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Individualised Patient Stratification Using Whole Genome Sequencing

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As the cost of sequencing the human genome falls, medical use of whole-genome sequencing in the clinic is rapidly approaching academic medical centres. In order to support the foreseeable flood of genomic information in the clinic an Oracle based database and business intelligence solution to detect genomic variants has been built at Erasmus university Medical Center (ErasmusMC), rotterdam, the Netherlands.

Introducing Innovation in Healthcare

These advances in genomic medicine are only possible due to the equal progress in the information technology sector. The diagnostic opportunities also bring new challenges to medical centres regarding privacy in general and the necessary ICT infrastructure to secure this privacy at the individual patient level.

Clinical genetic testing in adults is at present typically done for a few patients who, as a result of family history or clinical indications, are considered at risk of carrying genetic variations that are linked to a particular disease or disease predisposition. This is going to change and in the near future, when at relative low cost, all variants in coding and non-coding DNA is mapped at once. The vast amount of knowledge that is offered by whole-genome sequencing means that informed consent for this procedure is more complex than that for existing genetic testing.

The ICT Solution

In order to facilitate this genomics project an Oracle Exadata server was installed as the initial component for the ErasmusMC Translational Research Center (TRC) personal cloud architecture. The aim of this next generation sequencing data storage and analysis project is being used as proof of concept to evaluate outsourcing of IT services.

Additionally, the bioinformatics team is using this environment to benchmark the performance of the Exadata technology against an existing standard IT architecture. The project is a success in that the genomics solution started by the team of professor Peter van der Spek at ErasmusMC has attracted interest for adoption by organisations worldwide and now needs to be managed by an external company. To grow this solution to an international standard the support of Oracle is required.

Impact of the Project

As the focus in the clinic shifts from sequential testing of individual genes associated with a particular disease, to mapping all variants within an individual's whole genome to all known information on all diseases and traits, the consequence is the need for massive parallel processing and high volume data storage.

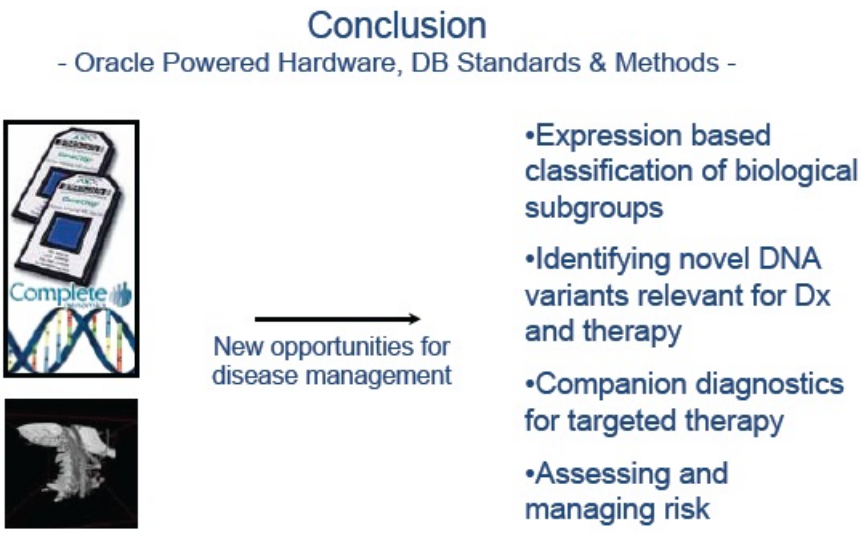
The added value to the clinic is for higher efficiency in diagnosis, with decreased time to delivery and a view of the whole genome not just the individual genes. Increased knowledge can result in medical or lifestyle changes that reduce risks, or it can affect the patient's life decisions or

strategies for coping. Risks of genetic testing also centre on the accuracy of the knowledge that patients (or others) take away from the tests and how that knowledge is used. Over time our knowledge will rapidly expand and new ways to re-annotate the genome data will be developed. Historical data for both the clinician and patient, which has an updated diagnosis should be made available via a secure web based application.

Genome sequencing will have its major impact in three areas listed below:

- Genetic testing of inherited conditions;
- Cancer diagnostics; and
- Pharmacogenetics.

Every individual will learn that he or she is a heterozygous carrier of more than one serious or lethal autosomal recessive disease. This information might affect a patient's lifestyle decisions, and have implications for existing children or other relatives.



Education

This technological solution offers a platform for training medical professionals (physician scientists) to deal with large volumes of sensitive patient related data and learn to diagnose clinical relevant variants within the genome. These actionable items will help reducing costs by providing the right drug to the right patient in therapy. To make informed decisions about whole-genome sequencing, patients will need to have the opportunity to ask questions and get accurate answers from knowledgeable and trained professionals.

The Erasmus MC team is proud to have won the 2nd place at the IT @ Networking Awards 2012.

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