

Prof. Peter Bauer, MD

Chief Medical & Genomic Officer

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PERSONAL STATEMENT

Prof. Peter Bauer, M.D. is member of the executive board of CENTOGENE GmbH, Rostock (Germany) where he has continuously broadened the portfolio and increased the efficiency in diagnostic processes through implementation of high-throughput Next-Generation Sequencing (NGS). Besides large screening projects, where thousands of samples receive limited genotyping and biomarker profiling, he designed and implemented the diagnostic Whole Genome Sequencing workflow at CENTOGENE becoming the first European diagnostic provider in rare diseases. Since 2017, as Chief Scientific Officer, he focused on bioinformatics integration of genomics, transcriptomics, and peptidomics for rare diseases and somatic cancer analysis. Moreover, he developed a strategy for patient-derived cellular models including inducible pluripotent stem cells (iPSCs), which are used to model rare disease ("study-in-a-dish"), enhance early biomarker identification, and help to validate potential compounds in preclinical settings. More recently, he developed dried-blood spot-based RNA sequencing for research and diagnostic cases and strengthened the Biodatabank with disease insight tools including a new platform approach for identification of modifier genes in rare diseases.

From 2001 to 2015, he has headed Molecular Genetics and the Core Facility for Applied Transcriptomics and Genomics, Institute of Medical Genetics and Applied Genomics, Tübingen. In diagnostics, he has established a broad portfolio of technologies and tests within the Institute focusing on neurodegenerative diseases, genetics of intellectual disability and cardiogenetics. The diagnostic laboratory uses all current molecular technologies including PCR / Sanger sequencing, MLPA, real-time PCR, SNP array diagnostics, and next-generation sequencing making the Institute of Medical Genetics and Applied Genomics a highly visible academic supplier of cutting edge molecular genetic testing. He still holds a teaching and research position at the Institute of Medical Genetics and Applied Genomics in Tübingen. From 2016 to 2021, he was holding a teaching position at the University Hospital in Tübingen, since September 2022, he joined the University Medicine Rostock with teaching in human genetics and clinical services for patients with rare tumor syndromes.

His research focus lies on the genomic analysis and diagnostics of neurodegenerative diseases, where he has authored more than 250 peer-reviewed publications. He is partner in several European networks dealing with the implementation of NGS in clinical practice (EMQN, TECHGENE, ad-hoc commission for genetic testing of the German Society of Human Genetics). Furthermore, he is committed to Next-Generation-Sequencing diagnostics and developing NGS guidelines. He has broad experience in Next-Generation-Sequencing application both in research and diagnostics.

PROFESSIONAL ACTIVITIES

Since 06/2022	Chief Medical & Genomic Officer Centogene GmbH, Rostock
12/2019 - 05/2022	Chief Genomic Officer Centogene GmbH, Rostock
01/2017 - 11/2019	Chief Scientific Officer Centogene, Rostock
01/2016 - 12/2016	Chief Operating Officer Centogene, Rostock
08/2001 - 12/2015	Head of Molecular Genetics Institute of Medical Genetics and Applied Genomics, University of Tübingen
09/1997 - 07/2001	Residency in Neurology Department of Neurology, University of Rostock

SCIENTIFIC CAREER

2010	Habilitation in Human Genetics Eberhard-Karls-University Tübingen
2006	Board Certification in Human Genetics Eberhard-Karls-University Tübingen
1997	Thesis in Medicine Humboldt-University, Berlin
04/1993 - 07/1997	Medicine Free University Berlin
04/1990 - 03/1993	Medicine Albert-Ludwigs-Universität, Freiburg im Breisgau
10/1988 - 03/1990	Fine Arts Federal Academy of Fine Arts, University of Karlsruhe

Other experience and professional memberships

Member, American Society of Human Genetics (ASHG)
 Member, German Society of Human Genetics (GfH)
 Member, European Society of Human Genetics (ESHG)
 President, German Society of Neurogenetics (DGNG)

Publications (*Publications since 2018*)

- Mayer AK, Balousha G, Sharkia R, Mahajnah M, Ayesh S, Schulze M, Buchert R, Zobor D, Azem A, Schöls L, Bauer P, Wissinger B. Unraveling the genetic cause of hereditary ophthalmic disorders in Arab societies from Israel and the Palestinian Authority. *Eur J Hum Genet.* 2020 Jan 2. doi: 10.1038/s41431-019-0566-3. [Epub ahead of print] PubMed PMID: 31896775.
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- Beetz C, Ameziane N, Kdissa A, Karageorgou V, Bauer P, Suleiman J, Sutton VR, El-Hattab AW. VPS26C homozygous nonsense variant in two cousins with neurodevelopmental deficits, growth failure, skeletal abnormalities, and distinctive facial features. *Clin Genet.* 2019 Dec 17. doi: 10.1111/cge.13690. [Epub ahead of print] PubMed PMID: 31845315.
- Rocha ME, Silveira TRD, Sasaki E, Sás DM, Lourenço CM, Kandaswamy KK, Beetz C, Rolfs A, Bauer P, Reardon W, Bertoli-Avella AM. Novel clinical and genetic insight into CXorf56-associated intellectual disability. *Eur J Hum Genet.* 2019 Dec 10. doi: 10.1038/s41431-019-0558-3. [Epub ahead of print] PubMed PMID: 31822863.
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- Al-Kindi A, Al-Shehhi M, Westenberger A, Beetz C, Scott P, Brandau O, Abbasi-Moheb L, Yüksel Z, Bauer P, Rolfs A, Grüning NM. A novel POC1A variant in an alternatively spliced exon causes classic SOFT syndrome: clinical presentation of seven patients. *J Hum Genet.* 2020 Jan;65(2):193-197. doi: 10.1038/s10038-019-0693-2. Epub 2019 Nov 26. PubMed PMID: 31767933.
- Sidransky E, Arkadir D, Bauer P, Dinur T, Lopez G, Rolfs A, Zimran A. Substrate reduction therapy for GBA1-associated Parkinsonism: Are we betting on the wrong mouse? *Mov Disord.* 2019 Nov 11. doi: 10.1002/mds.27903. [Epub ahead of print] PubMed PMID: 31710399.
- Cullufi P, Tabaku M, Beetz C, Tomori S, Velmishi V, Gjokopulli A, Bauer P, Wirth S, Rolfs A. Comprehensive clinical, biochemical and genetic screening reveals four distinct GBA genotypes as underlying variable manifestation of Gaucher disease in a single family. *Mol Genet Metab Rep.* 2019 Oct 26;21:100532. doi: 10.1016/j.ymgmr.2019.100532. eCollection 2019 Dec. PubMed PMID: 31709146; PubMed Central PMCID: PMC6831897.
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- Holger Hengel, Yvonne Schelling, Reinhard Keimer, Werner Deigendesch, Peter Bauer, Ludger Schöls: A previously identified missense mutation in STYXL1 is likely benign. *European Journal of Medical Genetics* 11/2018; DOI:10.1016/j.ejmg.2018.11.016
- Joanne Trinh, Katja Lohmann, Hauke Baumann, Alexander Balck, Max Borsche, Norbert Bruggemann, Leon Dure, Marissa Dean, Jens Volkmann, Sinem Tunc, Jannik Prasuhn, Heike Pawlack, Sophie Imhoff, Christine Lill, Meike Kasten, Peter Bauer, Arndt Rolfs, Christine Klein: Utility and implications of exome sequencing in early-onset Parkinson's disease. *Movement Disorders* 10/2018; DOI:10.1002/mds.27559
- Matthias Baumann, Herbert Schreiber, Beate Schlotter-Weigel, Wolfgang N. Löscher, Rolf Stucka, Daniela Karall, Tim M. Strom, Peter Bauer, Birgit Krabichler, Christine Fauth, Dieter Glaeser, Jan Senderek: MPV17 mutations in juvenile- and adult-onset axonal sensorimotor polyneuropathy. *Clinical Genetics* 10/2018; 95(1)., DOI:10.1111/cge.13462
- Nuria C. Bramswig, Aida M. Bertoli-Avella, Beate Albrecht, Aida I. Al Aqeel, Amal Alhashem, Nouriya Al-Sannaa, Maissa Bah, Katharina Bröhl, Christel Depienne, Nathalie Dorison, Diane Doummar, Nadja Ehmke, Hasnaa M. Elbendary, Svetlana Gorokhova, Delphine Héron, Denise Horn, Kiely James, Boris Keren, Alma Kuechler, Samira Ismail, Mahmoud Y. Issa, Isabelle Marey, Michèle Mayer, Jennifer McEvoy-Venneri, Andre Megarbane, Cyril Mignot, Sarar Mohamed, Caroline Nava, Nicole Philip, Cecile Ravix, Arndt Rolfs, Abdelrahim Abdrabou Sadek, Lara Segebrecht, Valentina Stanley, Camille Trautman, Stephanie Valence, Laurent Villard, Thomas Wieland, Hartmut Engels, Tim M. Strom, Maha S. Zaki, Joseph G. Gleeson, Hermann-Josef Lüdecke, Peter Bauer, Dagmar Wieczorek: Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). *Human Genetics* 08/2018; 137(80)., DOI:10.1007/s00439-018-1929-5
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- Heike Jacobi, Sophie Tezenas du Montcel, Peter Bauer, Paola Giunti, Arron Cook, Robyn Labrum, Michael H. Parkinson, Alexandra Durr, Alexis Brice, Perrine Charles, Cecilia Marelli, Caterina Mariotti, Lorenzo Nanetti, Lidia Sarro, Maria Rakowicz, Anna Sulek, Anna Sobanska, Tanja Schmitz-Hübsch, Ludger Schöls, Holger Hengel, Laszlo Baliko, Bela Melegh, Alessandro Filla, Antonella Antenora, Jon Infante, José Berciano, Bart P. van de Warrenburg, Dagmar Timmann, Sandra Szymanski, Sylvia Boesch, Wolfgang Nachbauer, Jun-Suk Kang, Massimo Pandolfo, Jörg B. Schulz, Audrey Tanguy Melac, Alhassane Diallo, Thomas Klockgether: Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. *Journal of Neurology* 06/2018; 265(10)., DOI:10.1007/s00415-018-8954-0
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- Suzanna G. M. Frints, Aysegul Ozanturk, Germán Rodríguez Criado, Ute Grasshoff, Bas de Hoon, Michael Field, Sylvie Manouvrier-Hanu, Scott E. Hickey, Molka Kammoun, Karen W. Gripp, Claudia Bauer, Christopher Schroeder, Annick Toutain, Theresa Mihalic Mosher, Benjamin J. Kelly, Peter White, Andreas Dufke, Eveline Rentmeester, Sungjin Moon, Daniel C Koboldt, Kees E. P. van Roozendaal, Hao Hu, Stefan A. Haas, Hans-Hilger Ropers, Lucinda Murray, Eric Haan, Marie Shaw, Renee Carroll, Kathryn Friend, Jan Liebelt, Lynne Hobson, Marjan De Rademaeker, Joep Geraedts, Jean-Pierre Fryns, Joris Vermeesch, Martine Raynaud, Olaf Riess, Joost Gribnau, Nicholas Katsanis, Koen Devriendt, Peter Bauer, Jozef Gecz, Christelle Golzio, Cristina Gontan, Vera M. Kalscheuer: Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. *Molecular Psychiatry* 05/2018; DOI:10.1038/s41380-018-0065-x
- I Sepahi, U Faust, M Sturm, K Bosse, M Kehrer, T Heinrich, K Grundman-Hauser, P Bauer, S Ossowski, H Susak, U Bick, E Schröck, D Niederacher, B Auber, C Sutter, N Arnold, E Hahnen, B Dworniczak, S Wang-Gorke, A Gehrig, BHF Weber, C Engel, J Lemke, H Huu Phuc Nguyen, O Riess, C Schroeder: Truncating variants in DNA-repair genes and their effect on AAO of hereditary breast cancer. *Senologie - Zeitschrift für Mammadiagnostik und -therapie* 05/2018; 15(02):e45-e46., DOI:10.1055/s-0038-1651801
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