

**Professor Bernd Wollnik**

**CURRENT POST:** Director, Institute of Human Genetics, University Medical Center Göttingen  
Professor of Medical Genetics

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**EDUCATION AND PROFESSIONAL QUALIFICATIONS**

1996 Dr. med.; Rheinische Friedrich-Wilhelms University Bonn, Germany  
1995 German Medical Exam (Dritter Abschnitt der Ärztlichen Prüfung)  
1990-1992 M.D. thesis studies, Rheinische Friedrich-Wilhelms University Bonn, Germany  
1988-1995 Undergraduate studies in Medicine, Rheinische Friedrich-Wilhelms University Bonn, Germany

**EMPLOYMENT HISTORY**

Since 2015 Professor of Medical Genetics, Director of Institute of Human Genetics, University Medical Center Göttingen, Germany  
2013-2015 Director of Center for Rare Diseases Cologne, University Hospital of Cologne, Germany  
2012-2015 University (Full) Professor for Medical Genetics, Institute of Human Genetics, University Hospital Cologne, Germany  
2004-2012 Junior research group leader, Center for Molecular Medicine Cologne (CMMC), Institute of Human Genetics, University of Cologne, Germany  
1999-2004 "Foreign Lecturer" position, Division of Medical Genetics, Istanbul University, Istanbul, Turkey  
1998-2004 Molecular Genetics Unit, Division of Medical Genetics, Istanbul University, Istanbul, Turkey  
1997-1999 DAAD Fellowship for Young Scientists, Division of Medical Genetics, Istanbul University, Istanbul, Turkey  
1997 Post-Doc Position, Center for Molecular Neurobiology, University Hamburg, Germany  
1995-1997 Practical Training (Arzt im Praktikum), Department of Neurology/Center for Molecular Neurobiology, University Hamburg, Germany

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**LARGE SCALE GRANTS (from 2012)**

2016-2020 **German Research Foundation (DFG)**  
SFB1002 Modulatory Units in Heart Failure; The role of genomic instability in the failing heart  
2015-2018 **E-RARE (German Ministry of Education and Research (BMBF)) EuroMicro: Primary monogenic microcephalies: from genetics to pathophysiology and the clinic**  
2017 **German Centre for Heart Research (DZHK)**  
Unravelling the genetic origin of the hypoplastic left heart syndrome by whole-genome sequencing approaches  
2016 **German State of Niedersachsen**  
Physics-to-Medicine Seed Grant 2016: Mitochondrial Dysfunction in Aging Phenotypes  
2015-2016 **German Research Foundation**  
CECAD - Excellence Cluster on Cellular Stress Response and Aging-Associated Diseases, Cologne University  
2014-2016 **Center for Molecular Medicine Cologne**  
Molecular pathogenesis of Hallermann-Streiff syndrome  
2012-2015 **E-RARE (German Ministry of Education and Research (BMBF))**  
CRANIRARE-2: An integrated clinical and scientific approach for craniofacial malformations  
2012-2015 **German Ministry of Education and Research (BMBF)**  
FACE: Research consortium on selected craniofacial malformation disorders

## TOP 10 PUBLICATIONS

1. Gordon CT, Xue S, Yigit G, ..., Amiel J\*, **Wollnik B\***, Reversade B\*. De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. *Nat Genet.* 2017; 49(2):249-255. (\*co-corresponding)
2. Harley ME, Murina O, Leitch A, ..., **Wollnik B**, Stewart GS, Jackson AP. TRAIIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. *Nat Genet.* 2016; 48(1):36-43.
3. Hatzold J, Beleggia F, Herzig H, Altmüller J, Nürnberg P, Bloch W, **Wollnik B**, Hammerschmidt M. Tumor suppression in basal keratinocytes via dual non-cell-autonomous functions of a Na,K-ATPase beta subunit. *eLife*; 2016 5, e14277.
4. Yigit G, Wieczorek D, Bögershausen N, ..., Thiele H, Nürnberg P, **Wollnik B**. A syndrome of microcephaly, short stature, polysyndactyly, and dental anomalies caused by a homozygous KATNB1 mutation. *Am J Med Genet A.* 2016; 170:728-33.
5. Bögershausen N, Tsai IC, Pohl E, ..., Li Y, Katsanis N, **Wollnik B**. RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. *J Clin Invest.* 2015; 125(9):3585-99.
6. Rosin N, Elcioglu NH, Beleggia F, ..., **Wollnik B**, Yigit G. Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. *Hum Mol Genet.* 2015; 24:3708-17.
7. Murray JE, Bicknell LS, Yigit G, ..., **Wollnik B**, Jackson AP. Extreme growth failure is a common presentation of ligase IV deficiency. *Hum Mutat.* 2014; 35:76-85.
8. Keupp K, Beleggia F, Kayserili H, ..., Kornak U, Marini J, **Wollnik B**. Mutations in WNT1 cause different forms of bone fragility. *Am J Hum Genet.* 2013; 92(4):565-74.
9. Kalay E, Yigit G, Aslan Y, ..., Jackson AP, Karagüzel A, **Wollnik B**. CEP152 is a novel genome-maintenance protein and its disruption causes genomic instability in Seckel syndrome. *Nat Genet.* 2011; 43(1):23-6.
10. Reversade B, Escande-Beillard N, Dimopoulou A, **Wollnik B**, Van Maldergem L, Mundlos S, Kornak U. Mutations in PYCR1 cause cutis laxa with progeroid features. *Nat Genet.* 2009; 41(9):1016-21.

## AWARDS/HONORS/OTHER EXCELLENCE CRITERIA (selected)

Since 2017	Member Else Kröner-Fresenius MD Program, University of Göttingen
Since 2017	Member Else Kröner-Fresenius Clinician Scientist Program, University of Göttingen
Since 2017	Member International Max Planck Research School for Genome Science, University of Göttingen
Since 2017	Board Member, Center for Rare Diseases Göttingen
Since 2015	Member of Georg-August-University School of Science, GAUSS (Molecular Medicine), Göttingen
2007-2015	PI Cluster of Excellence 229; Cellular Stress Response and Aging-Associated Diseases – CECAD, University of Cologne,
2004-2012	Center for Molecular Medicine Cologne (CMMC), junior research group
2002	Young Scientist Award, Turkish Academy of Science, Turkey
2001	International Scientific Research Award of Istanbul University, Turkey
1997-1999	DAAD Research fellowship for young scientists (Istanbul University, Turkey)