

Priv.-Doz. Dr. Barbara Vona

Group Leader

Tel: +49 (0)551-39-65128
Email: barbara.vona@med.uni-goettingen.de

DESCRIPTION OF THE 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 impairment. 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) that ended in 2018, I was recruited to start a Junior Group at the Tübingen Hearing Research Center (University of Tübingen) that started in 2019. In Tübingen, as the only auditory human geneticist among diverse hearing research experts, I absorbed in-depth knowledge in hearing science and mouse models of deafness. I completed my Habilitation in 2019 with the thesis “Identification and characterization of genes in connection with hereditary hearing disorders”. In 2021, privileged to join the Institute for Auditory Neuroscience and Institute of Human Genetics and at the University Medical Center Göttingen, I started my group in what I envisioned to be the right combination of two complimentary disciplines. Before starting, I made a long “wish list”, shaped and motivated by my previous experiences, to establish and contribute to frameworks for exciting science while enhancing the patient journey alongside like-minded colleagues with a common vision for the future. I significantly contributed to the patient registry “Hereditary Hearing Impairment Patient Registry” in Göttingen that currently includes the Otoferlin Registry (since 2023), as well as another for *CABP2* anticipated to be online by late 2024. Furthermore, I have founded the newly approved “Center for Rare Hearing Disorders” at the Center for Rare Diseases Göttingen to improve multidisciplinary care of patients while contributing crucial structures to support research and clinical trials. I am an editorial board member for *The Journal of the Association for Research in Otolaryngology* 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*, and *JAMA Neurology*. In the last year, I have been invited to give scientific talks at the German Society of Oto-Rhino-Laryngology, Head and Neck Surgery and the European Society of Human Genetics. I have supervised 28 trainees, from bachelor to PhD and medical student level and have extensive experience in training young scientists and medical professionals.

Due to synergistic opportunities to build new frameworks in the productive Göttingen environment, I was recently able to showcase my specialization in auditory human genomics and was accepted into the Heisenberg Programme at the DFG on July 29th, 2024 that the University Medical Center Göttingen has signaled to support with a W2 tenure-track professorship. I plan to use this incredible opportunity to continue working in important areas of the hereditary hearing loss while training the next generation of scientists and improving patient care for patients in Germany and worldwide.

RESEARCH STATEMENT

My research begins when routine molecular genetic diagnostics fails to achieve a genetic diagnosis. In light of emerging gene therapies, this work is imperative to correctly annotate genes and variants involved in the basic molecular physiology of hearing and deafness and contributes to knowing the number of patients and timeframe to address genetic deficits for pre-clinical development. There are several contributing factors for a failed genetic diagnosis that my group approaches from two areas.

Firstly, my group identifies new genes for hearing loss by analyzing the sequencing data of undiagnosed families. Over the past six years, we have contributed to the discovery of 25 novel human disease-associated genes with many more at various stages of scientific validation. I have aggregated impressive patient data resources, supported by undiagnosed patients from Germany and all over the world (eg. Australia, United Kingdom) directly asking to participate in this research. This great privilege has allowed me to quickly find families that replicate or confirm a novel gene and expand on or identify new phenotypes not yet linked to a specific gene. This work has broad implications to also further improve understanding of hearing loss gene-disease relationships. When we identify a new candidate gene, I utilize my extensive international collaborative network in the auditory human genetics field to find more families to strengthen human genetics data. Beyond genetic replication in families, our functional assays apply genome editing in cell lines to uncover the molecular biology, disease mechanisms and expression of genes/proteins of interest with and without patient variants. For the most interesting candidate genes based on inner ear expression, there is opportunity to also study mouse models for human hearing loss genes, which makes the Göttingen Campus a wonderful base for this work.

Secondly, I have begun a new line of research that embraces a wave of novel *in vitro* technologies termed “Multiplex Assays of Variant Effect” (MAVEs) to tackle an issue called “variants of uncertain significance”. When genetic variants are identified in patient sequencing data, they are assigned one of three basic interpretations: benign (not causal/not diagnostic), pathogenic (potentially causal for hearing loss/diagnostic), or variants of uncertain significance (precisely neither benign nor pathogenic but cannot be completely excluded in a diagnosis). Functional studies contribute helpful data to understand the pathogenicity of genetic variants. Following recently generated promising pilot data, I plan to design a “MAVE” assay to screen all possible variants in *CLRN1* on a high-throughput basis to measure CLRN1 transport to either the plasma membrane (for wild-type like variants) or the endoplasmic reticulum (for pathogenic variants) as a way to estimate pathogenicity. Furthermore, there are small molecule chaperones (eg. BF844) that can be tested in dose-response experiments that would allow for testing of all variants and how they respond to drugs, especially those anticipated to enter the clinical trials space soon.

RESEARCH EXPERIENCE

Group Leader

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

Since 09/2021 Institute of Human Genetics, University Medical Center Göttingen, Göttingen, Germany

- **Impact:**

- Accepted into the Heisenberg Programme (DFG) with commitment from UMG to support with a W2 tenure-track professorship
- Obtained approval for a new Center for Rare Hearing Disorders at the Center for Rare Diseases Göttingen
- Designed a SPATA patient registry for individuals with *SPATA5* (*AFG2A*) and *SPATA5L1* (*AFG2B*) diagnoses, hosted at the Coordination of Rare Diseases at Sanford (CoRDS).
- Obtained ethics approval for the *CABP2* registry to be online end of 2024
- 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
- Research collaboration in the interdisciplinary “Audiogenetik Sprechstunde” for patients with hearing impairment at the University Medical Center Göttingen
- 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
- Independently ran scientific projects
- Submitted and maintained ethical approval for experiments involving humans as the study leader
- Served as a main contact for clinicians disseminating 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 the 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

- 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 (Prof. T. Haaf)
- 2008 Master of Science, Pathobiology, University of Arizona, Tucson, AZ, USA
- 2006 Bachelor of Science, Microbiology (Honors), University of Arizona, Tucson, AZ, USA

SUPERVISING AND MENTORING EXPERIENCE

<u>Dates</u>	<u>University</u>	<u>Level</u>	<u>No. of Trainees¹</u>
12/08/2024 – present	University Medical Center Göttingen	Master Lab Rotation	28
15/07/2024 – 31/08/2024	University Medical Center 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 – present	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 – present***	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 – present	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

¹Intended 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

<u>Date</u>	<u>University</u>	<u>Department</u>
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

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

PROMOTION APPOINTMENT COMMITTEE REVIEWER

- 06/2023 – Served as a committee member to review a candidate W3 (Full)- Professor
10/2023 Clinic for Child and Adolescent Psychiatry and Psychotherapy at the University Medical Center Göttingen
- 11/2022 – Served as a committee member to review a candidate to be promoted from a
03/2023 W1-Professor (assistant professor) position to a tenure track W2-Professor (associate professor) position at the University Medical Center Göttingen

TEACHING EXPERIENCE

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

MSc, Master-level; BSc, Bachelor-level

I regularly engage in research-oriented teaching by having Bachelor, Master and medical students in my lab for internships and rotations (eg. 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
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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 04/2019	Member of the ClinGen Hearing Loss Variant Curation Expert Panel
Since 04/2019	Member of the ClinGen Hearing Loss Gene Curation Expert Panel
Since 03/2018	Variant curator for the Leiden Online Variation Database 3.0 (LOVD)

DIAGNOSTIC SERVICES

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

2024 – present	Co-developed and launched the SPATA Registry for patients with a diagnosis of <i>SPATA5</i> (<i>AFG2A</i>) or <i>SPATA5L1</i> (<i>AFG2B</i>) in collaboration with The SPATA Foundation
2023 – present	Co-developed and launched the Otoferlin Registry in collaboration with members at the University Medical Center Göttingen

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 to start by the end of 2024
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, 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

2024 – present	Role: Medical and Scientific Advisory Board with The SPATA Foundation
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CONSORTIUM MEMBERSHIP

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

EDITORIAL AND PEER REVIEW SERVICE

Editorial Board Member

07/2022 – present	<i>Journal of the Association for Research in Otolaryngology</i>
04/2015 – present	<i>Molecular Syndromology</i>

Editorial Reviewer

09/2018 – present	<i>Frontiers in Neuroscience</i>
09/2018 – present	<i>Frontiers in Psychology</i>

Ad hoc Peer Reviewer

Since 09/2024	<i>Molecular Therapy</i>
Since 05/2024	<i>Journal of Medical Genetics</i>
Since 01/2024	<i>Therapeutic Advances in Rare Disease</i>
Since 06/2023	<i>Nature Communications</i>
Since 01/2023	<i>Annals of Clinical and Translational Neurology</i>
Since 11/2022	<i>Cell and Bioscience</i>

Since 07/2022	<i>European Journal of Clinical Investigation</i>
Since 06/2022	<i>British Journal of Ophthalmology</i>
Since 05/2022	<i>Annals of Neurology</i>
Since 02/2022	<i>Clinical and Translational Medicine</i>
Since 07/2021	<i>Molecular Neurobiology</i>
Since 06/2021	<i>Journal of Hearing Science</i>
Since 03/2021	<i>BMC Research Notes</i>
Since 01/2021	<i>EbioMedicine</i>
Since 11/2020	<i>Expert Review of Molecular Diagnostics</i>
Since 09/2020	<i>Molecular Genetics and Genomic Medicine</i>
Since 08/2020	<i>International Journal of Developmental Neuroscience</i>
Since 06/2020	<i>Genes</i>
Since 05/2020	<i>Journal of Clinical Laboratory Analysis</i>
Since 04/2020	<i>Frontiers in Genetics</i>
Since 03/2020	<i>JAMA Neurology</i>
Since 12/2019	<i>Frontiers in Neurology</i>
Since 12/2019	<i>Journal of Cellular and Molecular Medicine</i>
Since 10/2019	<i>Journal of Human Genetics</i>
Since 10/2019	<i>Clinical Genetics</i>
Since 09/2019	<i>Computer Methods and Programs in Biomedicine</i>
Since 06/2019	<i>European Journal of Medical Genetics</i>
Since 05/2019	<i>Cell and Tissue Research</i>
Since 04/2019	<i>Frontiers in Molecular Neuroscience</i>
Since 04/2019	<i>Naunyn-Schmiedeberg's Archives of Pharmacology</i>
Since 03/2019	<i>Frontiers in Pediatrics</i>
Since 03/2019	<i>Nucleic Acids Research</i>
Since 01/2019	<i>Acta Oto-Laryngologica</i>
Since 01/2019	<i>Cytogenetic and Genome Research</i>
Since 10/2018	<i>Human Genetics</i>
Since 10/2018	<i>Sensors</i>
Since 06/2018	<i>Journal of Molecular Medicine</i>
Since 06/2018	<i>Audiology and Neuro-otology</i>
Since 05/2018	<i>BMC Medical Genetics</i>
Since 11/2017	<i>Journal of Genetics</i>
Since 11/2017	<i>International Journal of Neonatal Screening</i>
Since 10/2017	<i>International Journal of Molecular Sciences</i>
Since 09/2017	<i>Journal of Pediatric Genetics</i>

Since 07/2017	<i>SM Otolaryngology</i>
Since 11/2016	<i>Scientific Reports</i>
Since 11/2016	<i>Orphanet Journal of Rare Diseases</i>
Since 10/2016	<i>Molecular Syndromology</i>
Since 10/2016	<i>American Journal of Medical Genetics Part A</i>
Since 09/2016	<i>Frontiers in Neuroscience</i>
Since 08/2016	<i>Hearing Research</i>
Since 05/2016	<i>Meta Gene</i>
Since 04/2016	<i>European Journal of Human Genetics</i>
Since 03/2016	<i>Genetic Testing and Molecular Biomarkers</i>
Since 01/2016	<i>Human Molecular Genetics</i>
Since 09/2015	<i>Journal of Clinical and Experimental Genetics</i>
Since 07/2015	<i>Journal of Translational Medicine</i>
Since 08/2014	<i>PLOS ONE</i>
Since 08/2013	<i>Gene</i>

AD HOC GRANT PEER REVIEWER

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

Redfield SE, De-la-Torre P, Zamani M, Wang H, Khan H, Morris T, Shariati G, Karimi M, Kenna MA, Seo GH, Xu H, Lu W, Naz S, Galehdari H, Indzhukulian AA*, Shearer AE*, **Vona B***. PKHD1L1, a gene involved in the stereocilia coat, causes autosomal recessive nonsyndromic hearing loss. 2024. ***Hum Genet*** 143(3):311-329. doi: 10.1007/s00439-024-02649-2 (***corresponding authors, equal contribution**) **Impact factor: 3.800**

Lin SJ*, **Vona B***, Lau T, Huang K, Zaki MS, Aldeen HS, Karimiani EG, Rocca C, Noureldeen MM, Saad AK, Petree C, Bartolomaeus T, Abou Jamra R, Zifarelli G, Gotkhindikar A, Wentzensen IM, Liao M, Cork EE, Varshney P, Hashemi N, Mohammadi MH, Rad A, Neira J, Toosi MB, Knopp C, Kurth I, Challman TD, Smith R, Abdalla A, Haaf T, Suri M, Joshi M, Chung WK, Moreno-De-Luca A, Houlden H, Maroofian R, Varshney GK. 2023. Evaluating the association of biallelic OGDHL variants with significant phenotypic heterogeneity. 2023. ***Genome Med*** 15:102. doi:10.1186/s13073-023-01258-4 (***shared first author**) **Impact factor: 10.400**

Lin SJ*, **Vona B*#**, Porter HM, Izadi M, Huang K, Lacassie Y, Rosenfeld JA, Khan S, Petree C, Ali TA, Muhammad N, Khan SA, Muhammad N, Liu P, Haymon ML, Rüschenhoff F, Kong IK, Schnapp L, Shur N, Chorich L, Layman L, Haaf T, Pourkarimi E, Kim HG, Varshney GK#. Biallelic variants in WARS1 cause a highly variable neurodevelopmental syndrome and implicate a critical exon for normal auditory function. 2022. ***Human***

Mutation Oct;43(10):1472-1489. doi: 10.1002/humu.24435. (*equal contribution, #shared correspondence) Editor's choice. Issue cover. **Impact factor: 4.700**

Lin SJ*, Vona B*, Barbalho PG*, Kaiyrzhanov R, Maroofian R, Petree C, Severino M, Stanley V, Varshney P, Bahena P, Alzahrani F, Alhashem A, Pagnamenta AT, Aubertin G, Estrada-Veras JI, Díaz Hernández HA, Mazaheri N, Oza A, Thies J, Renaud DL, Dugad S, McEvoy J, Sultan T, Pais LS, Tabarki B, Villalobos-Ramirez D, Rad A, Genomics England Research Consortium, Galehdari H, Ashrafzadeh F, Sahebzamani A, Saeidi K, Torti E, Elloumi HZ, Mora S, Palculict TB, Yang H, Wren JD, Fowler B, Joshi M, Behra M, Burgess SM, Nath SK, Hanna MG, Kenna M, Merritt II L, Houlden H, Karimiani EG, Alkuraya FS, Haaf T, Zaki MS, Gleeson JG, Varshney GK. Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. 2021. **Genet Med** 23(10):1933-1943. doi: 10.1038/s41436-021-01239-1 (*shared first author) **Impact factor: 8.864**

Lin YC*, Niceta M*, Muto V*, Vona B*, Pagnamenta AT, Maroofian R, Beetz C, van Duyvenvoorde H, Dentici ML, Lauffer P, Vallian S, Ciolfi A, Pizzi S, Bauer P, Grüning NM, Bellacchio E, Del Fattore A, Petrini S, Shaheen R, Tiosano D, Halloun R, Pode-Shakked B, Albayrak HM, Işık E, Wit JM, Dittrich M, Freire BL, Bertola DR, Jorge AAL, Barel O, Sabir AH, Al Tenaiji AMJ, Taji SM, Al-Sannaa N, Al-Abdulwahed H, Digilio MC, Irving M, Anikster Y, Bhavani GSL, Girisha KM; Genomics England Research Consortium, Haaf T, Taylor JC, Dallapiccola B, Alkuraya FS, Yang RB, Tartaglia M. 2021. SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. 2021. **Am J Hum Genet** 108(1):115-133. doi: 10.1016/j.ajhg.2020.11.015 (*shared first author) **Impact factor: 11.043**

PUBLICATIONS

Rawlins LE, Maroofian R, Cannon SJ, Daana M, Zamani M, Ghani S, Leslie JS, Ubeyratna N, Khan N, Khan H, Scardamaglia A, Cloarec R, Khan SA, Umair M, Sadeghian S, Galehdari H, Al-Maawali A, Al-Kindi A, Azizimalamiri R, Shariati G, Ahmed F, Al-Futaisi A, Rodriguez Cruz PM, Salazar-Villacorta A, Ndiaye M, Diop AG, Sedaghat A, Saberi A, Hamid M, Zaki MS, Vona B, Owrang D, Alhashem AM, Obeid M, Khan A, Beydoun A, Najjar M, Tajsharghi H, Zifarelli G, Bauer P, Hakami WS, Al Hashem AM, Boustany RMN, Burglen L, Alavi S, Gunning AC, Owens M, Karimiani EG, Gleeson JG, Milh M, Salah S, Khan J, Haucke V, Wright CF, McGavin L, Elpeleg O, Shabbir MI, Houlden H, Ebner M, Baple EL, Crosby AH. Elucidating the clinical and genetic spectrum of inositol polyphosphate phosphatase INPP4A-related neurodevelopmental disorder. 2024. **Genet Med** [Accepted].

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INVITED EDITORIAL WORK

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PREPRINTS

Nataliya Di Donato, **NMA Consortium**, Andrew Thom, Andreas Rump, Johannes N. Greve, Marcus Kropp, Juan Cadinanos, Salvatore Calabro, Sara Cathey, Brian Chung, Heidi Cope, Maria Costales, Sara Cuvertino, Philine Dinkel, Kalliopi Erripi, Andrew E. Fry, Livia

Garavelli, Kaomei Guan, Sabine Hoffjan, Wibke G. Janzarik, Matti Koenig, Insa Kreimer, Karolina Kuenzel, Grazia Mancini, Purificacion Marin-Reina, Andrea Meinhardt, Indra Niehaus, Daniela Pilz, Ivana Ricca, Fernando Santos Simarro, Evelin Schrock, Anja Marquardt, Manuel H. Taft, Kamer Tezcan, Sofia Thunstrom, Judith Verhagen, Alain Verloes, Bernd Wollnik, Peter Krawitz, Tzung-Chien Hsieh, Leo Zeef, Michael Seifert, Michael Heide, Catherine B. Lawrence, Neil Roberts, Dietmar Manstein, Adrian S. Woolf, Siddharth Banka. Genomic and biological panoramas of non-muscle actinopathies. 2024. **medRxiv** doi.org/10.1101/2024.08.21.24310320

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INVITED PRESENTATIONS

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 5th, 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 North West 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 Cícilo 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.

CONFERENCE PRESENTATIONS

Vona B, Strenzke N, Moser T, Wollnik B. The establishment of a patient registry for Otoferlin-related hearing loss. 2024. European Society of Human Genetics Conference, Berlin, Germany, June 1-4, 2024.

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.

Eftymiou 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.

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- Strenzke N, **Vona B**, Wollnik B, Moser T. Etablierung eines weltweit zugänglichen Registers für die auditorische Synaptopathie DFNB9 (OTOF). 2024. Deutsche Gesellschaft für Audiologie e. V., Aalen, Germany, March 6-8, 2024.
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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.
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- Engert J, Doll J, Völker J, Bieniussa L, Scholl M, **Vona B**, Hagen R, Rak K. Molekulargenetische Analyse zur Identifizierung relevanter Faktoren der Neurogenese des Nucleus cochlearis der Ratte im zeitlichen Verlauf. Deutsche Gesellschaft für Hals-Nasen-Ohren-Heilkunde, Hannover, May 25-28, 2022.
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- Weingart J, Gallant P, Doll J, **Vona B**, Rad A, Maroofian R, Haaf T, Hofrichter MAH. The autosomal recessive hearing loss gene LHFPL5 – a game changer for the Mendelian rule? 2022. Deutsche Gesellschaft für Humangenetik, Würzburg, Germany, March 16-18, 2022.
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Vona B, Tropitzsch A, Stöbe P, Gamberdinger P, Schade-Mann T, Rad A, Müller M, Sturm M, Riess O, Haack TB, Löwenheim H. Towards personalized medicine through advancing molecular genetic diagnostic approaches. Association for Research in Otolaryngology Mid-Winter Meeting. Virtual. February 20-24, 2021. Selected for the Young Investigator Symposium

Vona B, Tropitzsch A, Rad A, Schneider F, Müller M; Schade-Mann T, Biskup S, Löwenheim H. A novel splicing variant confirms COL11A1 as a cause of autosomal dominant non-syndromic hearing loss in the DFNA37 locus. European Society of Human Genetics Conference, Virtual (Berlin, Germany), June 6-9, 2020.

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- Tropitzsch A, Müller M, Dofek S, Gernerding P, Löwenheim H, **Vona B**. Reclassification of the causes of hereditary hearing impairment based on past data. 91. Jahresversammlung in der HNO-Heilkunde, Kopf- und Hals-Chirurgie. Berlin, Germany, May 20-May 23, 2020.
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Hofrichter MAH, Mojarad M, Doll J, Grimm C, Eslahi A, Hosseini NS, Rajati M, Müller T, Dittrich M, Maroofian R, Haaf T, **Vona B**. Key function of the conserved p.Arg108 residue in S1PR2 (DFNB68) and its significance in sensorineural hearing loss. Deutsche Gesellschaft für Humangenetik. Weimar, Germany, March 6-8, 2019.

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Gopal SR, **Vona B**, Maroofian R, Azaiez H, Booth KT, Clancey K, Mazaheri N, Shariati G, Sedaghat A, Stepanyan R, Smith RJH, Haaf T, Galehdari H, Alagramam KN. Mutation in the *clarin-2* gene causes hearing loss in human and a zebrafish model reveals the likely cause of that hearing loss. Association for Research in Otolaryngology Mid-Winter Meeting. Baltimore, MD, USA, February 9-13, 2019.

Bademci G, Li C, Diaz-Horta O, Abad C, **Vona B**, Maroofian R, Subasioglu A, Mihci E, Alper O, Nur BG, Benham M, Incesulu A, Silan F, Tokgoz-Yilmaz S, Salehi M, Haaf T, Cengiz FB, Blanton SH, Duman D, Walz K, Zhai RG, Tekin M. Three novel hearing loss genes reveal previously unrecognized roles of their protein products in the perception of sound. American Society of Human Genetics. San Diego, CA, USA, October 16-20, 2018.

Läßig A, **Vona B**, Bartsch O. Genetisch bedingte Tiefschwerhörigkeit im Kindesalter mit auditorischer Neuropathie. Deutscher Gesellschaft für Phoniatrie und Pädaudiologie. Innsbruck, Austria, September 20-23, 2018.

Bahena P, **Vona B**, Maroofian R, Suri F, Mirrahimi M, Daftarian N, Sabbaghi H, Behboudi H, Darvish H, Haaf T. Molecular analysis of thirty-five Iranian patients with clinical suspicion of Usher syndrome. 4th International Symposium on Usher Syndrome. Mainz, Germany, July 19-21, 2018.

Bahena P, **Vona B**, Maroofian R, Mendiratta G, Croken M, Peng S, Ye X, Rezazadeh J, Lekszas C, Haaf T, Edelmann L, Shi L. Dual diagnosis of Ellis-van Creveld syndrome and hearing loss in a consanguineous family. European Society of Human Genetics. June 16-19, 2018.

Vona B, Liedtke D, Rak K, Katana R, Schrode KM, Lauermann K, Han M, Nanda I, Senthilan PR, Jürgens L, Neuner C, Hofrichter MAH, Schnapp L, Schröder J, Zechner U, Herms S, Hoffmann P, Müller T, Dittrich M, Bartsch O, Krawitz PM, Klopocki E, Löwenheim H, Shehata-Dieler W, Wang T, Müller M, Lauer AM, Worley PF, Göpfert MC, Haaf T. Disruption of *FRMPD4* is associated with hearing loss in humans and hints to functional

conservation. 11th Molecular Biology of Hearing and Deafness. Göttingen, Germany, May 16-19, 2018.

Back D, Schraven S, **Vona B**, Hofrichter MAH, Rahne T, Hagen R, Shehata-Dieler W. Phänotyp-Charakterisierung von 9 Patienten mit DFNB16-Gendefekt. 89. Jahresversammlung der Deutschen Gesellschaft für Hals-Nasen-Ohren-Heilkunde, Kopf- und Hals-Chirurgie e. V. Lübeck, Germany, May 9-12, 2018.

Hofrichter MAH, **Vona B**, Doll J, Bahena P, Röder T, Kolb S, Nanda I, Wolf B, Müller T, Dittrich M, Maroofian R, Haaf T. Familial locus heterogeneity in non-syndromic hearing loss: evidence from whole exome sequencing in Iranian families. Deutsche Gesellschaft für Humangenetik. Münster, Germany, March 14-16, 2018.

Doll J, **Vona B**, Hofrichter MAH, Schnapp L, Wolf B, Müller T, Dittrich M, Läßig AK, Bartsch O, Haaf T. Identification of a patient with CAPOS syndrome and auditory neuropathy. Deutsche Gesellschaft für Humangenetik. Münster, Germany, March 14-16, 2018.

Bahena P, **Vona B**, Maroofian R, Haaf T. Molecular analysis of twenty-seven Iranian patients with Usher syndrome. Deutsche Gesellschaft für Humangenetik. Münster, Germany, March 14-16, 2018.

Lekszas C, Maroofian R, **Vona B**, Nanda I, Haaf T. A novel reciprocal translocation t(9;11)(p24.3;p15.4) as a cause of transgenerational Beckwith-Wiedemann syndrome. Deutsche Gesellschaft für Humangenetik. Münster, Germany, March 14-16, 2018.

Vona B, Liedtke D, Rak K, Katana R, Jürgens L, Senthilan PR, Nanda I, Neuner C, Hofrichter MAH, Schnapp L, Schröder J, Zechner U, Herms S, Hoffmann P, Müller T, Dittrich M, Bartsch O, Krawitz PM, Klopocki E, Shehata-Dieler W, Göpfert MC, Haaf T. *FRMPD4* is associated with X-linked non-syndromic hearing loss. 54th Inner Ear Biology Workshop. Hannover, Germany, September 13-16, 2017.

Lekszas C, **Vona B**, Nanda I, Maroofian R, Haaf T. Case report: a potentially new skeletal dysplasia with autosomal recessive inheritance. International Conference on Children's Bone Health. Würzburg, Germany, June 10-13, 2017.

Reubelt D, **Vona B**, Hofrichter MAH, Hagen R, Shehata-Dieler W. Hereditäre Hörstörungen in der Pädaudiologie – Genotyp, Phänotyp und Behandlungsstrategien. 88. Jahresversammlung der Deutschen Gesellschaft für Hals-Nasen-Ohren-Heilkunde, Kopf- und Hals-Chirurgie e. V. Erfurt, Germany, May 24-27, 2017.

Vona B, Liedtke D, Rak K, Katana R, Jürgens L, Senthilan PR, Nanda I, Neuner C, Hofrichter MAH, Schnapp L, Schröder J, Zechner U, Herms S, Hoffmann P, Müller T, Dittrich M, Bartsch O, Krawitz PM, Klopocki E, Shehata-Dieler W, Göpfert MC, Haaf T. *FRMPD4* is associated with X-linked non-syndromic hearing loss in humans. Deutsche Gesellschaft für Humangenetik. Bochum, Germany, March 29-31, 2017.

Hofrichter MAH, **Vona B**, Maroofian R, Schnapp L, Doll J, Röder T, Nanda I, Chioza BA, Wolf B, Shehata-Dieler W, Kunstmann E, Schröder J, Müller T, Zechner U, Bartsch O, Dittrich M, Crosby AH, Haaf T. Genotype-phenotype correlation – the many facets of heterogeneous hearing loss in the context of molecular epidemiology. Deutsche Gesellschaft für Humangenetik. Bochum, Germany, March 29-31, 2017.

Pellegrino R, Kao C, Mafra F, Garifallou J, Kaminski C, Tian L, Garcia S, **Vona B**, Haaf T, Hakonarson H. Resolving the pseudogene mapping complexities of conventional exome

sequencing in hearing impairment using 10XGenomics approach. American Society of Human Genetics. Vancouver, Canada, October 18-22, 2016.

Hofrichter MAH, **Vona B**, Maroofian R, Nanda I, Chioza BA, Shehata-Dieler W, Kunstmann E, Schröder J, Crosby AH, Haaf T. The puzzle of hereditary hearing loss and the quest for the right diagnostic strategy. 53rd Inner Ear Biology Workshop. Montpellier, France, September 17-21, 2016.

Vona B, Lechno S, Hofrichter MAH, Hopf S, Läßig AK, Haaf T, Keilmann A, Zechner U, Bartsch O. Clinical and genetic evidence implying *PDZD7* as a non-syndromic hearing loss gene. Deutsche Gesellschaft für Humangenetik. Lübeck, Germany, March 16-18, 2016.

Hofrichter MAH, **Vona B**, Maroofian R, Nanda I, Chioza BA, Shehata-Dieler W, Kunstmann E, Schröder J, Crosby AH, Haaf T. The ambiguity of syndromic and non-syndromic hearing loss. Deutsche Gesellschaft für Humangenetik. Lübeck, Germany, March 16-18, 2016.

Vona B, Hofrichter MAH, Neuner C, Schröder J, Bartsch O, Zechner U, Shehata-Dieler W, Nanda I, Haaf T. The express lane from lab to clinic: high throughput sequencing in hearing impaired patients discloses informative mutations at lightning speed. 52nd Inner Ear Biology Workshop. Rome, Italy, September 12-15, 2015.

Hofrichter MAH, **Vona B**, Kunstmann E, Nanda I, Haaf T. A haystack full of needles: the complicated interpretation of rare genetic variants in a German family with autosomal dominant non-syndromic hearing loss. 52nd Inner Ear Biology Workshop. Rome, Italy, September 12-15, 2015.

Hofrichter MAH, **Vona B**, Gräf J, Schröder J, Shehata-Dieler W, Nanda I, Haaf T. A novel *de novo* mutation in the gene *CEACAM16* in a German family with hearing impairment. Deutsche Gesellschaft für Humangenetik. Graz, Austria, April 15-17, 2015.

Vona B, Hofrichter MAH, Neuner C, Schröder J, Müller T, Shehata-Dieler W, Klopocki E, Nanda I, Haaf T. A comprehensive approach for identifying mutations involved in non-syndromic hearing loss. Deutsche Gesellschaft für Humangenetik. Essen, Germany. March 19-21, 2014.

Hofrichter MAH, **Vona B**, Neuner C, Schröder J, Gehrig A, Kraus F, Shehata-Dieler W, Klopocki E, Nanda I, Haaf T. Excessive mutational fallout of *STRC* in patients with high frequency hearing loss. Deutsche Gesellschaft für Humangenetik. Essen, Germany. March 19-21, 2014.

Vona B, Neuner C, Nanda I, Müller T, Haaf T. A novel 3' cryptic splice site mutation in *GRHL2* causes post-lingual progressive autosomal dominant hearing loss. Deutsche Gesellschaft für Humangenetik. Dresden, Germany. March 20-22, 2013.

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

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

01/2023–02/2023	Abstract reviewer for the European Society of Human Genetics Conference, 2023, Glasgow, Scotland
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PROFESSIONAL COURSES

10/2018–02/2019	<i>Privatissimum</i> 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 (€)
Starting 2025	German Research Foundation, VO 2138/8-1, Heisenberg Award	532,500
06/2022–05/2025	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	fortune 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 11 th Molecular Biology of Hearing and Deafness in Göttingen, Germany, May 16-19, 2018	500
Total:		1,105,805€

Awards:

Date	Award	Organization
07/2024	Heisenberg Award	German Research Foundation (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
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SCIENCE COMMUNICATION AND PATIENT OUTREACH

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 Diseases at Sanford (CoRDS).

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
15/11/2023	UCL Brain Sciences	New study reveals molecular causes of rare neurological condition in children	A new study published in <i>Brain</i> 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 <i>Am J Hum Genet</i> 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 identified	Highlights of the <i>Am J Hum Genet</i> 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	<i>CLRN2</i> 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 Grosser	Tissue	Grossly describing and processing patient tissue specimens excised through biopsy for a pathology lab	LabCorp, Phoenix, AZ, USA
05/2004– 10/2004	Intern – Testing Lab	Nucleic Acid	Donor blood testing for West Nile Virus (nucleic acid testing)	Blood Systems Laboratories, Tempe, AZ, USA