



ERA-Instruments WP 4
Interaction with scientists

Task 4.3
User meetings on cutting edge techniques

Deliverable 4.6
Report on user meeting Tartu

Task leader
Estonian Ministry

Month 14 Year 2



Deliverable 4.6

User meetings on cutting edge techniques report

Month 14 Year 2

1	<i>Executive summary</i>	3
2	<i>Introduction</i>	3
3	<i>About the deliverable and the work package/task</i>	3
3.1	Objective	3
3.2	Approach	4
3.3	Results	5
4	<i>Conclusion</i>	6
	<i>Appendices</i>	7

Authors

Madis Saluveer, Archimedes Foundation, Estonia

1 Executive summary

To promote and ensure the proper operation of newly funded (or to be funded) instrumentation, user workshops with participation of scientists, manufacturers and administrators will enable to identify unpredicted technological problems, will facilitate the exchange of experience and expertise gained with the new instrumentation and this way will promote successful operation of the facilities.

The aim of the user workshop I was to discuss the infrastructural, organizational and funding requirements for establishing and developing a sequencing centre. The specific situation concerning **molecular analysis technologies** in Estonia was highlighted as a sample case.

As a result of the discussion, the following main issues were indicated:

- 1) When setting up a sequencing centre, obtaining two sequencing machines would be beneficial to guarantee uninterrupted sequencing.
- 2) In case of very large scale projects, it would be advisable to run them in the centres and to equip smaller laboratories with a less expensive machine for rapid analysis on a smaller scale.
- 3) Bioinformatics is currently a limiting factor. The software used is not standard and user-friendly; therefore bioinformatics specialists need to find solutions for specific (tailor-made) applications.

2 Introduction

Most important for all activities, both in the national and in the European context, are the needs and views of the scientific community (WP 4 of the project). All measures proposed in ERA Instruments aim at improving the availability of frontier instrumentation for as many scientists as possible as well as identifying the means and ways to achieve these goals (within the limits of the available national budgets). The activities should not only be embedded in the European context but also use input from the international context.

The ninth international Gene Forum with the sub-topic of "New Frontiers in Genomics", jointly organised by the Estonian Genome Foundation and the Estonian Biocentre, on June 12-13 2009 looked into the future of genetic research and paid a lot of attention to the new trend of gene maps identifying personal genetic predispositions. The conference which offered presentations by 18 renowned professionals, the world's leading gene and biotechnology specialists, attracted 210 researchers, doctors, students, health care officials, executives and investors, incl. a seventh from other Baltic states as well as from Scandinavia, Western Europe, and North and Central America.

Prof Andres Metspalu, head of the programme committee of the conference, Director of the Estonian Genome Project of the University of Tartu, expressed a belief that in five to six years there will be a database which doctors can use for assessment of the genetic risks of their patients.

3 About the deliverable and the work package/task

Task 4.3: User meetings on cutting edge techniques

Task Leader: Estonian Ministry (partner #8)

The task "4.3 User meetings on cutting edge techniques" with participation of scientists, manufacturers and administrators aims to promote and ensure the proper operation of newly funded instrumentation in order to identify technological problems, to facilitate the exchange of experience and expertise gained with the new instrumentation and, thus, to promote successful launch and operation of the facilities. The international character of these user meetings will enable to bring together many more experts than national meetings and in this way experience and expertise can be exchanged and pooled to a much larger degree than in case of smaller user meetings.

3.1 Objective

The objective of the task is the formation of discussion groups within user communities dedicated to in-depth discussion of newly funded or emerging instrumentation and technologies.

3.2 Approach

User meeting on molecular analysis technologies

Friday, June 12, 2009

Tartu, Estonia

The international Gene Forum with its focus on genetics and second generation sequencing provided an ideal environment for discussing issues arising around the very rapidly developing technologies of high throughput sequencing. These recent and current developments in sequencing technology are likely to have a major impact on a broad area of biological research.

The user meeting was linked to the conference „GeneForum 2009. New Frontiers in Genomics“with sessions on longevity, human genomics, genomics technologies and applications and genetics of complex traits. <http://www.geneforum.ee/GF2009/index.php>

The user meeting was organized in the form of panel discussion chaired by Prof Ants Kurg, Department of Biotechnology, University of Tartu, and Johannes Janssen, Deutsche Forschungsgemeinschaft, with the active participation and contribution of the participants.

The panellists were:

Prof Jun Wang, Beijing Genomics Institute at Shenzhen, China (JW)

Prof Michael S. Phillips, Genome Quebec; Universite de Montreal, Canada (MP)

Prof Ann-Christine Syvänen, Uppsala University, Sweden (AS)

Prof Ulf Landegren, Uppsala University, Sweden (UL)

Participants: All scientists participating in the user meeting were also present at the conference.

Overall objective

The aim of the discussion was to discuss the infrastructural, organizational and funding requirements for establishing and developing a sequencing centre. The specific situation in Estonia was discussed as a sample case.

The results of the discussion are bundled according to topics with a potential starter question / response at the beginning (boxed, response in bold).

Question 1.

There are three types of second generation sequencing machines currently on the market:

- SOLID 3 from Applied BioSystems Inc., (Foster City, CA, USA)
- Illumina Genome Analyzer II from Illumina Inc., (San Diego, CA, USA) utilizing Solexa technology, and
- Roche 454 GS FLX from 454 Life Sciences Roche (Branford, CT, USA).

In case there is a plan to establish a sequencing center and to purchase the relevant equipment now, which of these three options would be preferable?

Discussion

The large sequencing facilities are using mostly Illumina Genome Analyzer.

Ann-Christine Syvänen explains that 454 GS FLX has so far longer runs and might have applications for specific projects.

According to Jun Wang, SOLID has its benefits for expression analysis.

Ann-Christine Syvänen recommends for the Estonian Sequencing Centre to have two Solexa machines. Two machines are beneficial as the sequencing can run when one of the machines needs repairs or is being serviced. In the case the equipment is of the same type the operating and servicing would be easier.

Postscript: As a result of a call for new instrumentation 2009, the purchase of 2 Illumina Genome Analyzer Ix machines will be supported from the European Structural Funds in order to set up the sequencing centre at the University of Tartu, Estonia.

Question 2.

Do we need to concentrate the sequencing into large centers or spread the machines into laboratories?

Discussion

Ann-Christine Syvänen and Jun Wang expect that projects of very large scale will be run in the centres and every laboratory will have a machine for rapid analysis on a smaller scale. It is expected to look like computing nowadays with both PCs and supercomputing centres operating.

Ulf Landegren points out that it takes considerable effort for developing a new technology into a user friendly machine. It is therefore expected that the new generations of machines start in the large centres where there is more technology-related competence.

Question 3.

Which are the additional costs related to sequencing?

Discussion

Michael S. Phillips points out that bioinformatics is currently a limiting factor. The software is not standard and user-friendly; therefore a bioinformatics specialist needs to find solutions for particular applications. Considering the need for custom-made solutions, developing open source software is a trend.

Ann-Christine Syvänen points out that different applications have different requirements and costs for sample preparation.

Jun Wang provides an estimate for the distribution of work effort in their facility: 50 people for sample preparation, 10 workers for operating the sequencing machines, 100-150 people for bioinformatics. Michael S. Phillips expects that in the future wet-lab costs will decrease and bioinformatics will have a bigger share in the cost structure.

Question 4.

What are the future trends in the organizational structure of sequencing facilities?

Discussion

Both Jun Wang and Ulf Landegren expressed the opinion that commercial service centres will develop both nationally and internationally, providing customers second generation sequencing for a reasonable price.

3.3 Results

The user meeting outcomes (bearing in mind also the overall constraints characteristic of a small country and limited user community) can be summarised as follows:

- 1) In setting up a sequencing centre obtaining two sequencing machines would be beneficial to guarantee uninterrupted sequencing when one of the machines needs repairs or is being serviced.
- 2) In case of very large scale projects, it would be advisable to run them in the centres and funding permitting, to supply every laboratory with a less expensive machine for rapid analysis on a smaller scale.
- 3) It was a general conclusion that bioinformatics is currently a limiting factor. The software used is not standard and user-friendly; therefore bioinformatics specialists need to find solutions for specific applications. It was also pointed out that considering the need for custom-made solutions, developing open source software is a trend.

4 Conclusion

The ERA-Net user meeting was linked to the conference „GeneForum 2009. New Frontiers in Genomics“ which provided the user meeting participants the opportunity to update their knowledge on current issues in human genomics, genomics technologies and applications as well as genetics of complex traits. Secondly, taking Estonia as a sample case, the panellists could focus more on the needs and limits of setting up a large-scale sequencing facility in a country. Third, it also provided a chance for the participants to discuss the issues related to organising co-operation between different institutions in a small user community, to discuss problems of access and organising everyday operations of such a centre.

Appendices

Appendix 1

GENE FORUM 2009 Programme

Friday, 12. June 2009	
08:30-09:00	REGISTRATION AT THE DORPAT CONFERENCE CENTRE, LOCATED AT SOOLA 6
09:00-10:20	<p>OPENING SESSION <i>Chaired by Prof Andres Metspalu, Estonian Genome Project, University of Tartu, Estonia.</i></p> <p>Prof Alar Karis, Rector of the University of Tartu, Estonia <i>"Opening of the Conference"</i></p> <p>Keynote Prof Jun Wang, Beijing Genomics Institute at Shenzhen, China <i>"Sequencing, Sequencing, Sequencing"</i></p> <p>Prof Gerardo Jiménez-Sánchez, National Institute of Genomic Medicine, Mexico <i>"Genomic Medicine in Mexico"</i></p>
10:20-10:40	<i>Coffee/tea break</i>
10:40-12:30	<p>SESSION I - LONGEVITY <i>Chaired by Dr Jaanus Pikaní, Estonian Biotechnology Association, Estonia.</i></p> <p>Prof Eline Slagboom, University of Leiden, The Netherlands <i>"Longevity genetics"</i></p> <p>Prof Gil Atzmon, Albert Einstein College of Medicine, New York, USA <i>"Longevity Genes: past, present and future"</i></p> <p>Prof Jan Vijg, Albert Einstein College of Medicine, New York, USA <i>"Genome Instability, Aging and Longevity"</i></p>
12:30-13:50	<i>Lunch (served at the conference centre)</i>
13:50-15:00	<p>SESSION II - HUMAN GENOMICS I <i>Chaired by Prof Ants Kurg, Department of Biotechnology, University of Tartu, Estonia.</i></p> <p>Prof Donald Conrad, The Wellcome Trust Sanger Institute, United Kingdom <i>"Population genetic insights into the creation and functional impact of human copy number variation."</i></p> <p>Prof Sean Ennis, University College Dublin, Ireland & National Centre for Medical Genetics, Dublin, Ireland <i>"The Autism Genome Project: The Genetics of Autism Spectrum Disorder."</i></p>
15:00-16:50	<p>SESSION III - GENOMICS TECHNOLOGY AND APPLICATIONS <i>Chaired by Prof Ants Kurg, Department of Biotechnology, University of Tartu, Estonia.</i></p> <p>Prof Michael S. Phillips, Genome Quebec; Université de Montréal, Canada <i>"Pharmacogenomics: Paving the Way to Improved Drug Development and Healthcare"</i></p> <p>Prof Ann-Christine Svvänen, Uppsala University, Sweden</p>

"Allele-specific regulation of gene expression by DNA-methylation"

Prof Ulf Landegren, Uppsala University, Sweden

"Molecular tools for clinical interactomics"

16:50-17:15 *Coffee/tea break*

17:15-19:00 **SESSION IV - USER MEETING ON MOLECULAR ANALYSIS TECHNOLOGIES**
Chaired by Prof Ants Kurg, Department of Biotechnology, University of Tartu, Estonia.

Saturday, 13. June 2009

09:00-10:20 **SESSION V - HUMAN GENETICS**
Chaired by Prof Toomas Veidebaum, National Institute for Health Development, Estonia.

Prof Dorret Boomsma, VU University Amsterdam, The Netherlands

"Analysis of Complex Traits"

Prof Jüri Allik, University of Tartu, Estonia

"Psychology and Genetics"

10:20-10:40 *Coffee/tea break*

10:40-12:40 **SESSION VI - GENETICS OF COMPLEX TRAITS**
Chaired by Prof Pärt Peterson, Laboratory of Molecular Pathology, University of Tartu, Estonia.

Prof Markus Nöthen, University of Bonn, Germany

"Genetics of bipolar affective disorder"

Prof Ben Oostra, Erasmus University of Rotterdam, The Netherlands

"Genetic studies in isolated populations"

Dr Gerli Pielberg, Uppsala University, Sweden

"Domestic animals: a resource for identifying genes behind diseases"

12:40-14:00 *Lunch (served at the conference centre)*

14:00-16:00 **SESSION VII - HUMAN GENOMICS II**
Chaired by Prof Pärt Peterson, Laboratory of Molecular Pathology, University of Tartu, Estonia.

Dr. Brian Naughton, 23andMe Inc., USA

"Genetics gets personal: how to read your genome, and what it means for your health."

Prof Andres Metspalu, University of Tartu, Estonia

"The Estonian Biobank and studies on genetic structure of Europeans"

Prof Manfred Kayser, Erasmus University of Rotterdam, the Netherlands

"Forensic genomics: from bio-geographic ancestry testing to visible trait prediction"

16:00 **CLOSING OF THE CONFERENCE**

Appendix 2

List of user meeting participants

Mari	Nelis	Estonian Biocentre
Alar	Aints	Competence Centre for Cancer Research
Helene	Alavere	Estonian Genome Project, University of Tartu
Annely	Allik	Estonian Genome Project, University of Tartu
Wally	Anderson	Competence Centre for Cancer Research
Tarmo	Annilo	University of Tartu
Svetlana	Bankovska	BIOSAN LTD
Vasily	Bankovsky	BIOSAN LTD
Chandana	Basu Mallick	Estonian Biocentre
Marie-Denise	Breton	Centre National de la Recherche Scientifique
Gyaneshwer	Chaubey	Estonian Biocentre
Benoit	Dardelet	Centre National de la Recherche Scientifique
Elina	Floria	University of Tartu
Thomas	Gübitz	German Science Foundation (DFG)
Margot	Hein	University of Tartu
Jonas	Hälldin	Estonian Genome Project, University of Tartu
Johannes	Janssen	German Science Foundation (DFG)
Nina	Jeran	Estonian Biocentre
Mari	Järve	Estonian Biocentre
Aidula-Taie	Kaasik	Estonian Genome Project, University of Tartu
Aili	Kallastu	Competence Centre for Cancer Research
Kaisa	Kamarik	University of Tartu
Alar	Karis	University of Tartu
Aime	Keis	Estonian Genome Project, University of Tartu
Reeli-Kadri	Kullamaa	University of Tartu
Ants	Kurg	University of Tartu
Kadri	Lilienthal	Estonian Genome Project, University of Tartu
Rutt	Lilleoja	Tallinn University of Technology
Eduard	Malinov	University of Tartu
Andres	Metspalu	University of Tartu
Reet	Mändar	University of Tartu
Katrin	Männik	University of Tartu
Tiit	Nikopensius	University of Tartu
Eha	Nurk	National Institute for Health Development
Elin	Org	University of Tartu
Viiu	Paalme	Competence Centre for Cancer Research
Jüri	Parik	University of Tartu
Sven	Parkel	University of Tartu
Pille	Pata	Competence Centre for Cancer Research
Erwan	Pennarun	Estonian Biocentre
Pärt	Peterson	University of Tartu
Jaanus	Pikani	Estonian Genome Foundation
Janne	Pullat	Estonian Genome Project, University of Tartu
Kristina	Raud	Competence Centre for Cancer Research
Anu	Reigo	Estonian Genome Project, University of Tartu
Mari-Liis	Reim	University of Tartu
Maido	Remm	University of Tartu
Maigo	Riener	University of Tartu
Christian	Rolando	Centre National de la Recherche Scientifique
Hovhannes	Sahakyan	Estonian Biocentre

Andres	Salumets	University of Tartu
Rita	Shersta	BIOSAN LTD
Jüri	Siigur	National Institute of Chemical Physics and Biophysics
Tiina	Talvik	Tartu University Hospital
Mari-Liis	Tammesoo	Estonian Genome Project, University of Tartu
Maris	Teder-Laving	Estonian Biocentre
Tanel	Tenson	University of Tartu
Reigo	Timm	University of Tartu
Tõnis	Timmusk	Tallinn University of Technology
Neeme	Tõnisson	University of Tartu
Andres	Valkna	Competence Centre for Cancer Research
Toomas	Veidebaum	National Institute for Health Development
Hanna	Vihma	Tallinn University of Technology
Richard	Villems	University of Tartu
Tõnu	Vooder	Tartu University Hospital
Maris	Väli-Täht	Estonian Genome Foundation
Kaido	Värbu	GlaxoSmithKline Eesti OÜ
Tiit	Örd	University of Tartu
Tõnis	Örd	Estonian Biocentre