

Curriculum Vitae

Sandra Gräfin von Hardenberg PD Dr. rer. nat.

d.o.b. April 23th, 1982, in Minden, Germany

University Education

- 2025 Habilitation "Experimental Human Genetics", Hannover Medical School
- 2010 Dissertation, Institute of Human Genetics, University Medical Center Göttingen
- 2007 Diploma, Institute of Human Genetics, University Medical Center Göttingen
- 2002-2006 Studies in Biology at the University of Göttingen (Germany) and Lund (Sweden)
- 2001-2002 Studies in Mathematics and Biology for Secondary School Teaching at the Carl von Ossietzky University of Oldenburg (Germany)

Scientific Career

- Since 2025 Research Group Leader for the "Gene Discovery" Process at the Department of Human Genetics, Hannover Medical School
- Since 2020 Member „Arbeitsgemeinschaft Pädiatrische Immunologie“
- Since 2018 Research Associate and Co-Responsible Scientist for Human Genetic Diagnostics, Hannover Medical School
- 2017-2018 Research Associate at the Department of Obstetrics and Gynecology, Hannover Medical School
- Since 2016 Member „Deutsche Gesellschaft für Humangenetik“
- 2015-2017 Research Associate at the Department of Human Genetics, Hannover Medical School
- 2013-2015 Research Associate at the Department of Pharmaco- and Toxicogenomics, Hannover Medical School

Citation Record

Total citations: 761; h-index:15; h-index since 2021: 14 (Google Scholar February 2nd, 2026)

Top-10 selected Publications

- 1) Elsayed A, von Hardenberg S, Atschekzei F, Siek P, Witte T, Sogkas G, Ringshausen FC. A novel hemizygous nonsense variant in DOCK11 causes systemic inflammation and immunodeficiency. *Clin Immunol*. 2025 Jul;276:110504. doi: 10.1016/j.clim.2025.110504. Epub 2025 Apr 22. PMID: 40274249.
- 2) Schütz K, Schmidt A, Schwerk N, Renz DM, Gerard B, Schaefer E, Antal MC, Peters S, Griesse M, Rapp CK, Engels H, Cremer K, Bergmann AK, Schmidt G, Auber B, Kamp JC, Laenger F, von Hardenberg S. Variants in FGF10 cause early onset of severe childhood interstitial lung disease: A detailed description of four affected children. *Pediatr Pulmonol*. 2023 Nov;58(11):3095-3105. doi: 10.1002/ppul.26627. Epub 2023 Aug 10. PMID: 37560881.
- 3) Raidt J, Riepenhausen S, Pennekamp P, Olbrich H, Amirav I, Athanazio RA, Aviram M, Balinotti JE, Bar-On O, Bode SFN, Boon M, Borrelli M, Carr SB, Crowley S, Dehlink E, Diepenhorst S, Durdik P, Dworniczak B, Emiralioglu N, Erdem E, Fonnesu R, Gracci S, Große-Onnebrink J, Gwozdziwicz K, Haarman EG, Hansen CR, Hogg C, Holgersen MG, Kerem E, Körner RW, Kötz K, Kouis P, Loebinger MR, Lorent N, Lucas JS, Maj D, Mall MA, Marthin JK, Martinu V, Mazurek H, Mitchison HM, Nöthe-Menchen T, Özçelik U, Pifferi M, Pogorzelski A, Ringshausen FC, Roehmel JF, Rovira-Amigo S, Rumman N, Schlegtendal A, Shoemark A, Sperstad Kennelly S, Staar BO, Sutharsan S, Thomas S, Ullmann N, Varghese J, von Hardenberg S, Walker WT, Wetzke M, Witt M, Yiallourous P, Zschocke A, Ziętkiewicz E, Nielsen KG, Omran H. Analyses of 1236 genotyped primary ciliary dyskinesia individuals identify regional clusters of distinct DNA variants and significant genotype-phenotype correlations. *Eur Respir J*. 2024 Aug 8;64(2):2301769. doi: 10.1183/13993003.01769-2023. PMID: 38871375; PMCID: PMC11306806.
- 4) Elsayed A, von Hardenberg S, Atschekzei F, Graalman T, Jänke C, Witte T, Ringshausen FC, Sogkas G. Phenotypic and pathomechanistic overlap between tapasin and TAP deficiencies. *J Allergy Clin Immunol*. 2024 Oct;154(4):1069-1075. doi: 10.1016/j.jaci.2024.06.003. Epub 2024 Jun 10. PMID: 38866210.
- 5) Nashabat M, Nabavizadeh N, Saraçoğlu HP, Sarıbaş B, Avcı Ş, Börklü E, Beillard E, Yılmaz E, Uygur SE, Kayhan CK, Bosco L, Eren ZB, Steindl K, Richter MF, Bademci G, Rauch A, Fattahi Z, Valentino ML, Connolly AM, Bahr A, Viola L, Bergmann AK, Rocha ME, Peart L, Castro-Rojas DL, Bültmann E, Khan S, Giarrana ML, Teleanu RI, Gonzalez JM, Pini A, Schädlich IS, Vill K, Brugger M, Zuchner S, Pinto A, Donkervoort S, Bivona SA, Riza A; Undiagnosed Diseases Network; Streata I, Gläser D, Baquero-Montoya C, Garcia-Restrepo N, Kotzaeridou U, Brunet T, Epure DA, Bertoli-Avella A, Kariminejad A, Tekin M, von Hardenberg S, Bönnemann CG, Stettner GM, Zanni G, Kayserili H, Oflazer ZP, Escande-Beillard N. SNUPN deficiency causes a recessive muscular dystrophy due to RNA mis-splicing and ECM dysregulation. *Nat Commun*. 2024 Feb 27;15(1):1758. doi: 10.1038/s41467-024-45933-5. PMID: 38413582; PMCID: PMC10899626.
- 6) Scala M, Khan K, Beneteau C, Fox RG, von Hardenberg S, Khan A, Joubert M, Fievet L, Musquer M, Le Vaillant C, Holsclaw JK, Lim D, Berking AC, Accogli A, Giacomini T, Nobili L, Striano P, Zara F, Torella A, Nigro V, Cogné B, Salick MR, Kaykas A, Eggan K, Capra V, Béziau S, Davis EE, Wells MF. Biallelic loss- of-function variants in CACHD1 cause a novel neurodevelopmental syndrome with facial dysmorphism and multisystem congenital abnormalities. *Genet Med*. 2024 Apr;26(4):101057. doi:

- 10.1016/j.gim.2023.101057. Epub 2023 Dec 27. PMID: 38158856; PMCID: PMC11910193.
- 7) Kamp JC, Neubert L, Schupp JC, Braubach P, Wrede C, Laenger F, Salditt T, Reichmann J, Welte T, Ruhparwar A, Ius F, Schwerk N, Bergmann AK, von Hardenberg S, Griese M, Rapp C, Olsson KM, Fuge J, Park DH, Hoeper MM, Jonigk DD, Knudsen L, Kuehnel MP. Multilamellated Basement Membranes in the Capillary Network of Alveolar Capillary Dysplasia. *Am J Pathol.* 2024 Feb;194(2):180-194. doi: 10.1016/j.ajpath.2023.10.012. Epub 2023 Nov 27. PMID: 38029923; PMCID: PMC12178336.
 - 8) von Hardenberg S, Wallaschek H, Du C, Schmidt G, Auber B. A holistic approach to maximise diagnostic output in trio exome sequencing. *Front Pediatr.* 2023 May 19;11:1183891. doi: 10.3389/fped.2023.1183891. PMID: 37274821; PMCID: PMC10238563.
 - 9) Wan R, Fänder J, Zakaraia I, Lee-Kirsch MA, Wolf C, Lucas N, Olfe LI, Hendrich C, Jonigk D, Holzinger D, Steindor M, Schmidt G, Davenport C, Klemann C, Schwerk N, Griese M, Schlegelberger B, Stehling F, Happle C, Auber B, Steinemann D, Wetzke M, von Hardenberg S. Phenotypic spectrum in recessive STING-associated vasculopathy with onset in infancy: Four novel cases and analysis of previously reported cases. *Front Immunol.* 2022 Oct 6;13:1029423. doi: 10.3389/fimmu.2022.1029423. PMID: 36275728; PMCID: PMC9583393.
 - 10) Vavassori S, Chou J, Faletti LE, Haunerding V, Opitz L, Joset P, Fraser CJ, Prader S, Gao X, Schuch LA, Wagner M, Hoefele J, Maccari ME, Zhu Y, Elakis G, Gabbett MT, Forstner M, Omran H, Kaiser T, Kessler C, Olbrich H, Frosk P, Almutairi A, Platt CD, Elkins M, Weeks S, Rubin T, Planas R, Marchetti T, Koovely D, Klämbt V, Soliman NA, von Hardenberg S, Klemann C, Baumann U, Lenz D, Klein-Franke A, Schwemmle M, Huber M, Sturm E, Hartleif S, Häffner K, Gimpel C, Brotschi B, Laube G, Gungör T, Buckley MF, Kottke R, Staufner C, Hildebrandt F, Reu-Hofer S, Moll S, Weber A, Kaur H, Ehl S, Hiller S, Geha R, Roscioli T, Griese M, Pachlopnik Schmid J. Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. *J Allergy Clin Immunol.* 2021 Aug;148(2):381-393. doi: 10.1016/j.jaci.2021.03.045. Epub 2021 Apr 17. PMID: 33872655; PMCID: PMC8569286.