Institute of Molecular Biology

IMB TechTalk

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"Precision diagnostics: high-throughput sequencing and bioinformatics on the way to clinical routine"

07 September 2012, 11:00 (*s.t.*)

Attention! Change of weekday:
Friday instead of Tuesday

Institute of Molecular Biology (IMB)

Johannes Gutenberg University Campus Mainz

All are welcome to attend

Host: Dr. Bernhard Korn, Director CF & Technology, IMB

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Abstract:

Precision diagnostics: high-throughput sequencing and bioinformatics on the way to clinical routine

Driven by unmet diagnostic needs for many human pathologies as well as exploding costs in healthcare systems, novel molecular diagnostic tests are urgently required. High-throughput sequencing enables such diagnostic applications. However, given the steadily increasing amount of data, sophisticated computational tools become essential to promote high-throughput sequencing applications from research to routine testing.

Small non-coding RNA profiling for a variety of human pathologies represents one example where bio-computational analyses are inevitable to develop highly specific tests. To this end, we investigated over 2,000 samples using array based, sequencing and qRT-PCR experimental methods that have been evaluated with statistical learning approaches to discover minimally-invasive tests and to improve our understanding of human pathologies on a molecular level.

As second example of novel diagnostic applications, targeted re-sequencing, exome sequencing and whole genome sequencing of patients DNA is carried out. We applied targeted re-sequencing to a set of several hundred patients suffering from cardiac diseases and developed a fully automated bioinformatics pipeline including parameter optimization to make the high-dimensional data and the clinical value accessible for physicians.

In the near future, the respective genomics approaches together with epigenmoics, transcriptomics, proteomics and finally also imaging applications will require even more sophisticated bioinformatics and systems biology approaches to understand pathogenic process from a molecular, cellular and organ level.