

List of Publications

i. Original articles (published or accepted/in press)

1. **Vona B**, Nanda I, Neuner C, Müller T, Haaf T. 2013. Confirmation of GRHL2 as the gene for the DFNA28 locus. *Am J Med Genet A* **161A**:2060–2065. doi:10.1002/ajmg.a.36017
2. **Vona B**, Neuner C, El Hajj N, Schneider E, Farcas R, Beyer V, Zechner U, Keilmann A, Poot M, Bartsch O, Nanda I, Haaf T. 2014. Disruption of the ATE1 and SLC12A1 Genes by Balanced Translocation in a Boy with Non-Syndromic Hearing Loss. *Mol Syndromol* **5**:3–10. doi:10.1159/000355443
3. **Vona B**, Müller T, Nanda I, Neuner C, Hofrichter MAH, Schröder J, Bartsch O, Läßig A, Keilmann A, Schraven S, Kraus F, Shehata-Dieler W, Haaf T. 2014. Targeted next-generation sequencing of deafness genes in hearing-impaired individuals uncovers informative mutations. *Genet Med* **16**:945–953. doi:10.1038/gim.2014.65
4. **Vona B**, Nanda I, Neuner C, Schröder J, Kalscheuer VM, Shehata-Dieler W, Haaf T. 2014. Terminal chromosome 4q deletion syndrome in an infant with hearing impairment and moderate syndromic features: review of literature. *BMC Med Genet* **15**:72. doi:10.1186/1471-2350-15-72
5. **Vona B**, Hofrichter M a. H, Neuner C, Schröder J, Gehrig A, Hennermann JB, Kraus F, Shehata-Dieler W, Klopocki E, Nanda I, Haaf T. 2015. DFNB16 is a frequent cause of congenital hearing impairment: implementation of STRC mutation analysis in routine diagnostics. *Clin Genet* **87**:49–55. doi:10.1111/cge.12332
6. Hofrichter MAH, Nanda I, Gräf J, Schröder J, Shehata-Dieler W, **Vona B[#]**, Haaf T. 2015. A Novel de novo Mutation in CEACAM16 Associated with Postlingual Hearing Impairment. *Mol Syndromol* **6**:156–163. doi:10.1159/000439576 **#Corresponding author**
7. **Vona B[#]**, Lechno S, Hofrichter MAH, Hopf S, Lägig AK, Haaf T, Keilmann A, Zechner U, Bartsch O[#]. 2016. Confirmation of PDZD7 as a Nonsyndromic Hearing Loss Gene. *Ear Hear* **37**:e238-246. doi:10.1097/AUD.0000000000000278 **#Shared corresponding authors**
8. **Vona B**, Maroofian R, Mendiratta G, Croken M, Peng S, Ye X, Rezazadeh J, Bahena P, Lekszas C, Haaf T, Edelmann L, Shi L. 2017. Dual Diagnosis of Ellis-van Creveld Syndrome and Hearing Loss in a Consanguineous Family. *Mol Syndromol* **9**:5–14. doi:10.1159/000480458
9. Tranebjærg L, Strenzke N, Lindholm S, Rendtorff ND, Poulsen H, Khandelia H, Kopec W, Lyngbye TJB, Hamel C, Delettre C, Bocquet B, Bille M, Owen HH, Bek T, Jensen H, Østergaard K, Möller C, Luxon L, Carr L, Wilson L, Rajput K, Sirimanna T, Harrop-Griffiths K, Rahman S, **Vona B**, Doll J, Haaf T, Bartsch O, Rosewich H, Moser T, Bitner-Glindzicz M. 2018. The CAPOS mutation in ATP1A3 alters Na/K-ATPase function and results in auditory neuropathy which has implications for management. *Hum Genet* **137**:111–127. doi:10.1007/s00439-017-1862-z
10. Hofrichter MAH, Mojarad M, Doll J, Grimm C, Eslahi A, Hosseini NS, Rajati M, Müller T, Dittrich M, Maroofian R, Haaf T, **Vona B[#]**. 2018. The conserved p.Arg108 residue in S1PR2 (DFNB68) is fundamental for proper hearing: evidence from a consanguineous Iranian family. *BMC Med Genet* **19**:81. doi:10.1186/s12881-018-0598-5 **#Corresponding author**
11. **Vona B[#]**, Hofrichter MAH, Schröder J, Shehata-Dieler W, Nanda I, Haaf T. 2018. Hereditary hearing loss SNP-microarray pilot study. *BMC Res Notes* **11**:391. doi:10.1186/s13104-018-3466-7 **#Corresponding author**

12. Bademci G, Abad C, Incesulu A, Rad A, Alper O, Kolb SM, Cengiz FB, Diaz-Horta O, Silan F, Mihci E, Ocaik E, Najafi M, Maroofian R, Yilmaz E, Nur BG, Duman D, Guo S, Sant DW, Wang G, Monje PV, Haaf T, Blanton SH, Vona B, Walz K, Tekin M. 2018. MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss. *Hum Genet* **137**:479–486. doi:10.1007/s00439-018-1901-4
13. **Vona B[#]**, Maroofian R, Bellacchio E, Najafi M, Thompson K, Alahmad A, He L, Ahangari N, Rad A, Shahrokhzadeh S, Bahena P, Mittag F, Traub F, Movaffagh J, Amiri N, Doosti M, Boostani R, Shirzadeh E, Haaf T, Diodato D, Schmidts M, Taylor RW, Karimiani EG[#]. 2018. Expanding the clinical phenotype of IARS2-related mitochondrial disease. *BMC Med Genet* **19**:196. doi:10.1186/s12881-018-0709-3
#Shared corresponding authors
14. Szczepek AJ, Frejo L, **Vona B**, Trpchevska N, Cederroth CR, Caria H, Lopez-Escamez JA. 2019. Recommendations on Collecting and Storing Samples for Genetic Studies in Hearing and Tinnitus Research. *Ear Hear* **40**:219–226. doi:10.1097/AUD.0000000000000614
15. Back D, Shehata-Dieler W, **Vona B**, Hofrichter MAH, Schroeder J, Haaf T, Rahne T, Hagen R, Schraven SP. 2019. Phenotypic Characterization of DFNB16-associated Hearing Loss. *Otol Neurotol* **40**:e48–e55. doi:10.1097/MAO.0000000000002059
16. **Vona B[#]**. 2019. Heritability and Tinnitus. *JAMA Otolaryngol Head Neck Surg* **145**:229–230. doi:10.1001/jamaoto.2018.3946 **#Corresponding author**
17. Hedberg-Oldfors C, Abramsson A, Osborn DPS, Danielsson O, Fazlinezhad A, Nilipour Y, Hübbert L, Nennesmo I, Visuttijai K, Bharj J, Petropoulou E, Shoreim A, **Vona B**, Ahangari N, López MD, Doosti M, Banote RK, Maroofian R, Edling M, Taherpour M, Zetterberg H, Karimiani EG, Oldfors A, Jamshidi Y. 2019. Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. *Hum Mol Genet* **28**:1919–1929. doi:10.1093/hmg/ddz032
18. Lekszas C, Nanda I, **Vona B**, Böck J, Ashrafzadeh F, Donyadideh N, Ebrahimzadeh F, Ahangari N, Maroofian R, Karimiani EG, Haaf T. 2019. Unbalanced segregation of a paternal t(9;11)(p24.3;p15.4) translocation causing familial Beckwith-Wiedemann syndrome: a case report. *BMC Med Genomics* **12**:83. doi:10.1186/s12920-019-0539-y
19. Hofrichter MAH, Doll J, Habibi H, Enayati S, Vahidi Mehrjardi MY, Müller T, Dittrich M, Haaf T, **Vona B[#]**. 2019. Exome-wide copy number variation analysis identifies a COL9A1 in frame deletion that is associated with hearing loss. *Eur J Med Genet* **62**:103724. doi:10.1016/j.ejmg.2019.103724 **#Corresponding author**
20. Lekszas C, Foresti O, Raote I, Liedtke D, König E-M, Nanda I, **Vona B**, De Coster P, Cauwels R, Malhotra V, Haaf T. 2020. Biallelic TANGO1 mutations cause a novel syndromal disease due to hampered cellular collagen secretion. *Elife* **9**:e51319. doi:10.7554/eLife.51319
21. Doll J, Hofrichter MAH, Bahena P, Heihoff A, Segebarth D, Müller T, Dittrich M, Haaf T, **Vona B[#]**. 2020. A novel missense variant in MYO3A is associated with autosomal dominant high-frequency hearing loss in a German family. *Mol Genet Genomic Med* **8**:e1343. doi:10.1002/mgg3.1343 **#Corresponding author**
22. Sabbaghi H, Daftarian N, Suri F, Mirrahimi M, Madani S, Sheikhtaheri A, Khorrami F, Saviz P, Zarei Nejad M, Tivay A, Shahriari HA, Maleki A, Ahmadi SS, Sargazi M, Cremers FPM, Najafi M, **Vona B**, Haaf T, Bahena-Carbajal P, Moghadasi A, Naraghi H, Yaseri M, Kheiri B, Kalantarion M, Sabbaghi E, Salami M, Pazooki L, Zendedel K, Mojarrab S, Ahmadi H. 2020. The First Inherited Retinal Disease Registry in Iran: Research Protocol and Results of a Pilot Study. *Arch Iran Med* **23**:445–454. doi:10.34172/aim.2020.41

23. Rad A, Schade-Mann T, Gamberdinger 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**:25–30. doi:10.1002/humu.24136 **#Corresponding author**
24. Doll J, **Vona B[#]**, Schnapp L, Rüschenhoff F, Khan I, Khan S, Muhammad N, Alam Khan S, Nawaz H, Khan A, Ahmad N, Kolb SM, Kühlewein L, Labonne JDJ, Layman LC, Hofrichter MAH, Röder T, Dittrich M, Müller T, Graves TD, Kong I-K, Nanda I, Kim H-G[#], Haaf T. 2020. Genetic Spectrum of Syndromic and Non-Syndromic Hearing Loss in Pakistani Families. *Genes (Basel)* **11**:1329. doi:10.3390/genes11111329 **#Shared corresponding authors**
25. 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 N-M, 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 R-B, 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**:115–133. doi:10.1016/j.ajhg.2020.11.015 ***Shared first author**
26. Prasad S, **Vona B**, Diñeiro M, Costales M, González-Aguado R, Fontalba A, Diego-Pérez C, Subasioglu A, Bademci G, Tekin M, Cabanillas R, Cadiñanos J, Fridberger A. 2020. Radixin modulates the function of outer hair cell stereocilia. *Commun Biol* **3**:792. doi:10.1038/s42003-020-01506-y
27. Sokolova TN, Breder VV, Shumskaya IS, Suspitsin EN, Aleksakhina SN, Yanus GA, Tiurin VI, Ivantsov AO, **Vona B**, Raskin GA, Gamajunov SV, Imyanitov EN. 2021. Revisiting multiple erroneous genetic testing results and clinical misinterpretations in a patient with Li-Fraumeni syndrome: lessons for translational medicine. *Hered Cancer Clin Pract* **19**:2. doi:10.1186/s13053-020-00157-8
28. Efthymiou S, Dutra-Clarke M, Maroofian R, Kaiyrzhanov R, Scala M, Reza Alvi J, Sultan T, Christoforou M, Tuyet Mai Nguyen T, Mankad K, **Vona B**, Rad A, Striano P, Salpietro V, Guillen Sacoto MJ, Zaki MS, Gleeson JG, Campeau PM, Russell BE, Houlden H. 2021. Expanding the phenotype of PIGS-associated early onset epileptic developmental encephalopathy. *Epilepsia* **62**:e35–e41. doi:10.1111/epi.16801
29. **Vona B[#]**, Mazaheri N, Lin S-J, Dunbar LA, Maroofian R, Azaiez H, Booth KT, Vitry S, Rad A, Rüschenhoff 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**:915–931. doi:10.1007/s00439-020-02254-z **#Shared corresponding authors**
30. Ziegler A, Duclaux-Loras R, Revenu C, Charbit-Henrion F, Begue B, Duroure K, Grimaud L, Guihot AL, Desquiere-Dumas V, Zarhrate M, Cagnard N, Mas E, Breton A, Edouard T, Billon C, Frank M, Colin E, Lenaers G, Henrion D, Lyonnet S, Faivre L, Alembik Y, Philippe A, Moulin B, Reinstein E, Tzur S, Attali R, McGillivray G, White SM, Gallacher L, Kutsche K, Schneeberger P, Girisha KM, Nayak SS, Pais L, Maroofian R, Rad A, **Vona B**, Karimiani EG, Lekszas C, Haaf T, Martin L, Ruemmele F, Bonneau D, Cerf-Bensussan N, Del Bene F, Parlato M. 2021. Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. *Am J Hum Genet* **108**:1126–1137. doi:10.1016/j.ajhg.2021.04.020

31. Rad A, Najafi M, Suri F, Abedini S, Loum S, Karimiani EG, Daftarian N, Murphy D, Doosti M, Moghaddasi A, Ahmadi H, Sabbaghi H, Rajati M, Hashemi N, **Vona B**, Schmidts M. 2022. Identification of three novel homozygous variants in COL9A3 causing autosomal recessive Stickler syndrome. *Orphanet J Rare Dis* **17**:97. doi:10.1186/s13023-022-02244-6
32. Bahena P, Daftarian N, Maroofian R, Linares P, Villalobos D, Mirrahimi M, Rad A, Doll J, Hofrichter MAH, Koparir A, Röder T, Han S, Sabbaghi H, Ahmadi H, Behboudi H, Villanueva-Mendoza C, Cortés-Gonzalez V, Zamora-Ortiz R, Kohl S, Kuehlewein L, Darvish H, Alehabib E, Arenas-Sordo M de la L, Suri F^{*,#}, **Vona B^{*,#}**, Haaf T^{*,#}. 2022. Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. *Hum Genet* **141**:785–803. doi:10.1007/s00439-021-02303-1
****Shared last author, #Shared corresponding authors**
33. 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 null, Joshi M, Behra M, Burgess SM, Nath SK, Hanna MG, Kenna M, Merritt JL, 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**:1933–1943. doi:10.1038/s41436-021-01239-1 ***Shared first author**
34. Iqbal M, Maroofian R, Çavdarlı B, Riccardi F, Field M, Banka S, Bubshait DK, Li Y, Hertecant J, Baig SM, Dymont D, Efthymiou S, Abdullah U, Makhdoom EUH, Ali Z, Scherf de Almeida T, Molinari F, Mignon-Ravix C, Chabrol B, Antony J, Ades L, Pagnamenta AT, Jackson A, Douzgou S, Genomics England Research Consortium, Beetz C, Karageorgou V, **Vona B**, Rad A, Baig JM, Sultan T, Alvi JR, Maqbool S, Rahman F, Toosi MB, Ashrafzadeh F, Imannezhad S, Karimiani EG, Sarwar Y, Khan S, Jameel M, Noegel AA, Budde B, Altmüller J, Motameny S, Höhne W, Houlden H, Nürnberg P, Wollnik B, Villard L, Alkuraya FS, Osmond M, Hussain MS, Yigit G. 2021. Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. *Genet Med* **23**:2138–2149. doi:10.1038/s41436-021-01260-4
35. Richard EM, Bakhtiari S, Marsh APL, Kaiyrzhanov R, Wagner M, Shetty S, Pagnozzi A, Nordlie SM, Guida BS, Cornejo P, Magee H, Liu J, Norton BY, Webster RI, Worgan L, Hakonarson H, Li J, Guo Y, Jain M, Blesson A, Rodan LH, Abbott M-A, Comi A, Cohen JS, Alhaddad B, Meitinger T, Lenz D, Ziegler A, Kotzaeridou U, Brunet T, Chassevent A, Smith-Hicks C, Ekstein J, Weiden T, Hahn A, Zharkinbekova N, Turnpenny P, Tucci A, Yelton M, Horvath R, Gungor S, Hiz S, Oktay Y, Lochmuller H, Zollino M, Morleo M, Marangi G, Nigro V, Torella A, Pinelli M, Amenta S, Husain RA, Grossmann B, Rapp M, Steen C, Marquardt I, Grimm M, Grasshoff U, Korenke GC, Owczarek-Lipska M, Neidhardt J, Radio FC, Mancini C, Claps Sepulveda DJ, McWalter K, Begtrup A, Crunk A, Guillen Sacoto MJ, Person R, Schnur RE, Mancardi MM, Kreuder F, Striano P, Zara F, Chung WK, Marks WA, van Eyk CL, Webber DL, Corbett MA, Harper K, Berry JG, MacLennan AH, Gecz J, Tartaglia M, Salpietro V, Christodoulou J, Kaslin J, Padilla-Lopez S, Bilguvar K, Munchau A, Ahmed ZM, Hufnagel RB, Fahey MC, Maroofian R, Houlden H, Sticht H, Mane SM, Rad A, **Vona B**, Jin SC, Haack TB, Makowski C, Hirsch Y, Riazuddin S, Kruer MC. 2021. Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. *Am J Hum Genet* **108**:2006–2016. doi:10.1016/j.ajhg.2021.08.003

36. Kaiyrzhanov R, Zaki MS, Maroofian R, Dominik N, Rad A, **Vona B**, Houlden H. 2021. A Novel Homozygous ADCY5 Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. *Mov Disord Clin Pract* **8**:1140–1143. doi:10.1002/mdc3.13310
37. Yap ZY, Efthymiou S, Seiffert S, Vargas Parra K, Lee S, Nasca A, Maroofian R, Schrauwen I, Pendziwiat M, Jung S, Bhoj E, Striano P, Mankad K, **Vona B**, Cuddapah S, Wagner A, Alvi JR, Davoudi-Dehaghani E, Fallah M-S, Gannavarapu S, Lamperti C, Legati A, Murtaza BN, Nadeem MS, Rehman MU, Saeidi K, Salpietro V, von Spiczak S, Sandoval A, Zeinali S, Zeviani M, Reich A, SYNaPS Study Group, University of Washington Center for Mendelian Genomics, Jang C, Helbig I, Barakat TS, Ghezzi D, Leal SM, Weber Y, Houlden H, Yoon WH. 2021. Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. *Am J Hum Genet* **108**:2368–2384. doi:10.1016/j.ajhg.2021.11.003
38. Tropitzsch A, Schade-Mann T, Gamerding P, Dofek S, Schulte B, Schulze M, Battke F, Fehr S, Biskup S, Heyd A, Müller M, Löwenheim H, **Vona B**[#], Holderried M. 2022. Diagnostic Yield of Targeted Hearing Loss Gene Panel Sequencing in a Large German Cohort With a Balanced Age Distribution from a Single Diagnostic Center: An Eight-year Study. *Ear Hear* **43**:1049–1066. doi:10.1097/AUD.0000000000001159
#Corresponding author
39. Pater JA, Penney C, O’Rielly DD, Griffin A, Kamal L, Brownstein Z, **Vona B**, Vinkler C, Shohat M, Barel O, French CR, Singh S, Werdyani S, Burt T, Abdelfatah N, Houston J, Doucette LP, Squires J, Glaser F, Roslin NM, Vincent D, Marquis P, Woodland G, Benoukraf T, Hawkey-Noble A, Avraham KB, Stanton SG, Young T-L. 2022. Autosomal dominant non-syndromic hearing loss maps to DFNA33 (13q34) and co-segregates with splice and frameshift variants in ATP11A, a phospholipid flippase gene. *Hum Genet*. doi:10.1007/s00439-022-02444-x
40. van der Knoop MM, Maroofian R, Fukata Y, van Ierland Y, Karimiani EG, Lehesjoki AE, Muona M, Paetau A, Miyazaki Y, Hirano Y, Selim L, de França M, Fock RA, Beetz C, Ruivenkamp CAL, Eaton AJ, Morneau-Jacob FD, Sagi-Dain L, Shemer-Meir L, Peleg A, Haddad-Halloun J, Kamphuis DJ, Peeters-Scholte CMPCD, Kurul SH, Horvath R, Lochmüller H, Murphy D, Waldmüller S, Spranger S, Overberg D, Muir AM, Rad A, **Vona B**, Abdulwahad F, Maddirevula S, Povolotskaya IS, Voinova VY, Gowda VK, Srinivasan VM, Alkuraya FS, Mefford HC, Alfarhel M, Haack TB, Striano P, Severino M, Fukata M, Hilhorst-Hofstee Y, Houlden H. 2022. Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. *Brain* **145**:2301–2312. doi:10.1093/brain/awac116
41. **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^{*}. 2022. Biallelic KITLG variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. *J Eur Acad Dermatol Venereol* **36**:1606–1611. doi:10.1111/jdv.18207 ***Equal contribution**
42. 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, Hjelmberg JVB, Hjerling-Leffler J, Hoffman HJ, Kaprio J, Kettunen J, Krebs K, Kähler AK, Lallemand F, Launer LJ, Lee I-M, Leonard H, Li C-M, Löwenheim 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, Goedegebuere 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**:1077–1091. doi:10.1016/j.ajhg.2022.04.010, **Top five most important paper**

43. Tábara LC, Al-Salmi F, Maroofian R, Al-Futaisi AM, Al-Murshedi F, Kennedy J, Day JO, Courtin T, Al-Khayat A, Galedari H, Mazaheri N, Protasoni M, Johnson M, Leslie JS, Salter CG, Rawlins LE, Fasham J, Al-Maawali A, Voutsina N, Charles P, Harrold L, Keren B, Kunji ERS, **Vona B**, Jelodar G, Sedaghat A, Shariati G, Houlden H, Crosby AH, Prudent J, Baple EL. 2022. TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. *Brain* **145**:3095–3107. doi:10.1093/brain/awac123
44. Bögershausen N, Krawczyk HE, Jamra RA, Lin S-J, Yigit G, Hüning I, Polo AM, **Vona B**, Huang K, Schmidt J, Altmüller J, Luppe J, Platzer K, Dörgeloh BB, Busche A, Biskup S, Mendes MI, Smith DEC, Salomons GS, Zibat A, Bültmann E, Nürnberg P, Spielmann M, Lemke JR, Li Y, Zenker M, Varshney GK, Hillen HS, Kratz CP, Wollnik B. 2022. WARS1 and SARS1: Two tRNA synthetases implicated in autosomal recessive microcephaly. *Hum Mutat* **43**:1454–1471. doi:10.1002/humu.24430
45. Lin SJ*, **Vona B***.#, Porter HM, Izadi M, Huang K, Lacassie Y, Rosenfeld JA, Khan S, Petree C, Ali TA, Muhammad Nazif, Khan SA, Muhammad Noor, Liu P, Haymon M-L, Rüschenhoff F, Kong I-K, Schnapp L, Shur N, Chorich L, Layman L, Haaf T, Pourkarimi E, Kim H-G, Varshney GK#. 2022. Biallelic variants in WARS1 cause a highly variable neurodevelopmental syndrome and implicate a critical exon for normal auditory function. *Hum Mutat* **43**:1472–1489. doi:10.1002/humu.24435 ***Shared first author, #Shared corresponding authors**
46. Kaiyrzhanov R, Perry L, Rocca C, Zaki MS, Hosny H, Araujo Martins Moreno C, Phadke R, Zaharieva I, Camelo Gontijo C, Beetz C, Pini V, Movahedinia M, Zanoteli E, DiTroia S, Vuillaumier-Barrot S, Isapof A, Mehrjardi MYV, Ghasemi N, Sarkozy A, Muntoni F, Whalen S, **Vona B**, Houlden H, Maroofian R. 2022. GGPS1-associated muscular dystrophy with and without hearing loss. *Ann Clin Transl Neurol* **9**:1465–1474. doi:10.1002/acn3.51633
47. Yao S, Zhou X, **Vona B**, Fan L, Zhang C, Li D, Yuan H, Du Y, Ma L, Pan Y. 2022. Skeletal Class III Malocclusion Is Associated with ADAMTS2 Variants and Reduced Expression in a Familial Case. *Int J Mol Sci* **23**:10673. doi:10.3390/ijms231810673
48. **Vona B**#, Regele S, Rad A, Strenzke N, Pater JA, Neumann K#, Sturm M, Haack TB, Am Zehnhoff-Dinnesen AG. 2023. Unraveling haplotype errors in the DFNA33 locus. *Front Genet* **14**:1214736. doi:10.3389/fgene.2023.1214736 **#Shared corresponding authors**
49. Pauly MG, Brüggemann N, Efthymiou S, Grözinger A, Diaw SH, Chelban V, Turchetti V, **Vona B**, Tadic V, Houlden H, Münchau A, Lohmann K. 2023. Not to Miss: Intronic Variants, Treatment, and Review of the Phenotypic Spectrum in VPS13D-Related Disorder. *Int J Mol Sci* **24**:1874. doi:10.3390/ijms24031874
50. Saadi SM, Cali E, Khalid LB, Yousaf H, Zafar G, Khan HN, Sher M, Vona B, Abdullah U, Malik NA, Klar J, Efthymiou S, Dahl N, Houlden H, Toft M, Baig SM, Fatima A, Iqbal Z. 2023. Genetic Investigation of Consanguineous Pakistani Families Segregating Rare Spinocerebellar Disorders. *Genes (Basel)* **14**:1404. doi:10.3390/genes14071404
51. Tropitzsch A, 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. 2023. Variability in Cochlear Implantation Outcomes in a Large German Cohort With

- a Genetic Etiology of Hearing Loss. *Ear Hear* **44**:1464–1484. doi:10.1097/AUD.0000000000001386
52. Engert J, Doll J, **Vona B**, Ehret Kasemo T, Spahn B, Hagen R, Rak K, Voelker J. 2023. mRNA Abundance of Neurogenic Factors Correlates with Hearing Capacity in Auditory Brainstem Nuclei of the Rat. *Life (Basel)* **13**:1858. doi:10.3390/life13091858
 53. Yao S, Zhou X, Gu M, Zhang C, Bartsch O, **Vona B**, Fan L, Ma L, Pan Y. 2023. FGFR1 variants contributed to families with tooth agenesis. *Hum Genomics* **17**:93. doi:10.1186/s40246-023-00539-8
 54. Uctepe E, **Vona B**, Esen FN, Sonmez FM, Smol T, Tümer S, Mancılar H, Geylan Durgun DE, Boute O, Moghbeli M, Ghayoor Karimiani E, Hashemi N, Bakhshoodeh B, Kim HG, Maroofian R, Yesilyurt A. 2024. Bi-allelic truncating variants in CASP2 underlie a neurodevelopmental disorder with lissencephaly. *Eur J Hum Genet* **32**:52–60. doi:10.1038/s41431-023-01461-2
 55. 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 Deirdre, Kelly Dominic, 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**:94. doi:10.1186/s13073-023-01240-0, **Top five most important paper**
 56. Kaiyrzhanov R, Rad A, Lin S-J, Bertoli-Avella A, Kallemeijn WW, Godwin A, Zaki MS, Huang K, Lau T, Petree C, Efthymiou S, Ghayoor Karimiani E, Hempel M, Normand EA, Rudnik-Schöneborn S, Schatz UA, Baggelaar MP, Ilyas M, Sultan T, Alvi JR, Ganieva M, Fowler B, Anicai R, Akay Tayfun G, Al Saman A, Alswaid A, Amiri N, Asilova N, Shotelersuk V, Yeetong P, Azam M, Babaei M, Bahrami Monajemi G, Mohammadi P, Samie S, Banu SH, Basto JP, Kortüm F, Bauer M, Bauer P, Beetz C, Garshasbi M, Hameed Issa A, Eyaid W, Ahmed H, Hashemi N, Hassanpour K, Herman I, Ibrohimov S, Abdul-Majeed BA, Imdad M, Isrofilov M, Kaiyal Q, Khan S, Kirmse B, Koster J, Lourenço CM, Mitani T, Moldovan O, Murphy D, Najafi M, Pehlivan D, Rocha ME, Salpietro V, Schmidts M, Shalata A, Mahroum M, Talbeya JK, Taylor RW, Vazquez D, Vetro A, Waterham HR, Zaman M, Schrader TA, Chung WK, Guerrini R, Lupski JR, Gleeson J, Suri M, Jamshidi Y, Bhatia KP, **Vona B**, Schrader M, Severino M, Guille M, Tate EW, Varshney GK, Houlden H, Maroofian R. 2023. Bi-allelic ACBD6 variants lead to a neurodevelopmental syndrome with progressive and complex movement disorders. *Brain* **147**(4):1436-1456. doi:10.1093/brain/awad380
 57. 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**:102. doi:10.1186/s13073-023-01258-4 ***Shared first author**

58. Maroofian R, Zamani M, Kaiyrzhanov R, Liebmann L, Ghayoor Karimiani E, **Vona B**, Huebner AK, Calame DG, Misra VK, Sadeghian S, Azizimalamiri R, Mohammadi MH, Zeighami J, Heydaran S, Beiraghi Toosi M, Akhondian J, Babaei M, Hashemi N, Schnur RE, Suri M, Setzke J, Wagner M, Brunet T, Grochowski CM, Emrick L, Chung WK, Hellmich UA, Schmidts M, Lupski JR, Galehdari H, Severino M, Houlden H, Hübner CA. 2023. Biallelic variants in SLC4A10 encoding the sodium-dependent chloride-bicarbonate exchanger NCBE lead to a neurodevelopmental disorder. *Genet Med* **26**(3):101034. doi:10.1016/j.gim.2023.101034
59. Chen H, Monga M, Fang Q, Slitin L, Neef J, Chepurwar SS, Mingroni Netto RC, Lezirovitz K, Tabith A, Benseler F, Brose N, Kusch K, Wichmann C, Strenzke N, **Vona B**, Preobraschenski J, Moser T. 2023. Ca²⁺-binding to the C2E domain of otoferlin is required for hair cell exocytosis and hearing. *Protein Cell* **15**(4):305-312. doi:10.1093/procel/pwad058
60. Efthymiou S, Scala M, Nagaraj V, Ochenkowska K, Komdeur FL, Liang RA, Abdel-Hamid MS, Sultan T, Barøy T, Van Ghelue M, **Vona B**, Maroofian R, Zafar F, Alkuraya FS, Zaki MS, Severino M, Duru KC, Tryon RC, Brauteset LV, Ansari M, Hamilton M, van Haelst MM, van Haaften G, Zara F, Houlden H, Samarut É, Nichols CG, Smeland MF, McClenaghan C. 2024. Novel loss-of-function variants expand ABCC9-related intellectual disability and myopathy syndrome. *Brain* **147**(5):1822-1836. doi:10.1093/brain/awae010
61. Mendia C, Peineau T, Zamani M, Felgerolle C, Yahiaoui N, Christophersen N, Papal S, Maudoux A, Maroofian R, Patni P, Nouaille S, Bowl MR, Delmaghani S, Galehdari H, **Vona B**, Dulon D, Vitry S, El-Amraoui A. 2024. Clarin-2 gene supplementation durably preserves hearing in a model of progressive hearing loss. *Mol Ther* **32**:800–817. doi:10.1016/j.ymthe.2024.01.021
62. Ramzan M, Zafeer MF, Abad C, Guo S, Owrang D, Alper O, Mutlu A, Atik T, Duman D, Bademci G, **Vona B**, Kalcioğlu MT, Walz K, Tekin M. 2024. Genetic heterogeneity in hereditary hearing loss: Potential role of kinociliary protein TOGARAM2. *Eur J Hum Genet* **32**(6):639-646. doi:10.1038/s41431-024-01562-6
63. **Vona B**^{*,#}. 2024. Rethinking non-syndromic hearing loss and its mimics in the genomic era. *Eur J Hum Genet*. doi:10.1038/s41431-024-01579-x ***First author, #Corresponding author**
64. Koparir A, Lekszas C, Keseroglu K, Rose T, Rappl L, Rad A, Maroofian R, Narendran N, Hasanazadeh A, Karimiani EG, Boschann F, Kornak U, Klopocki E, Özbudak EM, **Vona B**, Haaf T, Liedtke D. 2024. Zebrafish as a model to investigate a biallelic gain-of-function variant in MSGN1, associated with a novel skeletal dysplasia syndrome. *Hum Genomics* **18**:23. doi:10.1186/s40246-024-00593-w
65. 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. doi:10.1007/s00439-024-02649-2 ***Equal contribution, #Shared corresponding authors**
66. Chelban V, Aksnes H, Maroofian R, LaMonica LC, Seabra L, Siggervåg A, Devic P, Shamseldin HE, Vandrovcova J, Murphy D, Richard A-C, Quenez O, Bonneville A, Zanetti MN, Kaiyrzhanov R, Salpietro V, Efthymiou S, Schottlaender LV, Morsy H, Scardamaglia A, Tariq A, Pagnamenta AT, Pennavaria A, Krogstad LS, Bekkelund ÅK, Caiella A, Glomnes N, Brønstad KM, Tury S, Moreno De Luca A, Boland-Auge A, Olaso R, Deleuze J-F, Anheim M, Cretin B, **Vona B**, Alajlan F, Abdulwahab F, Battini J-L, İpek R, Bauer P, Zifarelli G, Gungor S, Kurul SH, Lochmuller H, Da'as SI, Fakhro KA, Gómez-Pascual A, Botía JA, Wood NW, Horvath R, Ernst AM, Rothman JE,

- McEntagart M, Crow YJ, Alkuraya FS, Nicolas G, SYNaPS Study Group, Arnesen T, Houlden H. 2024. Biallelic NAA60 variants with impaired n-terminal acetylation capacity cause autosomal recessive primary familial brain calcifications. *Nat Commun* **15**:2269. doi:10.1038/s41467-024-46354-0
67. Ma L, Zhou X, Siyue Y, **Vona B**, Fan L, Lou S, Li D, Wang L, Zhang X, Mao J. 2024. METTL3-dependent m6A modification of PSEN1 mRNA regulates craniofacial development through the Wnt/ β -catenin signaling pathway. *Cell Death Dis* **15**(3):229. doi: 10.1038/s41419-024-06606-9
68. Moser T, Chen H, Kusch K, Behr R, **Vona B**. 2024. Gene therapy for deafness: are we there now? *EMBO Mol Med* **16**(4):675-677. doi: 10.1038/s44321-024-00058-6
69. **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. doi: 10.1038/s12276-024-01247-6, **Top five most important paper**
70. Rad A, Bartsch O, Bakhtiari S, Zhu C, Xu Y, Monteiro FP, Kok F, Vulto-van Silfhout AT, Kruer MC, Bowl MR, **Vona B**[#]. 2024. Expanding the spectrum of phenotypes for MPDZ: Report of four unrelated families and review of the literature. *Clin Genet* **106**(4):413-426. doi: 10.1111/cge.14563 **#Corresponding author**
71. Alerasool M, Eslahi A, **Vona B**, Kahaei MS, Mojaver NK, Rajati M, Pasdar A, Ghasemi MM, Saburi E, Ardehaie RM, Aval MH, Tale MR, Nourizadeh N, Afzalzadeh MR, Niknezhad HT, Mojarrad M. 2024. Genetic landscape of hearing loss in prelingual deaf patients of eastern Iran: Insights from exome sequencing analysis. *Clin Genet* **106**(6):693-701. doi: 10.1111/cge.14599.
72. Akram R, Anwar H, Muzaffar H, Turchetti V, Lau T, Vona B, Makhdoom EUH, Iqbal J, Mahmood Baig S, Hussain G, Efthymiou S, Houlden H. 2024. A Novel MAG Variant Causes Hereditary Spastic Paraplegia in a Consanguineous Pakistani Family. *Genes* **15**:1203. doi:10.3390/genes15091203
73. 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. 2025. Elucidating the clinical and genetic spectrum of inositol polyphosphate phosphatase INPP4A-related neurodevelopmental disorder. *Genet Med* **27**(2):101278. doi: 10.1016/j.gim.2024.101278.
74. Rahman F, Marsili L, Pasquetti D, Rad A, Nadeem Anjum M, Oprea G, Cheema HA, **Vona B**, Augusto Alves C, Houlden H, Maqbool S, Efthymiou S, Smol T, Maroofian R. 2025. Bi-allelic MYMX variants cause a syndromic congenital myopathy with recognizable facial palsy, growth restriction, and dysmorphism. *Eur J Hum Genet* **33**:552–555. doi:10.1038/s41431-024-01759-9
75. Uctepe E, Mancilar H, Esen FN, Unverengil GG, **Vona B**, Yesilyurt A. 2025. A Homozygous MYH1 Variant Underlies Autosomal Recessive Isolated Recurrent Rhabdomyolysis. *Am J Med Genet A* **197**:e63952. doi:10.1002/ajmg.a.63952
76. Li D, Tian Y, **Vona B**, Yu X, Lin J, Ma L, Lou S, Li X, Zhu G, Wang Y, Du M, Wang L, Pan Y. 2025. A TAF11 variant contributes to non-syndromic cleft lip only through modulating neural crest cell migration. *Hum Mol Genet* **34**:392–401. doi:10.1093/hmg/ddae188

77. Tshering KC, DiStefano MT, Oza AM, Ajuyah P, Webb R, Edoth E, Broeren E, Ratliff J, Gitau V, Paris K, Aburyan A, Alexander J, Albano V, Bai D, Booth KTA, Buonfiglio PI, Charfeddine C, Dalamón V, Castillo ID, Moreno-Pelayo MA, Duzkale H, Dorshorst B, Faridi R, Kenna M, Lewis MA, Luo M, Lu Y, Mkaouar R, Matsunaga T, Nara K, Pandya A, Redfield S, Roux I, Schimmenti LA, Schrauwen I, Shaaban S, Shen J, **Vona B**, Smith RJ, Rehm HL, Azaiez H, Abou Tayoun AN, Amr SS, ClinGen Hearing Loss Clinical Domain Working Group. 2025. ClinGen recuration of hearing loss-associated genes demonstrates significant changes in gene-disease validity over time. *Genet Med* **27**:101392. doi:10.1016/j.gim.2025.101392
78. Efthymiou S, Leo CP, Deng C, Lin S-J, Maroofian R, Lin R, Karagoz I, Zhang K, Kaiyrzhanov R, Scardamaglia A, Owrang D, Turchetti V, Jahnke F, Huang K, Petree C, Derrick AV, Rees MI, Alvi JR, Sultan T, Li C, Jacquemont M-L, Tran-Mau-Them F, Valenzuela-Palafoll M, Sidlow R, Yoon G, Morrow MM, Carere DA, O'Connor M, Fleischer J, Gerkes EH, Phornphutkul C, Isidor B, Rivier-Ringenbach C, Philippe C, Kurul SH, Soydemir D, Kara B, Sunnetci-Akkoyunlu D, Bothe V, Platzer K, Wieczorek D, Koch-Hogrebe M, Rahner N, Thuresson A-C, Matsson H, Frykholm C, Bozdoğan ST, Bisgin A, Chatron N, Lesca G, Cabet S, Tümer Z, Hjortshøj TD, Rønde G, Marquardt T, Reunert J, Afzal E, Zamani M, Azizimalamiri R, Galehdari H, Nourbakhsh P, Chamanrou N, Chung S-K, Suri M, Benke PJ, Zaki MS, Gleeson JG, Calame DG, Pehlivan D, Yilmaz HI, Gezdirici A, Rad A, Abumansour IS, Oprea G, Bereketoğlu MB, Banneau G, Julia S, Zeighami J, Ashoori S, Shariati G, Sedaghat A, Sabri A, Hamid M, Parvas S, Tajudin TA, Abdullah U, Baig SM, Chung WK, Glazunova OO, Sabine S, Cheema HA, Zifarelli G, Bauer P, Sidpra J, Mankad K, **Vona B**, Fry AE, Varshney GK, Houlden H, Fu D. 2025. Bi-allelic pathogenic variants in TRMT1 disrupt tRNA modification and induce a neurodevelopmental disorder. *Am J Hum Genet* **112**:1117–1138. doi:10.1016/j.ajhg.2025.03.015
79. Shadab M, Ben-Mahmoud A, Martínez Völter LN, Abbasi AA, Ku B, Ejaz A, Latif Z, Gupta V, Owrang D, Jang M-H, Zhang Z, Mohammad R, Houlden H, Kim H-G, **Vona B**. 2025. Recurrent and Novel Pathogenic Variants in Genes Involved with Hearing Loss in the Pakistani Population. *Mol Diagn Ther* **29**:519–537. doi:10.1007/s40291-025-00782-w
80. Bastille I, Lee L, Moncada-Reid C, Yu W-M, Sitko A, Yung A, Zamani M, Christophersen N, Maroofian R, Galehdari H, Babai N, **Vona B**, Moser T, Goodrich L. 2025. Combinatorial transcriptional regulation establishes subtype-appropriate synaptic properties in auditory neurons. *Cell Rep* **44**:115796. doi:10.1016/j.celrep.2025.115796
81. Andreae H[#], Curcio M, Owrang D, Esmaeelpour S, Jahnke F, Benseler F, Brose N, **Vona B**[#]. 2025. Protocol for a minigene splice assay using the pET01 vector. *STAR Protoc* **6**:103908. doi:10.1016/j.xpro.2025.103908 **#Shared corresponding authors**
82. 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 H-G, Ekmekci C, Karimiani EG, Imannezhad S, Eghbal F, Badv RS, Schwaibold EMC, Dehghani M, Mehrjardi MYV, Metanat Z, Eslamiyeh H, Khouj E, Alhajj SMN, 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, Alves CAPF, 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**:1240–1255. doi:10.1038/s41556-025-01708-8 ***Shared first author, Top five most important paper**
83. **Vona B**, Strenzke N. 2025. Gradual recovery of auditory brainstem responses in the first DFNB9 patients with successful virus-mediated gene therapy. *Med* **6**:100775. doi:10.1016/j.medj.2025.100775

84. **Vona B**, Wollnik B, Strenzke N, Pangršič T, Moser T. 2025. Is CABP2-Associated Hearing Loss (DFNB93) a Gene Therapy Target? Preclinical Progress and Patient Registry. *MedComm* **6**:e70363. doi:10.1002/mco2.70363, **Top five most important paper**
85. Dekker J, Schot R, Aldinger KA, Everman DB, Washington C, Jones JR, Sullivan JA, Spillmann RC, Shashi V, Vitobello A, Denommé-Pichon AS, Mosca-Boidron AL, Perrin L, Auvin S, Zaki MS, Gleeson JG, Meave N, Wallace C, Nambot S, Delanne J, Ruggiero SM, Helbig I, Fitzgerald MP, Leventer RJ, Grange DK, Argilli E, Sherr EH, Prakash S, Neilson DE, Nicita F, Sferra A, Bertini ES, Aiello C, Brockmann K, Kuranov AB, Kaulfuss S, Basit S, Alluqmani M, Almatrafi A, Friedman JM, Guimond C, Mohammed F, Sharma P, Goel D, Wirth T, Anheim M, Bahena P, Koparir A, Kolokotronis K, **Vona B**, Haaf T, Kunstmann E, Maroofian R, Sczakiel HL, Boschann F, Misra-Isrie M, Louie RJ, Stolerma ES, Sanchez-Lara PA, Mergler S, Oegema R, Zarate YA, Kariminejad A, Tajsharghi H, Zeidler S, Kievit AJA, Bouman A, Cappuccio G, Brunetti-Pierri N, Stuurman KE, Swols DM, Tekin M, Upadia J, Martin DM, Craven D, Hiatt SM, van de Pol LA, D'Arco F, Margot H, Wilke M, Yousefi S, Barakat TS, van Veghel-Plandsoen MM, Aronica E, Anink J, Rogers SL, Slep KC, Doherty D, Dobyns WB, Mancini GMS. 2025. A clinical and genotype-phenotype analysis of MACF1 variants. *Am J Hum Genet* [Online ahead of print]. doi: 10.1016/j.ajhg.2025.08.010.
86. 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].

ii. Reviews

87. **Vona B**[#], Nanda I, Hofrichter MAH, Shehata-Dieler W, Haaf T. 2015. Non-syndromic hearing loss gene identification: A brief history and glimpse into the future. *Mol Cell Probes* **29**:260–270. doi:10.1016/j.mcp.2015.03.008 **#Corresponding author**
88. **Vona B**[#], Nanda I, Shehata-Dieler W, Haaf T. 2017. Genetics of Tinnitus: Still in its Infancy. *Front Neurosci* **11**:236. doi:10.3389/fnins.2017.00236 **#Corresponding author**
89. **Vona B**, Müller M, Dofek S, Holderried M, Löwenheim H, Tropitzsch A. 2019. A Big Data Perspective on the Genomics of Hearing Loss. *Laryngorhinootologie* **98**:S32–S81. doi:10.1055/a-0803-6149
90. **Vona B**[#], Doll J, Hofrichter MAH, Haaf T, Varshney GK[#]. 2020. Small fish, big prospects: using zebrafish to unravel the mechanisms of hereditary hearing loss. *Hear Res* **397**:107906. doi:10.1016/j.heares.2020.107906 **#Shared corresponding authors**
91. **Vona B**, Rad A, Reisinger E. 2020. The Many Faces of DFNB9: Relating OTOF Variants to Hearing Impairment. *Genes* **11**:1411. doi:10.3390/genes11121411
92. Peixoto Pinheiro B, **Vona B**, Löwenheim H, Rüttiger L, Knipper M, Adel Y. 2021. Age-related hearing loss pertaining to potassium ion channels in the cochlea and auditory pathway. *Pflugers Arch* **473**:823–840. doi:10.1007/s00424-020-02496-w
93. Wolf BJ, Kusch K, Hunniford V, **Vona B**, Kühler R, Keppeler D, Strenzke N, Moser T. 2022. Is there an unmet medical need for improved hearing restoration? *EMBO Mol Med* **14**:e15798. doi:10.15252/emmm.202215798
94. Shadab M[#], Abbasi AA, Ejaz A, Ben-Mahmoud A, Gupta V, Kim H-G, **Vona B**[#]. 2024. Autosomal recessive non-syndromic hearing loss genes in Pakistan during the previous three decades. *J Cell Mol Med* **28**(8):e18119. doi:10.1111/jcmm.18119 **#Shared corresponding authors**

95. Guan C, Shaikh M, Warnecke A, **Vona B**[#], Albert JT[#]. 2024. A burden shared: The evolutionary case for studying human deafness in *Drosophila*. *Hear Res* **450**:109047. doi: 10.1016/j.heares.2024.109047 **#Shared corresponding authors**

iii. Pre-prints

96. Magrinelli F, Tesson C, Angelova PR, Salazar-Villacorta A, Rodriguez JA, Scardamaglia A, Chung BH, Jacconelli M, **Vona B**, Esteras N, Kwong AK, Courtin T, Maroofian R, Alavi S, Nirujogi R, Severino M, Lewis PA, Efthymiou S, O'Callaghan B, Buchert R, Sofan L, Lis P, Pinon C, Breedveld GJ, Chui MM, Murphy D, Pitz V, Makarios MB, Cassar M, Hassan BA, Iftikhar S, Rocca C, Bauer P, Tinazzi M, Svetel M, Samanci B, Hanağası HA, Bilgiç B, Obeso JA, Kurtis MM, Cogan G, Başak AN, Kiziltan G, Gül T, Yalçın G, Elibol B, Barišić N, Ng EW, Fan SS, HersHKovitz T, Weiss K, Raza Alvi J, Sultan T, Azmi Alkhawaja I, Froukh T, E Alrukban HA, Fauth C, Schatz UA, Zöggeler T, Zech M, Stals K, Varghese V, Gandhi S, Blauwendraat C, Hardy JA, Lesage S, Bonifati V, Haack TB, Bertoli-Avella AM, Steinfeld R, Alessi DR, Steller H, Brice A, Abramov AY, Bhatia KP, Houlden H. 2024. PSMF1 variants cause a phenotypic spectrum from early-onset Parkinson's disease to perinatal lethality by disrupting mitochondrial pathways. *medRxiv* 20:2024.06.19.24308302. doi: 10.1101/2024.06.19.24308302
97. Gogate N, Jolly A, Rosenfeld JA, Bahena-Carbajal P, Bernstein JA, Bonner D, Busa T, Cristian I, D'Souza P, Friedman J, Gorokhova S, Haaf T, Herman I, Isin UU, Jhangiani SN, Johnson I, Lenberg J, Macnamara EF, Maroofian R, Undiagnosed Diseases Network, Melissa Racobaldo, Redlich OL, Tifft C, Tos T, **Vona B**, Zambrano RM, Wentzensen IM, Wigby K, Pehlivan D, Gibbs RA, Lupski JR, Posey JE. 2025. Domain specific phenotypic expansion associated with variants in MACF1. *MedRxiv Prepr Serv Health Sci* 2025.06.26.25330137. doi:10.1101/2025.06.26.25330137
98. Hale EB, **Vona B**, Goodyear RJ, Osgood RT, Amr SS, Mojica K, Vera-Monroy R, Callahan K, Gudlewski KL, Quadros R, Ohtsuka M, McGee J, Walsh EJ, Morton CC, Gurumurthy C, Saunders JE, Richardson GP, Indzhukulian AA. 2025. The TECTB-C225Y Variant Causing Autosomal Dominant Deafness in a Nicaraguan Family Enhances Sensitivity to Noise-Induced Hearing Loss in Mice. *MedRxiv* 2025.08.13.25333146. doi:10.1101/2025.08.13.25333146