DR. MED. BERNT POPP

Curriculum Vitae

As a physician and scientist I am currently working at the BIH at Charité Berlin and at Labor Berlin — Charité Vivantes GmbH.

From a human genetics perspective, my scientific interests are in rare diseases of neuronal development, rare tumors and the kidney. I particularly enjoy working bioinformatically on the analysis of data from high-throughput sequencing and on the curation of genetic diseases, variants and genes.

My current scientific focus within the CADS research project is the development of digital tools to support clinical decision-making and the analysis of genetic data, particularly in the field of nephrological diseases. This includes the development of algorithms for the detection of genetic variants and the integration of patient data to improve the treatment of rare kidney diseases.

ACADEMIC AND MEDICAL-CLINICAL CAREER

since 01/2023

Project lead Innovation in Human Genetics (Senior physician equivalent) at Labor Berlin — Charité Vivantes GmbH (50%)
Labor Berlin — Charité Vivantes GmbH — Berlin, Germany

Department of Human Genetics at Labor Berlin — Charité Vivantes GmbH

since 07/2022

Senior physician and Postdoc at Charité – Universitätsmedizin Berlin (50%)
Berlin Institute of Health (BIH) — Berlin, Germany

Translational Research Area - Research Group “Hypertension and Molecular Biology of Endocrine Tumors” (Prof. Dr. med. Ute Scholl)

04/2022 | 07/2021

Senior physician and head of the genetics outpatient clinic at MVZ Dresden
Medizinisches Versorgungszentrum des Universitätsklinikums Dresden — Dresden, Germany

Head of the genetic outpatient clinic as senior physician

06/2021 | 04/2021

Specialist in Human Genetics
at the Institute of Human Genetics, University of Leipzig Medical Center, Leipzig, Germany — Leipzig, Germany

Deputy Team Leader Genetic Diagnostics - Clinical Genomics

KONTAKT

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COMPETENCE

Specialist in human genetics with 12 years of experience in genetic diagnostics and counseling.

Scientist with experience in high throughput sequencing, data analysis, rare diseases of neural development and kidney.

Bioinformatician with experience in in R, Bash, Python, Javascript, workflow automation, virtualization and Linux administration.

Last change 2025-03-17.

03/2021

Recognition as a specialist in Human Genetics
Completed specialist training and examination by the Saxony State Medical Association

Dresden, Germany

Allowed to use the title “Specialist in Human Genetics” (German: “Facharzt für Humangenetik”)

since 01/2020

DFG (German Research Foundation) rotation position
Scientific-medical assistant at the Human Genetics Institute of the University Hospital Leipzig

Leipzig, Germany

Within the funds raised for the project “Exome Pool-Seq and systems biology approach to identify and characterize genes and networks in neurodevelopmental disorders”

03/2021 | 06/2019

Resident
at the Institute of Human Genetics, University of Leipzig Medical Center, Leipzig, Germany

Leipzig, Germany

Training as a specialist in human genetics

09/2020 | 09/2019

Clinical year
as an assistant physician in the field of nephrology at the Clinic and Polyclinic for Endocrinology, Nephrology, Rheumatology at the University Hospital Leipzig

Leipzig, Germany

Training as a specialist in human genetics

05/2019 | 02/2013

Resident and scientific-medical assistant
at the Institute of Human Genetics, University Hospital Erlangen, Friedrich-Alexander-Universität Erlangen-Nürnberg (FAU), Erlangen, Germany

Erlangen, Germany

Training as a specialist in human genetics

11/2015 | 02/2013

Dissertation Dr. med.
Institute of Human Genetics, University Hospital Erlangen, Friedrich-Alexander-Universität Erlangen-Nürnberg (FAU), Erlangen, Germany

Erlangen, Germany

Title of the dissertation „De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females” (summa cum laude)

11/2012

License as physician (Appropation)
after completing medical studies at the FAU Erlangen-Nuremberg
Erlangen, Germany

EDUCATION

10/2021 | 10/2019

Part-time study Bachelor of Science Computer Science
at the state distance-learning university FernUniversität in Hagen
Hagen, Germany

11/2012 | 10/2005

Medical School
FAU Erlangen-Nürnberg (Staatsexamen) Erlangen, Germany

06/2005 | 09/1996

General university entrance qualification (Allgemeine Hochschulreife)
Peter-Vischer-Gymnasium, Nuremberg Nuremberg, Germany

MEMBER OF PROFESSIONAL ASSOCIATIONS

- GfH - Gesellschaft für Humangenetik
- ESHG - European Society of Human Genetics
- ASHG - American Society of Human Genetics

PUBLICATIONS

Only first and last author positions (including equal contributions) are listed here. A complete and always up-to-date list can be found at Google Scholar.

Bartolomaeus, T., Hentschel, J., Jamra, R. A., & Popp, B. (2023). Re-evaluation and re-analysis of 152 research exomes five years after the initial report reveals clinically relevant changes in 18%. *European Journal of Human Genetics*, *31*(10), 1154–1164. https://doi.org/10.1038/s41431-023-01425-6

Bosch, E., Popp, B., Güse, E., Skinner, C., Van Der Sluijs, P. J., Maystadt, I., Pinto, A. M., Renieri, A., Bruno, L. P., Granata, S., Marcelis, C., Baysal, Ö., Hartwich, D., Holthöfer, L., Isidor, B., Cogne, B., Wieczorek, D., Capra, V., Scala, M., De Marco, P., Ognibene, M., Abou Jamra, R., Platzer, K., Carter, L. B., Kuismin, O., Van Haeringen, A., Maroofian, R., Valenzuela, I., Cuscó, I., Martinez-Agosto, J. A., Rabani, A. M., Mefford, H. C., Pereira, E. M., Close, C., Anyane-Yeboa, K., Wagner, M., Hannibal, M. C., Zacher, P., Thiffault, I., Beunders, G., Umair, M., Bhola, P. T., McGinnis, E., Millichap, J., Van De Kamp, J. M., Prijoles, E. J., Dobson, A., Shillington, A., Graham, B. H., Garcia, E.-J., Kelly Galindo, M., Ropers, F. G., Nibbeling, E. Ar., Hubbard, G., Karimov, C., Goj, G., Bend, R., Rath, J., Morrow, M. M., Millan, F., Salpietro, V., Torella, A., Nigro, V., Kurki, M., Stevenson, R. E., Santen, G. W. E., Zweier, M., Campeau, P. M., Severino, M., Reis, A., Accogli, A., & Vasileiou, G. (2023). Elucidating the clinical and molecular spectrum of SMARCC2-associated NDD in a cohort of 65 affected individuals. *Genetics in Medicine*, 100950. https://doi.org/10.1016/j.gim.2023.100950

Lehmann, C., Pehnke, S., Weimann, A., Bachmann, A., Dittrich, K., Petzold, F., Fürst, D., De Fallois, J., Landgraf, R., Henschler, R., Lindner, T. H., Halbritter, J., Doxiadis, I., Popp, B., & Münch, J. (2023). Extended genomic HLA typing identifies previously unrecognized mismatches in living kidney transplantation. *Frontiers in Immunology*, *14*, 1094862. https://doi.org/10.3389/fimmu.2023.1094862

Popp, B., Brugger, M., Poschmann, S., Bartolomaeus, T., Radtke, M., Hentschel, J., Di Donato, N., Rump, A., Gburek-Augustat, J., Graf, E., Wagner, M., Sorge, I., Lemke, J. R., Meitinger, T., Abou Jamra, R., Strehlow, V., & Brunet, T. (2023). The constitutional gain-of-function variant p.Glu1099Lys in NSD2 is associated with a novel syndrome. *Clinical Genetics*, *103*(2), 226–230. https://doi.org/10.1111/cge.14241

Roessler, F., Beck, A. E., Susie, B., Tobias, B., Begtrup, A., Biskup, S., Caluseriu, O., Delanty, N., Fröhlich, C., Greally, M. T., Karnstedt, M., Klöckner, C., Kurtzberg, J., Schubert, S., Schulze, M., Weidenbach, M., Westphal, D. S., White, M., Wolf, C. M., Zyskind, J., Popp, B., & Strehlow, V. (2023). Genetic and phenotypic spectrum in the NONO-associated syndromic disorder. *American Journal of Medical Genetics Part A*, *191*(2), 469–478. https://doi.org/10.1002/ajmg.a.63044

Klau, J., Abou Jamra, R., Radtke, M., Oppermann, H., Lemke, J. R., Beblo, S., & Popp, B. (2022). Exome first approach to reduce diagnostic costs and time – retrospective analysis of 111 individuals with rare neurodevelopmental disorders. *European Journal of Human Genetics*, *30*(1), 117–125. https://doi.org/10.1038/s41431-021-00981-z

Lieberwirth, J. K., Büttner, B., Klöckner, C., Platzer, K., Popp, B., & Abou Jamra, R. (2022). AutoCaSc: Prioritizing candidate genes for neurodevelopmental disorders. *Human Mutation*, *43*(12), 1795–1807. https://doi.org/10.1002/humu.24451

Münch, J., Engesser, M., Schönauer, R., Hamm, J. A., Hartig, C., Hantmann, E., Akay, G., Pehlivan, D., Mitani, T., Coban Akdemir, Z., Tüysüz, B., Shirakawa, T., Dateki, S., Claus, L. R., Van Eerde, A. M., Smol, T., Devisme, L., Franquet, H., Attié-Bitach, T., Wagner, T., Bergmann, C., Höhn, A. K., Shril, S., Pollack, A., Wenger, T., Scott, A. A., Paolucci, S., Buchan, J., Gabriel, G. C., Posey, J. E., Lupski, J. R., Petit, F., McCarthy, A. A., Pazour, G. J., Lo, C. W., Popp, B., & Halbritter, J. (2022). Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. *Kidney International*, *101*(5), 1039–1053. https://doi.org/10.1016/j.kint.2022.01.028

Neuser, S., Krey, I., Schwan, A., Abou Jamra, R., Bartolomaeus, T., Döring, J., Syrbe, S., Plassmann, M., Rohde, S., Roth, C., Rehder, H., Radtke, M., Le Duc, D., Schubert, S., Bermúdez-Guzmán, L., Leal, A., Schoner, K., & Popp, B. (2022). Prenatal phenotype of PNKP-related primary microcephaly associated with variants affecting both the FHA and phosphatase domain. *European Journal of Human Genetics*, *30*(1), 101–110. https://doi.org/10.1038/s41431-021-00982-y

Popp, B., Bienvenu, T., Giurgea, I., Metreau, J., Kraus, C., Reis, A., Fischer, J., Bralo, M. P., Tenorio-Castaño, J., Lapunzina, P., Almoguera, B., Lopez-Grondona, F., Sticht, H., & Zweier, C. (2022). The recurrent TCF4 missense variant p.(Arg389Cys) causes a neurodevelopmental disorder overlapping with but not typical for Pitt-Hopkins syndrome. *Clinical Genetics*, *102*(6), 517–523. https://doi.org/10.1111/cge.14206

Popp, B., Ekici, A. B., Knaup, K. X., Schneider, K., Uebe, S., Park, J., Bafna, V., Meiselbach, H., Eckardt, K.-U., Schiffer, M., Reis, A., Kraus, C., & Wiesener, M. (2022). Prevalence of hereditary tubulointerstitial kidney diseases in the GermanChronicKidneyDisease study. *European Journal of Human Genetics*, *30*(12), 1413–1422. https://doi.org/10.1038/s41431-022-01177-9

Neuser, S., Brechmann, B., Heimer, G., Brösse, I., Schubert, S., O'Grady, L., Zech, M., Srivastava, S., Sweetser, D. A., Dincer, Y., Mall, V., Winkelmann, J., Behrends, C., Darras, B. T., Graham, R. J., Jayakar, P., Byrne, B., Bar-Aluma, B. E., Haberman, Y., Szeinberg, A., Aldhalaan, H. M., Hashem, M., Al Tenaiji, A., Ismayl, O., Al Nuaimi, A. E., Maher, K., Ibrahim, S., Khan, F., Houlden, H., Ramakumaran, V. S., Pagnamenta, A. T., Posey, J. E., Lupski, J. R., Tan, W., ElGhazali, G., Herman, I., Muñoz, T., Repetto, G. M., Seitz, A., Krumbiegel, M., Poli, M. C., Kini, U., Efthymiou, S., Meiler, J., Maroofian, R., Alkuraya, F. S., Abou Jamra, R., Popp, B., Ben-Zeev, B., & Ebrahimi-Fakhari, D. (2021). Clinical, neuroimaging, and molecular spectrum of *TECPR2* -associated hereditary sensory and autonomic neuropathy with intellectual disability. *Human Mutation*, *42*(6), 762–776. https://doi.org/10.1002/humu.24206

Popp, B., Erber, R., Kraus, C., Vasileiou, G., Hoyer, J., Burghaus, S., Hartmann, A., Beckmann, M. W., Reis, A., & Agaimy, A. (2020). Targeted sequencing of FH-deficient uterine leiomyomas reveals biallelic inactivating somatic fumarase variants and allows characterization of missense variants. *Modern Pathology*, *33*(11), 2341–2353. https://doi.org/10.1038/s41379-020-0596-y

Hebebrand, M., Hüffmeier, U., Trollmann, R., Hehr, U., Uebe, S., Ekici, A. B., Kraus, C., Krumbiegel, M., Reis, A., Thiel, C. T., & Popp, B. (2019). The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. *Orphanet Journal of Rare Diseases*, *14*(1), 38. https://doi.org/10.1186/s13023-019-1020-x

Hebebrand, M., Vasileiou, G., Krumbiegel, M., Kraus, C., Uebe, S., Ekici, A. B., Thiel, C. T., Reis, A., & Popp, B. (2019). A biallelic truncating *AEBP1* variant causes connective tissue disorder in two siblings. *American Journal of Medical Genetics Part A*, *179*(1), 50–56. https://doi.org/10.1002/ajmg.a.60679

Popp, B., Agaimy, A., Kraus, C., Knaup, K. X., Ekici, A. B., Uebe, S., Reis, A., Wiesener, M., & Zweier, C. (2019). Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. *BMC Cancer*, *19*(1), 435. https://doi.org/10.1186/s12885-019-5633-1

Vasileiou, G., Hoyer, J., Thiel, C. T., Schaefer, J., Zapke, M., Krumbiegel, M., Kraus, C., Zweier, M., Uebe, S., Ekici, A. B., Schneider, M., Wiesener, M., Rauch, A., Faschingbauer, F., Reis, A., Zweier, C., & Popp, B. (2019). Prenatal diagnosis of *HNF1B* -associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome? *Prenatal Diagnosis*, *39*(12), 1136–1147. https://doi.org/10.1002/pd.5556

Popp, B., Krumbiegel, M., Grosch, J., Sommer, A., Uebe, S., Kohl, Z., Plötz, S., Farrell, M., Trautmann, U., Kraus, C., Ekici, A. B., Asadollahi, R., Regensburger, M., Günther, K., Rauch, A., Edenhofer, F., Winkler, J., Winner, B., & Reis, A. (2018). Need for high-resolution GeneticAnalysis in iPSC: Results and Lessons from the ForIPSConsortium. *Scientific Reports*, *8*(1), 17201. https://doi.org/10.1038/s41598-018-35506-0

Popp, B., Ekici, A. B., Thiel, C. T., Hoyer, J., Wiesener, A., Kraus, C., Reis, A., & Zweier, C. (2017). Exome Pool-Seq in neurodevelopmental disorders. *European Journal of Human Genetics*, *25*(12), 1364–1376. https://doi.org/10.1038/s41431-017-0022-1

Popp, B., Trollmann, R., Büttner, C., Caliebe, A., Thiel, C. T., Hüffmeier, U., Reis, A., & Zweier, C. (2016). Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. *European Journal of Medical Genetics*, *59*(10), 549–553. https://doi.org/10.1016/j.ejmg.2016.09.002

Popp, B., Støve, S. I., Endele, S., Myklebust, L. M., Hoyer, J., Sticht, H., Azzarello-Burri, S., Rauch, A., Arnesen, T., & Reis, A. (2015). De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. *European Journal of Human Genetics*, *23*(5), 602–609. https://doi.org/10.1038/ejhg.2014.150