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Frequent variations, small effects

The familial occurrence of very rare diseases is one of the characteristics of human genetics. Mendelian rules apply to the inheritance of such phenotypes and so far they have been described for more than a 1000 different diseases. The identification of mutated genes for such disorders has helped to understand basic principles of disease etiology and has helped to diagnose and in some cases to treat patients. The process of assigning the sequence variation in particular genes to particular disease phenotypes is ongoing and numerous reports at the ESHG meeting in Munich are dealing with the identification of rare mutations in one of the 30000 human gene, most of which have not even got a name so far.

More and more, human geneticists are also dealing with a type of sequence variation that is common and which can be related to common disorders. Frequent variation means that a considerable proportion of individuals in a given population bears a particular sequence which has an effect on disease susceptibility. 20% of a population for instance might have an increased risk for a common disorders such as stroke, while 80% has a relatively reduced risk. Such sequence variations are not called mutations, - this term is reserved for the rare variants - but polymorphisms, meaning frequent sequence variation. An international research consortium is currently out to determine and list the common sequence variation in human populations (HapMap project). Recent studies clearly show that the most of such variation is located in regulatory regions of genes and that the effects exerted by such variation is very small. In most cases, the effects are hardly measurable. Very large numbers of patients with particular phenotypes have to be compared with healthy controls in order to demonstrate these effects. Progress in this field will be reported at the meeting for several so called complex disorders including asthma, cancer and depression.

At the ESHG meeting, one symposium (S2, Sunday 17:00) and several workshops (W3, Sunday, 13:15 and W11, Monday, 13:15) will deal with aspects of common sequence variation in man.

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