

## Jahrestagung 2004

deutsche gesellschaft für humangenetik  
Geschäftsstelle  
Goethestr. 29, 80336 München  
Telefon 0049-89-5502 7855  
Telefax 0049-89-5502 7856  
organisation@gfhev.de  
www.gfhev.de

# European Society of Human Genetics Deutsche Gesellschaft für Humangenetik

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Kontakt:

**GfH-Pressestelle**  
Dipl.-Soz. Christine Scholz  
Goethestr. 29  
80336 München  
Tel 089-5502 7855  
Fax 089-5502 7856  
organisation@gfhev.de

**GSF-Forschungszentrum für Umwelt  
und Gesundheit, Presse- und Öffentlichkeitsarbeit**  
Ingolstädter Landstrasse 1  
85764 Neuherberg  
Tel 089-3187-2460  
Fax 089-3187-3324  
oea@gsf.de

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### **Epigenetics**

In the past few years, the DNA sequence of various species including yeast, plants, worm, fly, mouse and man has been determined. In an ongoing effort, an inventory of variations in the human and mouse DNA sequences is being made. These variations account for most of the heritable diversity of a species, which is the subject of genetics.

Although most cells of an organism contain the same DNA sequence, not all of the genes are used in every cell at every time. At various stages during development, dramatic changes in the pattern of gene expression occur, which then are stably inherited through several cell divisions. This variation of gene expression is the subject of epigenetics, a term which was introduced in 1953 by Conrad Waddington. As these patterns are potentially reversible, they can not be explained by DNA sequence variation. Thus, there must be a code above the DNA sequence level that specifies the selective use of genetic information during development. The epigenetic code is based on the modification of DNA and chromatin, for example by methylation.

Both the DNA sequence and the methylation pattern of a gene can be affected by a mutation. An inappropriate change of the methylation pattern is called an epimutation. In fact, it has been estimated that epimutations occur much more frequently than DNA sequence mutations. Epimutations contribute to many diseases including cancer and mental retardation. There is also growing concern that exogenous effects such as nutrition and assisted reproduction may increase the rate of epimutations.

Epigenetics is a rapidly growing field. At the ESHG meeting, one symposium (S3, Sunday 17:00)) and the ESHG award lecture (by Prof. Bernhard Horsthemke, Essen, Tuesday 13:45) deal with epigenetics.

**For more information please contact:**

Dr. Bernhard Horsthemke, Univ. Essen