

Prof. Dr. rer. nat. Barbara Vona

GENERAL INFORMATION

Address: Institute for Auditory Neuroscience
University Medical Center Göttingen
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Current position: Group leader of the Hearing Genomics Group at the Institute for Auditory Neuroscience & InnerEarLab

ACADEMIC EDUCATION

2011-2014 PhD Human Genetics, University of Würzburg

2006-2008 Master of Science, Pathobiology, University of Arizona (Tucson, AZ, USA)

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

SCIENTIFIC DEGREES

2022 Umhabilitation, Human Genetics, University Medical Center Göttingen

2019 Habilitation, Human Genetics, University of Würzburg (Prof. T. Haaf)

2014 PhD (Dr. rer. nat.), Human Genetics, University of Würzburg (Prof. T. Haaf)

2008 Master of Science in Pathobiology, University of Arizona (Tucson, AZ, USA)

2006 Bachelor of Science in Microbiology, University of Arizona (Tucson, AZ, USA)

PROFESSIONAL CAREER AFTER COMPLETING DEGREE

Since 2026 Professor of Genetics of Hearing, University Medical Center Göttingen

Since 2025 Visiting scientist, Harvard Medical School, Boston, MA, USA

Since 2025 Visiting scientist, The Broad Institute, MIT & Harvard, Cambridge, MA, USA

Since 2024 Accepted into the Heisenberg Program (German Research Foundation)

Since 2022 Group Leader

2021-2022 Fellow, Institute for Auditory Neuroscience & InnerEarLab, University Medical Center Göttingen

2018-2021 Junior Group Leader, Tübingen Hearing Research Center, University of Tübingen

2014-2018 Postdoctoral fellow, Institute of Human Genetics, University of Würzburg

MISCELLANEOUS Further Scientific Activities (*selected*)

Since 2026 International Rare Diseases Research Consortium (IRDIRC) Task Force on Optimizing the Use of Data Sources and Registries

Since 2026 Nominated by the German Society of Human Genetics as an expert for amending the pediatrics directive for newborn hearing screening in Germany

Since 2025 Nominated by the German Society of Human Genetics to amend the German S2k Guidelines for Cochlear Implant Treatment

Since 2025 Coordinator of the ClinGen Splicing Working Group

Since 2025 Member of the ClinGen/AVE (Atlas of Variant Effects) Functional Data Working Group

Since 2025 Scientific Advisory Board for the Cure KARS – Laia Foundation

Since 2025 Associate Editor, Molecular Syndromology

Since 2024 Establishment of the Center for Rare Hearing Disorders within the Centers for Rare Diseases framework at UMG

Since 2024 Medical and Scientific Advisory Board of The SPATA Foundation

Since 2023	Developer and study manager for the “Hereditary Hearing Impairment Patient Registry” for patients with Otoferlin (<i>OTOF</i>) and <i>CABP2</i> associated hearing impairment. ClinicalTrials.gov IDs: NCT05946057, NCT06680934
Since 2023	Member of the Atlas of Variant Effects Alliance
Since 2022	Associate Editor, Journal of the Association for Research in Otolaryngology
Since 2022	Genomics England PanelApp Gene Reviewer for Auditory Neuropathy
Since 2022	Genomics England PanelApp Gene Reviewer for Hearing Loss
Since 2021	Member of the Consortium for Gene Diagnostics
Since 2019	Member of the ClinGen Hearing Loss Variant Curation Expert Panel
Since 2019	Member of the ClinGen Hearing Loss Gene Curation Expert Panel

SELECTED PUBLICATIONS (*with scientific assurance*)

- 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 Alhaji 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. (2025) A programmed decline in ribosome levels governs human early neurodevelopment. *Nat Cell Biol* 27(8):1240-1255
- Vona B**[#] (2025) Rethinking non-syndromic hearing loss and its mimics in the genomic era. *Eur J Hum Genet* Eur J Hum Genet 33(2):147-150.
- Vona B**, Wollnik B, Strenzke N, Moser T (2024) Catching up but still miles behind—a patient registry for otoferlin. *Exp Mol Med* 56(6):1472-1473.
- 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**^{**#} (2024) PKHD1L1, a gene involved in the stereocilia coat, causes autosomal recessive nonsyndromic hearing loss. *Hum Genet* 143(3):311-329.
- 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. *Genome Med* 15(1):102.
- Vona B***, Schwartzbaum DA^{**}, Rodriguez AA^{**}, Lewis SS, Toosi MB, Radhakrishnan P, Bozan N, Akin R, Doosti M, Manju R, Duman D, Sineni CJ, Nampoothiri S, Karimiani EG, Houlden H, Bademci G, Tekin M*, Girisha KM*, Maroofian R*, Douzougou S* (2022) Biallelic KITLG variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. *J Eur Acad Dermatol Venereol* 36(9):1606-1611.
- 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, Hernández HAD, 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, Ben Fowler, Joshi M, Behra M, Burgess SM, Nath SK, Hanna MG, Kenna M, Merritt JL 2nd, Houlden H, Karimiani EG, Zaki MS, Haaf T, Alkuraya FS, Gleeson JG, Varshney GK (2021) Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. *Genet Med* 23(10):1933-1943.
- Vona B**[#], Mazaheri N, Lin SJ, Dunbar LA, Maroofian R, Azaiez H, Booth KT, Vitry S, Rad A, Rüschemdorf F, Varshney P, Fowler B, Beetz C, Alagramam KN, Murphy D, Shariati G,

- Sedaghat A, Houlden H, Petree C, VijayKumar S, Smith RJH, Haaf T, El-Amraoui A*, Bowl MR*#, Varshney GK*, Galehdari H* (2021) A biallelic variant in CLRN2 causes nonsyndromic hearing loss in humans. *Hum Genet* 140(6):915-931.
9. 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. *Am J Hum Genet* 108(1):115-133.
 10. Rad A, Schade-Mann T, Gamerdinger P, Yanus GA, Schulte B, Müller M, Imyanitov EN, Biskup S, Löwenheim H, Tropitzsch A, **Vona B#** (2021) Aberrant COL11A1 splicing causes prelingual autosomal dominant nonsyndromic hearing loss in the DFNA37 locus. *Hum Mutat* 42(1):25-30.

* equal contribution, ** equal contribution, # corresponding author