hereditary hearing impairment,” University of Tübingen, Study ID: 197/2019BO1

2015 – 2018 Role: Project Leader
Ethics Commission approved protocol for “Identification and Functional Characterization of Genes Involved in Congenital Hearing Loss,” University of Würzburg. Study ID: 46/15

BOARD MEMBERSHIP

2025 – present	Cure KARS – Laia Foundation, Independent Scientific Advisory Board
2024 – present	European Consortium of the Inner Ear, Management Board
2024 – present	The SPATA Foundation, Medical and Scientific Advisory Board

CONSORTIUM MEMBERSHIP

2024 – present	European Consortium of the Inner Ear Member
2017 – 2022	Academic partner for doctoral student supervision with the European School on Interdisciplinary Tinnitus Research (ESIT) network
2015 – 2018	Collaborative Member of the genetics work group of the TINNET (Tinnitus research network) consortium

EDITORIAL AND PEER REVIEW SERVICE

Editorial Board Member

04/2025 – present	*Scientific Reports*
07/2022 – present	*Journal of the Association for Research in Otolaryngology*
04/2015 – present	*Molecular Syndromology*

Editorial Reviewer

09/2018 – present	*Frontiers in Neuroscience*
09/2018 – present	*Frontiers in Psychology*

***Ad hoc* Peer Reviewer**

Since 08/2025	*American Journal of Human Genetics*
Since 05/2025	*MedComm*
Since 04/2025	*NAR Genomics and Bioinformatics*
Since 01/2025	*New England Journal of Medicine*
Since 11/2024	*Molecular Medicine*
Since 09/2024	*Molecular Therapy*
Since 09/2024	*Journal of Otolaryngology – Head & Neck Surgery*
Since 05/2024	*Journal of Medical Genetics*
Since 01/2024	*Therapeutic Advances in Rare Disease*
Since 06/2023	*Nature Communications*
Since 01/2023	*Annals of Clinical and Translational Neurology*
Since 11/2022	*Cell and Bioscience*
Since 07/2022	*European Journal of Clinical Investigation*
Since 06/2022	*British Journal of Ophthalmology*
Since 05/2022	*Annals of Neurology*

Since 02/2022 *Clinical and Translational Medicine*
Since 07/2021 *Molecular Neurobiology*
Since 06/2021 *Journal of Hearing Science*
Since 03/2021 *BMC Research Notes*
Since 01/2021 *eBioMedicine*
Since 11/2020 *Expert Review of Molecular Diagnostics*
Since 09/2020 *Molecular Genetics and Genomic Medicine*
Since 08/2020 *International Journal of Developmental Neuroscience*
Since 06/2020 *Genes*
Since 05/2020 *Journal of Clinical Laboratory Analysis*
Since 04/2020 *Frontiers in Genetics*
Since 03/2020 *JAMA* Neurology
Since 12/2019 *Frontiers in Neurology*
Since 12/2019 *Journal of Cellular and Molecular Medicine*
Since 10/2019 *Journal of Human Genetics*
Since 10/2019 *Clinical Genetics*
Since 09/2019 *Computer Methods and Programs in Biomedicine*
Since 06/2019 *European Journal of Medical Genetics*
Since 05/2019 *Cell and Tissue Research*
Since 04/2019 *Frontiers in Molecular Neuroscience*
Since 04/2019 *Naunyn-Schmiedeberg's Archives of Pharmacology*
Since 03/2019 *Frontiers in Pediatrics*
Since 03/2019 *Nucleic Acids Research*
Since 01/2019 *Acta Oto-Laryngologica*
Since 01/2019 *Cytogenetic and Genome Research*
Since 10/2018 *Human Genetics*
Since 10/2018 *Sensors*
Since 06/2018 *Journal of Molecular Medicine*
Since 06/2018 *Audiology and Neurotology*
Since 05/2018 *BMC Medical Genetics*
Since 11/2017 *Journal of Genetics*
Since 11/2017 *International Journal of Neonatal Screening*
Since 10/2017 *International Journal of Molecular Sciences*
Since 09/2017 *Journal of Pediatric Genetics*
Since 07/2017 *SM Otolaryngology*
Since 11/2016 *Scientific Reports*
Since 11/2016 *Orphanet Journal of Rare Diseases*

Since 10/2016	*Molecular Syndromology*
Since 10/2016	*American Journal of Medical Genetics Part A*
Since 09/2016	*Frontiers in Neuroscience*
Since 08/2016	*Hearing Research*
Since 05/2016	*Meta Gene*
Since 04/2016	*European Journal of Human Genetics*
Since 03/2016	*Genetic Testing and Molecular Biomarkers*
Since 01/2016	*Human Molecular Genetics*
Since 09/2015	*Journal of Clinical and Experimental Genetics*
Since 07/2015	*Journal of Translational Medicine*
Since 08/2014	*PLOS ONE*
Since 08/2013	*Gene*

AD HOC **GRANT PEER REVIEWER**

11/2024	Swiss National Science Foundation
09/2018, 04/2019, 05/2024	Action on Hearing Loss/Royal National Institute for Deaf People
03/2018	Institute of Physiology and Pathology of Hearing, Warsaw, Poland
06/2016	University of Sharjah

TOP FIVE RECENT PUBLICATIONS

Vona B, Wollnik B, Strenzke N, Pangršič T, Moser T. Is CABP2-Associated Hearing Loss (DFNB93) a Gene Therapy Target? Preclinical Progress and Patient Registry. 2025. ***MedComm*** 6(9):e70363. doi: 10.1002/mco2.70363. **Impact factor: 10.700**

Ni C*, Wei Y*, **Vona B***, Park D, Wei Y, Schmitz DA, Ding Y, Sakurai M, Ballard E, Li L, Liu Y, Kumar A, Xing C, Qin S, Kim S, Foglizzo M, Zhao J, Kim HG, Ekmekci C, Karimiani EG, Imannezhad S, Eghbal F, Badv RS, Schwaibold EMC, Dehghani M, Vahidi Mehrjardi MY, Metanat Z, Eslamiyeh H, Khouj E, Nasser Alhajj SM, Chedrawi A, Ramzan K, Hashmi JA, Alluqmani MM, Basit S, Veltra D, Marinakis NM, Niotakis G, Vorgia P, Sofocleous C, Lee H, Jeong WC, Umair M, Bilal M, Pinheiro Ferreira Alves CA, Sieber M, Kruer M, Houlden H, Alkuraya FS, Zeqiraj E, Greenberg RA, Cenik C, Yu L, Maroofian R, Wu J, Buszczak M. A programmed decline in ribosome levels governs human early neurodevelopment. 2025. ***Nat Cell Biol*** 27(8):1240-1255. (***shared first author**) **Impact factor: 19.100**

Vona B, Wollnik B, Strenzke N, Moser T. Catching up but Still Miles Behind – a Patient Registry for Otoferlin. 2024. ***Exp Mol Med*** 56(6):1472-1473. doi: 10.1038/s12276-024-01247-6 **Impact factor: 12.900**

Pagnamenta AT, Camps C, Giacopuzzi E, Taylor JM, Hashim M, Calpena E, Kaisaki PJ, Hashimoto A, Yu J, Sanders E, Schwessinger R, Hughes JR, Lunter G, Dreau H, Ferla M, Lange L, Kesim Y, Ragoussis V, Vavoulis DV, Allroggen H, Ansorge O, Babbs C, Banka S, Baños-Piñero B, Beeson D, Ben-Ami T, Bennett DL, Bento C, Blair E, Brasch-Andersen C, Bull KR, Cario H, Cilliers D, Conti V, Davies EG, Dhalla F, Dacal BD, Dong Y, Dunford JE, Guerrini R, Harris AL, Hartley J, Hollander G, Javaid K, Kane M, Kelly D, Kelly D,

Knight SJL, Kreins AY, Kvikstad EM, Langman CB, Lester T, Lines KE, Lord SR, Lu X, Mansour S, Manzur A, Maroofian R, Marsden B, Mason J, McGowan SJ, Mei D, Mlcochova H, Murakami Y, Németh AH, Okoli S, Ormondroyd E, Ousager LB, Palace J, Patel SY, Pentony MM, Pugh C, Rad A, Ramesh A, Riva SG, Roberts I, Roy N, Salminen O, Schilling KD, Scott C, Sen A, Smith C, Stevenson M, Thakker RV, Twigg SRF, Uhlig HH, van Wijk R, **Vona B**, Wall S, Wang J, Watkins H, Zak J, Schuh AH, Kini U, Wilkie AOM, Popitsch N, Taylor JC. Structural and non-coding variants increase the diagnostic yield of clinical whole genome sequencing for rare diseases. 2023. ***Genome Med*** 15(1):94. doi: 10.1186/s13073-023-01240-0 **Impact factor: 10.400**

Trpchevska N*, Freidin MB*, Broer L*, Oosterloo BC, Yao S, Zhou Y, **Vona B**, Bishop C, Bizaki-Vallaskangas A, Canlon B, Castellana F, Chasman DI, Cherny S, Christensen K, Concas MP, Correa A, Elkon R; Estonian Biobank Research Team, Mengel-From J, Gao Y, Giersch ABS, Girotto G, Gudjonsson A, Gudnason V, Heard-Costa NL, Hertzano R, Hjelmborg JVB, Hjerling-Leffler J, Hoffman HJ, Kaprio J, Kettunen J, Krebs K, Kähler AK, Lallemend F, Launer LJ, Lee IM, Leonard H, Li CM, Lowenheim H, Magnusson PKE, van Meurs J, Milani L, Morton CC, Mäkitie A, Nalls MA, Nardone GG, Nygaard M, Palviainen T, Pratt S, Quaranta N, Rämö J, Saarentaus E, Sardone R, Satizabal CL, Schweinfurth JM, Seshadri S, Shiroma E, Shulman E, Simonsick E, Spankovich C, Tropitzsch A, Lauschke VM, Sullivan PF, Goedegebure A, Cederroth CR**, Williams FMK**, Nagtegaal AP**. Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. 2022. ***Am J Hum Genet*** Jun 2;109(6):1077-1091. doi: 10.1016/j.ajhg.2022.04.010 (***equal contribution, **equal contribution**) **Impact factor: 9.800**

PUBLICATIONS

Pan Y, Li X, Li D, Lou S, Lin J, Gao Y, **Vona B**, Mi C, Wang L, Ma L, Du M. 2025.Genetic Regulation of ARID3B Confers Cleft Lip with/without Cleft Palate Susceptibility Through LLPS-mediated Transcriptional Program. ***Cell Rep.*** [Accepted].

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INVITED EDITORIAL WORK

Co-editing a special issue for ***Scientific Reports*** on hearing disorders and deafness. Submission deadline December 31, 2025.

Co-edited a special issue for ***Medizinische genetik***, the official journal of the German Society of Human Genetics, about the genetics of hearing loss and other auditory disorders. Publication date: August 2020.

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INVITED PRESENTATIONS

Vona B. A human genetics journey toward unravelling the genomics of auditory function. Neuroscience PhD Series. Medical University Innsbruck, Innsbruck, Austria, April 2nd, 2025.

Vona B, Wollnik B. Human genetic diagnostics for hearing loss. Deutsche Gesellschaft für Audiologie & Arbeitsgemeinschaft Deutschsprachiger Audiologen und Neuro-Otologen Annual Meeting, Göttingen, Germany, March 19th, 2025. Invited Tutorial.

Vona B. Perspectives on precision medicine for hereditary hearing loss: from gene discovery to gene therapy. International Congress of the Iranian Medical Sciences Students, Urmia, Iran, September 7th, 2024. Keynote Speaker.

Vona B. A human genetics journey toward unravelling the genomics of auditory function. Spring Seminar Series. Creighton University, Omaha, NE, USA, May 15th, 2024.

Vona B. Genetic diagnosis of hearing loss. NHS Northwest Genomic Laboratory Hub Seminar. Liverpool, England, December 7th, 2023.

Vona B. From diversity to discovery: Illuminating inner ear genetics and therapeutics through patient engagement. Inner Ear Biology Workshop, London, England, September 3rd, 2023.

Vona B. Genetic diagnosis of hearing loss. European Society of Human Genetics. Glasgow, Scotland, June 13th, 2023.

Vona B. Cochlear implantation from the genetic perspective. Deutsche Gesellschaft für Hals-Nasen-Ohren-Heilkunde, Kopf- und Hals-Chirurgie, Leipzig, Germany, May 18th, 2023.

Vona B. A genomic view of hereditary hearing loss. University College London Ear Institute Seminar Series, Virtual, March 4th, 2022.

Vona B. A genomic view of hereditary hearing loss. Hamad Bin Khalifa University Seminar Series, Virtual, February 16th, 2022.

Vona B. Advances in hearing loss diagnostics testing: a gateway to therapeutic development. Inner Ear Disorders Therapeutics Summit, Virtual, May 25th, 2021.

Vona B. Genetics of Hearing Loss: A sneak peek at two candidate genes. Radboud University Medical Center, Department of Human Genetics, Nijmegen, the Netherlands, November 11th, 2019.

Vona B. Genetic diagnosis of hearing loss. Hospital Universitario San Cicilio Conference, University of Granada, Granada, Spain, February 25th, 2019.

Hertzano R, **Vona B**. Session co-chair: Inner-Ear Genomics and Gene Regulation. Association for Research in Otolaryngology Mid-Winter Meeting, Baltimore, MD, USA, February 9-13, 2019.

Vona B. From diagnostics to novel gene discovery—a gene hunter's guide to hereditary hearing loss. Progress in Neuroscience Research of the Retina and Inner Ear, Center for Neurosensory Systems, Tübingen, Germany, January 24th, 2019.

Vona B, Nanda I, Haaf T. Advances in hearing loss gene mutation detection: the gateway to therapeutic target development. New Horizons in Vision and Hearing Research, Tübingen, Germany, March 5-7, 2018.

Vona B. Genetics of Deafness. Arbeitsgemeinschaft Deutschsprachiger Audiologen, Neurootologen und Otologen (ADANO) Herbsttagung, Tübingen, Germany, September 21-22, 2017.

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Koparir A, Bahena P, Hofrichter MAH, Zamani M, Tovornik S, Koparir E, Torbati P, Rad A, Owrang D, Kalay I, Dragicevic N, Chamanrou N, Martínez Völter LN, Christophersen N, Baranzehi T, Loum S, Shadab M, Alidadiani N, Shahrooz G, Haack T, Alavi A, Kremer H, Kordi-Tamandani D, Karimiani EG, Hebestreit H, Galehdari H, Flandin S, Sheata Dieler W, Maroofian R, Haaf T, **Vona B**. Clinical and genetic heterogeneity of syndromic hearing loss with an emphasis on syndromic genes mimicking non-syndromic hearing loss. 2025. European Society of Human Genetics, Milan, Italy, May 24-27, 2025.

Bögershausen N, **Vona B**, Cyganek L, Fedorenko A, Take P, Strohmeyer C, Hoerauf H, Wollnik B. InsightRP2: a global patient registry for RP2-associated X-linked retinitis pigmentosa. 2025. European Society of Human Genetics, Milan, Italy, May 24-27, 2025.

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Andreae H, Jahnke F, Zhang Z, Benseler F, **Vona B**. Using the pET01 exon trapping vector to unmask splicing effects of synonymous OTOF variants. 2025. Deutsche Gesellschaft für Humangenetik, Innsbruck, Austria, April 2-4, 2025.

Curcio M, Andreae H, Zhang Z, Owrang D, Benseler F, **Vona B**. Uncovering the splicing effects of Otoferlin missense variants and their potential impact on hearing loss. 2025. Deutsche Gesellschaft für Humangenetik, Innsbruck, Austria, April 2-4, 2025.

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von Hardenberg, S., Wiemers A., Phillipe C, S. Goldacker S., **Vona B.**, Auber, B. Uncovering a novel syndromic immune disorder. European Society for Immunodeficiencies. 2024. Marseille, France, October 16-19, 2024.

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Christophersen N, Zamani M, Mazaheri N, Rad A, Maroofian R, Galehdari H, **Vona B**. The discovery of the second family with Clarin-2-related autosomal recessive hearing loss. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

Jahnke F, Zhang Z, Owrang D, **Vona B**. Using the pSPL3 exon trapping vector to unmask splicing effects of otoferlin coding variants. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

Kalay I, **Vona B**. Homozygosity for a Dominant Novel NPR2 Splice-Altering Variant in a Turkish Family. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

Efthymiou S, Deng C, Leo C, Zhang K, Qing Lin R, Karagoz I, Scardamaglia A, Owrang D, Maroofian R, **Vona B**, Fry A, Houlden H, Fu D. Novel bi-allelic TRMT1 variants perturb tRNA modifications expanding the clinical spectrum of intellectual disability. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

Najarzadeh Torbati P, Karimiani EG, Zamani M, Kaiyrzhanov R, Liebmann L, **Vona B**, Huebner AK, Calame D, Misra VK, Sadeghian S, Azizimalamiri R, Mohammadi MH, Zeighami J, Heydaran S, Toosi MB, Akhondian J, Babaei M, Hashemi N, Schnur R, Suri M, Setzke J, Wagner M, Brunet T, Grochowski CM, Emrick L, Chung WK, Hellmich U, Schmidts M, Lupski J, Galehdari H, Severino M, Houlden H, Hübner C, Maroofian R. Biallelic variants in SLC4A10 lead to a neurodevelopmental disorder. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

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Tshering KC, DiStefano MT, Oza AM, Ajuyah Robertson P, Webb R, Edoh E, Broeren E, Ratliff J, Gitau V, Azaiez H, Booth KTA, Kenna M, Schimmenti LA, Smith RJ, Rehm HL, Abou Tayoun AN, Amr SS on behalf of the **ClinGen Hearing Loss Clinical Domain Working Group**. Recuration of hearing loss associated- genes demonstrate significant change in gene-disease validity over time. 2024. Curating the Clinical Genome. Baltimore, MD, USA, May 9-10, 2024.

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Taylor JC, Pagnamenta AT, Camps C, Giacopuzzi E, Taylor JM, Hashim M, Calpena E, Kaisaki PJ, Hashimoto A, Yu J, Kini U, Wilkie AOM, Popitsch N, **OxClinWGS Consortium**. 2023. American Society of Human Genetics, Washington DC, USA, November 1-5, 2023.

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Huang K, Lin SJ, Petree C, Vona B, Varshney GK. Functional analysis of wfs1-deficient zebrafish shed light on the etiology of Wolfram syndrome. 2023. Society for Developmental Biology Conference, Chicago, IL, USA, July 20-23, 2023.

Lin SJ, **Vona B**, Lau T, Maroofian R, Varshney GK. Evaluating the association of biallelic OGDHL variants with a clinically heterogeneous syndrome. 2023. European Society of Human Genetics Conference, Glasgow, Scotland, June 10-13, 2023.

Tasan H, Rasheed A, Raqeeb A, Setzke J, **Vona B**, Seo GH, Kriouile Z, Alvi JR, Cali E, Salpietro V, Sultan T, Maroofian R, Naz S, Houlden H, Ferdinandusse S, Efthymiou S. Novel biallelic variants expand the phenotypic spectrum of FAR1-related disorder. 2023. European Society of Human Genetics Conference, Glasgow, Scotland, June 10-13, 2023.

Vona B, Schwartzbaum DA, Rodriguez AA, Lewis SS, Toosi MB, Radhakrishnan P, Bozan N, Akın R, Doosti M, Manju R, Duman D, Sineni CJ, Nampoothiri S, Karimiani EG, Houlden H, Bademci G, Tekin M, Girisha KM, Maroofian R, Douzgou S. Biallelic KITLG variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. 2023. Deutsche Gesellschaft für Humangenetik, Kassel, Germany, March 15-17, 2023.

Zhang Z, Owrang D, **Vona B**. Unmasking splicing effects of Otoferlin missense variants. 2023. Deutsche Gesellschaft für Humangenetik, Kassel, Germany, March 15-17, 2023.

Owrang D, Rad A, Lin SJ, Kolb SM, Oprea G, Skrahina V, Huang K, Waheed N, Hussain M, Riaz S, Bahena P, Haaf T, Varshney GK, Schindler D, **Vona B**. Mutations affecting Glycine-X-Y repeats in the COL4A6 gene cause hearing loss. 2023. Deutsche Gesellschaft für Humangenetik, Kassel, Germany, March 15-17, 2023.

Schade-Mann T, Gamerdinger P, Dofek S, Schulte B, Schulze M, Fehr S, Biskup S, Haack TB, Stöbe P, Heyd A, Harre J, Lesinski-Schiedat A, Büchner A, Lenarz T, Warnecke A, Müller M, **Vona B**, Dahlhoff E, Löwenheim H, Holderried M, Tropitzsch A. Variability in Cochlear implantation outcomes in a large German cohort With a genetic etiology of hearing loss. 2023. Association for Research in Otolaryngology Mid-Winter Meeting. Orlando, FL, USA, February 11-15, 2023.

Koparir A, Lekszas C, Keseroglu K, Rad A, Narendran N, Hasanzadeh A, Karimiani EG, Ertugrul Ozbudak M, **Vona B**, Haaf T. A biallelic gain-of-function variant in *MSGN1* causes a new skeletal dysplasia syndrome. European Society of Human Genetics Conference, Vienna, Austria, June 11-14, 2022.

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impaired patients discloses informative mutations at lightning speed. 52nd Inner Ear Biology Workshop. Rome, Italy, September 12-15, 2015.

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Eirich K, Nanda I, **Vona B**, Haaf T, Schindler D. How and why frequent large deletions arise in the *FANCA* gene. Fanconi Anemia Symposium, Denver, CO, USA, September 27-30, 2012.

Vona B, and Billington SJ. Bacteriophage and transposon association of *erm*(X) in *Arcanobacterium pyogenes*. Arizona-Nevada Branch of the American Society for Microbiology Conference. Las Vegas, NV, USA, February 25, 2006.

PROFESSIONAL AFFILIATIONS

08/2025–present	German Society for Audiology
01/2025–present	American Society of Human Genetics
04/2023–present	Atlas of Variant Effects Alliance
11/2021–present	Consortium for Gene Diagnostics
01/2020–present	European Society of Human Genetics
07/2018–08/2021	Junior Academy, Medical Faculty, University of Tübingen
01/2018–present	Association for Research in Otolaryngology
01/2015–present	German Society of Human Genetics

SERVICE IN PROFESSIONAL SOCIETIES

03/2025–present	Communications Committee, Association for Research in Otolaryngology
01/2023–02/2023	Abstract reviewer for the European Society of Human Genetics Conference, 2023, Glasgow, Scotland

PROFESSIONAL COURSES

10/2018–02/2019	*Privatissimum* laboratory animal science course, University of Tübingen
11/2012	Basic Gene Mapping Course at the Max Delbrück Center for Molecular Medicine, Berlin, Germany

GRANTS AND AWARDS

Grants:

Running Dates	Funding organization	Amount (€)
04/2025-03/2030	German Research Foundation, VO 2138/8-1, Heisenberg Award	571,500
04/2025-12/2028	Collaborative Research Consortium, CRC 1690 "Disease Mechanisms and Functional Restoration of Sensory and Motor Systems" Co-PI of Project A03	243,775
06/2022–05/2026	German Research Foundation, VO 2138/7-1, Project Number: 469177153	451,500
12/2019–05/2021	Ministry of Science, Research and Art Baden-Württemberg Research Seed Capital (RiSC) Grant (12/2019): MWK project number D.24.01169	22,064
11/2019–04/2021	*f*ortüne intramural funding (11/2019): University of Tübingen Project number 2545-1-0	88,241
10/2018	Center for Rare Diseases Tübingen: Funding for genome sequencing of 11 families	11,000
05/2018	Travel grant: Travel award to attend the 11th Molecular Biology of Hearing and Deafness in Göttingen, Germany, May 16-19, 2018	500
Total:		**1,388,580€**

Awards:

Date	Award	Organization
09/2025	Top Reviewer for the 2nd quarter of 2025 for *Molecular Medicine*	Springer Nature
07/2024	Heisenberg Award	Deutsche Forschungsgemeinschaft (DFG)
09/2019	Top Peer Reviewer Award: Top 1% peer reviewer in the field of Molecular Biology and Genetics	Publons
25/02/2006	Awarded best Undergraduate poster presentation	Arizona-Nevada Branch of the American Society for Microbiology Conference in Las Vegas, Nevada, USA

SCIENCE COMMUNICATION AND PATIENT OUTREACH

21/06/2025: Participation in the 6th Göttingen Night of Science public outreach event

19/05/2025: Participation as a Pint of Science Göttingen Invited Speaker (a global science outreach initiative). Title of Talk: Gene & Tonic: A Spirited Look at Hereditary Hearing Loss and Deafness in Humans.

Since 10/2023: Collaboration with the SPATA Foundation, a patient led advocacy group for individuals with *SPATA5* (*AFG2A*) and *SPATA5L1* (*AFG2B*) diagnoses, on a deep phenotyping and natural history study. In preparation of these studies, I collaborated with Prof. Henry Houlden (UCL Queen Square Institute of Neurology) and Prof. Michael Buszczak (UT Southwestern) to create the SPATA Registry to be released via Coordination of Rare Diesases at Sanford (CoRDS).

09/07/2022: Participation in the 5th Göttingen Night of Science public outreach event.

Since 02/2022: Content contributor of the Twitter page @Otoferlin_Group that aims to talk about Otoferlin-associated hearing impairment in ways patients can engage.

IN THE NEWS

Date	Media outlet	Title	Summary
04/08/2025	UT Southwestern Medical Center	Ribosome level changes detected in early brain development	A study published in *Nature Cell Biology* discovered that ribosome levels decrease during neuroepithelial differentiation, a very early step in human brain development, making differentiating cells particularly vulnerable to changes in ribosome biogenesis during this time. Variants in a newly discovered gene called *AIRIM* uncovered a key role of the protein in generating ribosomes. https://www.utsouthwestern.edu/newsroom/articles/year-2025/aug-ribosome-early-brain-development.html
27/11/2024	UMG Press	University Medical Center Göttingen establishes a new center for rare hearing disorders	The Center for Rare Hearing Disorders starts within the Center for Rare Diseases in Göttingen. It aims to establish new treatment and research infrastructures for hearing disorders within the UMG and to improve care of affected patients. https://www.umg.eu/presse/news-detail/news-detail/detail/news/universitaetsmedizin-goettingen-gruendet-neues-zentrum-fuer-seltene-hoerstoerungen/
15/11/2023	UCL Brain Sciences	New study reveals molecular causes of rare neurological	A new study published in *Brain* describes a new gene responsible for key molecular defects underlying a rare brain condition in children.
18/05/2022	Erasmus MC	Discovery revives old theory of the origins of hearing loss	Highlights of the *Am J Hum Genet* publication that identified a new part of the inner ear that is involved in age-related hearing impairment based on large-scale GWAS studies
16/05/2022	King's College London	Hope for treatments against hearing loss as 10 genes	Highlights of the *Am J Hum Genet* publication that identified a new part of the inner ear that is involved in age-related hearing impairment based on large-scale GWAS studies
01/02/2021	University College London Ear Institute	*CLRN2* confirmed as new deafness gene	Press-release showcasing a new gene discovery publication: https://www.ucl.ac.uk/ear/news/2021/feb/clrn2-confirmed-new-deafness-gene

11/06/2019	Schnecke-Online	HNO-Kongress 2019: „CI-Patienten werden künftig alles über die App regeln“	Highlighted my involvement and work in the genetics of hearing loss https://www.schnecke-online.de/themen/Forschung/dghno_kongress-2019?page_69=13
23/01/2019	MDalert.com	Tinnitus linked to genetic factors	Interview about new adoption study data linking genetic factors to tinnitus

PROFESSIONAL LABORATORY EXPERIENCE

Dates	Position	Role	Employer
05/2005–06/2010	Clinical Lab Specialist IV – Nucleic Acid Testing, Red Cell Serology Labs	Donor blood testing for infectious diseases: HIV-1, HIV-2, Hepatitis C Virus, West Nile Virus nucleic acid testing; routine blood typing and antibody testing for syphilis and cytomegalovirus	Blood Systems Laboratories, Tempe, AZ, USA
06/2004–07/2005	Anatomical Tissue Grosser	Grossly describing and processing patient tissue specimens excised through biopsy for a pathology lab	LabCorp, Phoenix, AZ, USA
05/2004–10/2004	Intern – Nucleic Acid Testing Lab	Donor blood testing for West Nile Virus (nucleic acid testing)	Blood Systems Laboratories, Tempe, AZ, USA