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# European Society of Human Genetics Deutsche Gesellschaft für Humangenetik

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### **ESHG Award for Bernhard Horsthemke (born 16.2.1953 in Ahlen, Westfalen)**

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Bernhard Horsthemke, PhD  
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Laudatio by Han Brunner

The European Society of Human Genetics will present this year its annual award to professor Bernhard Horsthemke for his many important contributions to the understanding of the molecular basis of human genetic diseases. Dr Horsthemke and his group have uncovered the genetic basis of the tricho-rhino-phalangeal syndrome, and hereditary multiple exostosis, as well as contributing to our understanding of retinoblastoma and uveal melanoma.

The work of Bernhard Horsthemke on microdeletion syndromes has been especially important in elucidating the involvement of imprinting in the Angelman and Prader-Willi syndromes. Dr Horsthemke has published 50 scientific papers on these syndromes alone, reflecting his dedication to understanding them at the molecular level through careful and often elegant molecular genetic experiments. These relatively common mental retardation syndromes specifically occur when the abnormal chromosome 15 is inherited from the father (in Prader Willi syndrome), or from the mother (Angelman syndrome). This curious dependence on the sex of the parent transmitting the disease is termed imprinting and the scientific community owes a great deal to Dr. Horsthemke for his painstaking work in clarifying this unusual genetic mechanism.

Over the past 15 years the work of Dr. Horsthemke and his colleagues has allowed them to develop a rapid and reliable genetic test for the Angelman and Prader-Willi syndromes which is currently being used by many diagnostic labs worldwide today. Of the greatest fundamental importance is a paper from 1995 (Buiting et al. Nature Genetics 9:395-400, 1995) in which the authors describe the existence of an imprinting centre on chromosome 15 that explains this curious and previously unexplained effect of the parental origin of the mutation.

A recent paper by Horsthemke and colleagues (Cox et al. Am J Hum Genet: 71-162-164, 2002) suggested that the risk of imprinting defects may be increased in children born after intra-cytoplasmic sperm injection (ICSI). This hypothesis has also attracted considerable attention and is the focus of a number of ongoing studies worldwide.

In Dr. Horsthemke, the ESHG honours a scientist, whose work proves that the natural qualities of a true scientist are to be of unlimited curiosity and to keep asking new questions even after working for many years on a problem. Dr. Horsthemke is a role model for young European scientists. It is for this combination of outstanding scientific accomplishments and inspiration to others that Dr. Horsthemke receives this year's ESHG award.