

## A unique blend in Bled

The success of the series of biannual Mutation Detection meetings is in part due to the characteristic and perhaps unique combination of biotechnology, genome research and clinical diagnostics. In addition to these features, Mutation Detection 2001 included more presentations on single nucleotide polymorphism (SNP) identification and typing, as well as discussion of fundamental issues concerning their use in studies on common diseases and various types of genetic predispositions.

As stressed by Ed Southern in his keynote address, DNA provides a simple but powerful entry point into a multitude of biological questions. However, only a limited portion of the genome's DNA has been explored. While coding sequences attract much attention, the consequences of non-coding mutations remain to be investigated. New biological insights are expected from studies concerning changes affecting context, methylation and more general levels of gene expression. Methodological platforms must meet the needs of large-scale analyses, ease of automation, flexibility and production of high-quality data directly readable by computers, while maintaining costs within affordable levels.

Scanning methods for unknown mutations should be able to cover a wide range of applications and to satisfy user demands that differ substantially. Several problems remain to be solved in terms of higher throughput, better scalability, increased simplicity and lower costs, as stressed by Richard Cotton in his overview. Methods for known mutations or for SNP typing, reviewed by Ann-Christine Syvanen, are amenable to increased automation and simplicity, thanks to various solid-phase formats and to the availability of various homogeneous assays. The potential of methods not requiring PCR amplification was also emphasized during various discussions.

In the field of clinical diagnostic tests, rapidity may be a factor that could change the attitude of clinicians in several areas. As pointed out by Jean Amos, there is a need for testing rare mutations at lower costs and for kits or arrays for use with moderate sample numbers.



New SNP discovery and SNP typing is faced with the problem of SNP quality and validation. The issue of cut-off frequencies for minor alleles emerged on several occasions during the meeting. Moreover, the main limitations may be not in the technology, but in the quality of study designs and their ability to evaluate the correlations to be tested.

What are the new trends in methodologies? Better multiplexing, ability to obtain reliable data from DNA pools and increased use of methods that do not rely on amplification of template DNA are among the examples that were cited repeatedly during the meeting. Simplified procedures used to enter mutations into databases were also discussed. The progress of the HUGO Mutation Database Initiative was reported and a new programme has been envisaged to facilitate collection of DNA variations from diagnostic laboratories.

The beautiful scenery of the Alps and of the Bled lake will remain closely associated with the meeting in the minds of all participants. However, perhaps the strongest impression throughout the meeting and particularly during the social events was the warm hospitality of our hosts and the encounter with their rich culture and tradition. Damjan Glavac, Metka Ravnik-Glavac and all their local collaborators deserve major credit for the great success of the meeting.

Mutation Detection 2001, the VI International Symposium on Mutations in the Human Genome, was held in Bled, Slovenia 3-7 May 2001 and attracted over 120 participants. The meeting was organized by Damjan Glavac (far right), Metka Ravnik-Glavac (second right), Ann-Christine Syvanen (centre) and Richard G H Cotton (far left), seen here with Ed Southern (second left). This report was contributed by Mario Tosi and Graham Taylor.

The Book of Abstracts is available at <http://www.mutations2001.bled.si>

The next Mutation Detection meeting will take place in July 2003 at the Great Barrier Reef, Australia.