

Priv.-Doz. Dr. rer. nat. Barbara Vona

GENERAL INFORMATION

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

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 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 2023 Developer and study manager of the Otoferlin Registry (ClinicalTrials.gov Identifier: NCT05946057)
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 2019 Member of the ClinGen Hearing Loss Variant Curation Expert Panel
Since 2019 Member of the ClinGen Hearing Loss Gene Curation Expert Panel
Since 2018 Variant curator for the Leiden Online Variation Database 3.0 (LOVD)
Since 2015 Editorial Board, Molecular Syndromology

SELECTED PUBLICATIONS (*with scientific assurance*)

1. 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, Indzhykulian 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.
2. **Vona B**# (2024) Rethinking non-syndromic hearing loss and its mimics in the genomic era. *Eur J Hum Genet* [Online ahead of print].
3. 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.
4. Pagnamenta AT, Camps C, Giacomuzzi 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 (2023) Structural and non-coding variants increase the diagnostic yield of clinical whole genome sequencing for rare diseases. *Genome Med* 15(1):94.
5. **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*, Douzgou 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.
6. 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, Giroto G, Gudjonsson A, Gudnason V, Heard-Costa NL, Hertzano R, Hjelmberg JVB, Hjerling-Leffler J, Hoffman HJ, Kaprio J, Kettunen J, Krebs K, Kähler AK, Lallemand 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** (2022) Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. *Am J Hum Genet* 109(6):1077-1091.
7. 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.

8. **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 non-syndromic 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