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## Curriculum Vitae Professor Dr. Bernhard Horsthemke

**Name:** Bernhard Horsthemke

**Born:** 16 February 1953



**Main areas of research: genetic and epigenetic variation in the development of disease, gene expression differences, genomic imprinting**

Bernhard Horsthemke has made major contributions to the field of human genetics. His group has developed microcloning techniques for the analysis of specific chromosomal regions and identified several disease genes. A major focus of his work at the Institut für Humangenetik in Essen for the past twenty years has been the study of epimutations in human disease.

### Academic and Professional Career

- since 2001 Director, Institute of Human Genetics, University Hospital Essen, Germany
- since 2000 Full Professor of Human Genetics, University Duisburg-Essen, Germany
- 1992 - 2000 Associate Professor of Human Genetics, University Essen, Germany
- 1989 Habilitation (Human Genetics), University Essen, Germany
- 1986 - 1992 Assistant Professor, University Essen, Germany
- 1984 - 1986 Postdoc at St. Mary's Hospital, London, UK
- 1982 PhD, Technical University Berlin, Germany
- 1972 - 1978 Study of Chemistry, Technical University Berlin, Germany

### Project coordination, Membership in collaborative research projects

- since 2009 Coordinator of the BMBF research network "Imprinting diseases"
- since 2008 Member of the BMBF research network "Obesity"
- 2002 - 2008 Coordinator of the DFG priority program "Epigenetics"

1995 - 2001     Coordinator of the DFG priority program "Molecular Dymorphogenesis"

#### **Functions in Scientific Societies and Committees**

since 2012     Member of the DFG review board  
2010 - 2012     Vicepresident, German Society of Human Genetics  
1998 - 2000     Vicepresident, German Society of Human Genetics

#### **Honours and Awarded Memberships**

2012             Max-Delbrück-Lecture of the German Society of Genetics  
2007             Dr. Claudia Benton Award of the Angelman Syndrome Foundation, USA  
2004             Member of the National Academy of Sciences Leopoldina  
2004             Award of the European Society of Human Genetics  
1984             Long-term Fellowship of the European Molecular Biology Organization

#### **Major Scientific Interests**

Bernhard Horsthemke has made major contributions to the field of human genetics. His group has developed microcloning techniques for the analysis of specific chromosomal regions and identified several disease genes. A major focus of his work at the Institut für Humangenetik in Essen for the past twenty years has been the study of epimutations in human disease. His group was the first to demonstrate that tumour suppressor genes cannot only be inactivated by DNA mutations, but also by DNA methylation, and that defects in genomic imprinting lead to recognizable syndromes. His group then pioneered the use of DNA methylation testing in clinical practice. Based on the study of familiar cases of imprinting disorders, he developed the concept of an imprinting centre, which controls the domain-wide establishment and maintenance of genomic imprints.