

Jahrestagung 2004

European Society of Human Genetics Deutsche Gesellschaft für Humangenetik

München, 12.-15.6.2004

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

Zeichen: 3651

**Prof. Veronica Van Heyningen, Edinburgh
President ESHG**

A continuous theme of this year's conference is the elucidation of the myriad different ways in which genetic changes can cause disease. These diseases are often very rare, but the mechanisms are frequently relevant to much more common diseases. When a machine – the body – is running smoothly it is difficult to understand how the mechanism works. When things go wrong it is relatively easier to dissect how the machine has failed. That is why genetics is such an important tool for understanding biology.

Here we are discussing human genetic mechanisms, but the appreciation of the unity of biology has been one of the major surprises and delights of the genomic era, and some of that is strongly reflected in several talks at this meeting. The detailed, often computer-based, study of genome architecture has given us important insight into how chromosomal abnormalities, often cryptic, can arise and how genomes evolve – as we will hear from **James Lupski (in Plenary talk L08)**. Some of these features cause very small, invisible deletions which can lead to disease. New methods using microarrays are being developed to search for such deletions (**Anita Rauch C39**) in previously unexplained birth defects, including often mental retardation. Similar microarrays are also used for studying gene expression in tumours such as the childhood Wilms tumour (**C43 and C44**) and these analyses can help develop prognostic techniques to optimise tumour treatment.

Now that the genome is more or less fully available, at least as a string of 6 billion letters (3 billion from each parent, a 3 and 9 zeros.), we are in a better position to look for variation that may be associated with predisposition to more complex common diseases, the development of which is influenced by many genes, as well as environmental factors and probably also by chance. Such diseases include asthma and eczema, the frequency of which is unaccountably increasing in our populations. **Bill Cookson (speaker S6, in a symposium on common diseases)** won't mind my saying that he has been working for quite a few years trying, with increasing success, to identify some of the more major genes which function differently in people with asthma. Other ideas that have been around for some time but are now gaining further credence include the concept that fetal nutrition might influence the types of diseases we succumb to in adult life – we hear more about this and possible links to subtle alteration of genomic organisation, from **Randy L. Jirtle (speaker S9 in a symposium on model organisms)**.

The wealth of variation in the human population is an important feature to ensure the survival of our and other species, as conditions change: we hear from **Suzanne Rutherford (S28)** about mechanisms which can help hide and buffer some of this variation normally. However, we have to be ready to deal with variation in our responses to drug treatment and that is a new area that is now being increasingly explored by the pharmaceutical industry for example – this is discussed by **Karl Lindpaintner (S19)**.

Finally, this meeting is actually two over-lapping and intertwined meetings, and you will have noted that we have many sessions which discuss how individuals and societies deal with the impact of all this new knowledge and again variation is one of the recurrent themes: **Segolene Ayme (L04)** will discuss the variation in genetic testing and counselling around Europe. And we have a whole symposium tomorrow afternoon (**Talks S1, S2, S3**) on how people manage when the genetic risk of sudden cardiac death has been identified in their families.