integrated informatics solutions that integrate, store, and analyze clinical and omics data from diverse sources – generated in-house as well as public consortiums. Many researchers and clinicians must rely on bioinformaticians to perform mundane data management tasks in order to validate a simple hypothesis. Oracle Health Sciences Translational Research Center provides a complete and scalable informatics solution, with centralized data storage and analysis across genetic information areas (genomics, transcriptomics, and proteomics), vendor platforms, biological data types, and clinical data sources. Organizations such as Cancer Research UK, Erasmus MC, MD Anderson Cancer Center and UPMC have adopted this solution and are evaluating treatment responses for similar patients in a self-sufficient manner, ultimately shortening the biomarker development cycle and accelerating the adoption of personalized medicine.

http://dx.doi.org/10.1016/j.nhtm.2014.11.011

## Metabolic phenotyping in mouse and man: Mind the differences!

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### Abstract

Metabolic phenotyping comprises the quantification of endogenous metabolites in biofluids, cells, and tissues. It provides insights into normal as well as aberrant metabolic pathways and biological processes, which is important for the understanding of disease phenotypes. It also allows the identification of biological markers, which can serve as early disease indicators and therapeutic markers for the evaluation of treatment effects. As metabolic markers are not species-restricted, the concept of metabolic phenotyping is highly applicable for translational research. Species independence allows the use of established animal and cell culture models for various diseases within a preclinical context. However, differences in the metabolic set-up of study organisms compared to humans needs to be taken into consideration to prevent misleading conclusions from otherwise valid experimental designs. To determine species-related metabolic differences, a targeted metabolomics approach was applied using a mass spectrometry platform for the quantification of a predefined set of endogenous metabolites, i.e. amino acids, biogenic amines, phosphatidylcholines, sphingomyelins, hexoses, steroid hormones and others. Results from this species comparison on the metabolic level will be presented. Overall, the validity of metabolic phenotyping will be demonstrated, despite or even because of species-dependent characteristics. It has the potential to explain why findings in animal models cannot always be directly translated into clinical settings and might, therefore, facilitate the establishment of suitable models of disease.

http://dx.doi.org/10.1016/j.nhtm.2014.11.012

## Bioinformatics challenges in the adoption of next generation sequencing for translational molecular diagnostics

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### Abstract

Next generation sequencing (NGS) technologies are now widely used in medical research. NGS provides an unprecedented opportunity for high throughput analysis of genetic variations warranting their use in molecular diagnostics. However, among other obstacles, their adoption in clinics poses challenges in the provision for accurate and timely data analysis. A bioinformatics workflow for the analysis of the large amount of data from raw reads to final annotated variants suggesting their functional consequences with clinically actionable or reportable sensitivities and specificities is important. We have developed a number of targeted panels using NGS for the diagnosis of a number of haematological abnormalities and cancer. I will present the experience of development of bioinformatics workflows for the routine use of these panels for a busy molecular diagnostic service.

http://dx.doi.org/10.1016/j.nhtm.2014.11.013

## Identification & characterization of tumor cells isolated from body fluids

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### Abstract

The appearance of malignant cells in body fluids like urine, blood or body-cavity fluids are a clear indication for the existence of a tumor and urine or body-cavity fluid cytology are routine diagnostics today. Cytologic examination of the cellular features of fluids is a valuable adjunct to patient diagnosis and the staging and management of tumors. The German-language literature contains the earliest references to the cytology of malignant cells in fluid specimens. Preparation of the specimen has evolved from unstained wet smears to protocols that generally include centrifugation and the generation of stained smears and a cell block. The smears may be alcohol-fixed direct smears, cytospins, or a liquid-based preparation, and they are usually stained with the Papanicolaou method. Additional techniques, such as immunocytochemistry and flow cytometry, provide significant help in this differential diagnosis. Evaluation of microscopic images after Papanicolaou staining eluded digital pathology, an image-based information environment enabled by computer technology that allows for extracting information from a digital slide. With the advent of full-slide scanning digital methods are regarded as promising way to achieve better, faster and cheaper diagnosis, prognosis and prediction of cancer and other important diseases. One important feature are combinations with immunostaining, FISH technology etc., to elude additional information from the specimen. Circulating tumor cells (CTCs) can be found in the bloodstream and are always ready to attach to endothelial cells lining blood vessels and extravasate to enter tissues and organs to form a metastatic site. They show plastic phenotype and a small number of these cells undergo the epithelial-to-mesenchymal (EMT) program. De-differentiation and dissemination from the primary tumor is a basic prerequisite for colonization and growth of distant metastasis. Phenotypic and functional plasticity of cancer cells and the ability to adapt permanently to demanding conditions provide great challenge for identification and characterization of CTC´s from blood. Their clear identification and characterization is, however, also an important prerequisite to obtain valuable information for diagnosis and prognosis by downstream analytical methods. A novel platform for identification and morphological characterization of cancer cells in body fluids by digital methods is presented.

http://dx.doi.org/10.1016/j.nhtm.2014.11.014

## Next-generation tissue microarrays (ngTMA) in translational research

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### Abstract

Over the last two decades, prognostic and predictive biomarker studies in clinical and translational research settings have become