

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**
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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
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 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
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 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
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 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, Mojarab 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, Gamerding 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, Podeshsked 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, Top five most important paper**
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 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**
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 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**,

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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, Top five most important paper**
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Disorder and Movement Abnormalities. *Mov Disord Clin Pract* **8**:1140–1143.
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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-Meiri 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, Alfaridhi 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, 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**:1606–1611.
doi:10.1111/jdv.18207 ***Equal contribution**
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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
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