The proposal of the Estonian Genome Center of University of Tartu to organize the workshop "Biobanking, New Technologies, Complex Genetic Traits and Personalized Medicine", August 27-29, in Tartu, Estonia.

I Abstract and Scientific Summary

Abstract

The 3rd Young Investigator workshop **"Biobanking, New Technologies, Complex Genetic Traits and Personalized Medicine"** will be held in Tartu, Estonia, August 27-29, 2012. Specific topics to be covered include latest development in biobanking, new sequencing and analysis methods, GWAS, CNV, exome and full genome analysis and potential applications for personalized medicine.

Scientific summary

The 3rd Young Investigator workshop "Biobanking, New Technologies, Complex Genetic Traits and Personalized Medicine" will be held in Tartu, Estonia, August 27-29, 2012.

The overall aim of this workshop is to bring together young investigators (PhD students, postdocs and young researchers) in the framework of various disciplines and outstanding faculty, which would inspire them to go beyond their traditional research and cross the borders of their own disciplines. Such interdisciplinary approach draws together laboratories working mainly in the field of complex genetics, pharmacogenetics and using different technologies in their efforts to explain the "phenomena of complex genetics".

The participating young investigators come from both Estonia and abroad (45 people in past 2 years), in addition to 15-16 speakers from Europe and US.
Each participant should present a poster and give a short overview of their work (1-2 min). The speakers will have 25 minutes to explain the principles and prospective of their study. Each slot in the program will be moderated by a different presenter of that day. This gives the participants an opportunity to get a more detailed insight into the day's subject.
We will use the new conference hall of the Estonian Biocentre, which seats 220 people. This is a brand new facility, where we have the Estonian Genome Center together with the Estonian Biobank. In addition to the main lecture hall, we can use the large atrium for the tea/coffee breaks.

The ESF support (4500 EUR) will be used for covering travel expenses of the speakers only. The rest of the workshop budget will be covered by the Estonian Genome Center of the University of Tartu. The workshop budget incorporates travel and accommodation expenses of all speakers (up to 16 speakers from Europe, US and Canada), catering expenses (two dinners, lunches and coffee breaks) of the speakers and participants, rental of the conference hall and audio-visual equipment, in-country transportation costs of the speakers (transfer from the airport to Tartu and back). The estimated workshop budget totals to 22 000 EUR.

II Programme

The 3rd OpenGENE Young Investigator Workshop in The Baltic Region
"Biobanking, New Technologies, Complex Genetic Traits and Personalized Medicine"
August 27-29, 2012, Tartu, Estonia

26.08.12, Sunday
20.30 - 22.00 Welcome dinner

27.08.12, Monday

15.00 - 15.30 Coffee/tea break
15.30 - 16.30 Lecture
16.30 - 17.30 Lecture
17.30 - 18.00 Lecture
18.30 - 19.00 Lecture
19.00 - 20.00 Buss leaves from Tartu, Riia 23 to Otepää
20.00 – 22.00 Dinner at Lounge "Pühajärve Restaurant" (GMP Club)

28. 08.12, Tuesday

10.00 - 10.30 Coffee/tea break
10.30 - 11.00 Lecture
11.00 - 11.30 Lecture
11.30 – 12.00 Lecture
12.30 - 13.00 Lecture
13.05 - 14.00 Lunch
14.00 – 14.30 Lecture
14.30 – 15.00 Lecture
15.00 – 15.30 Lecture
15.30 – 16.00 Lecture
16.00 – 16.15 Coffee/tea break
16.15 - 17.30 Discussions
18.00 - 20.00 City tour in Tartu and Dinner

29.08.12, Wednesday

09.00 – 09.30 Coffee/tea break
09.30 – 10.00 Lecture
10.00 – 10.30 Lecture
10.30 – 11.00 Lecture
11.00 - 11.30 Lecture
11.30 - 13.00 Discussions
13.00 - 13.30 Conclusions
13.30 - 14.15 Lunch at Riia 23 building
14.30 Buss leaves from Tartu (Riia 23) to the Tartu and Tallinn Airport

* Main lectures are 25 min., rest of the time will be used for questions and discussion.

III Provisional list of speakers

Prof. Thomas Meitinger	Inst. of Human Genetics, Helmoltz Zentrum, München
Prof. Elio Riboli	Imperial College London
Prof. Bartha Maria Knoppers	Faculty of Medicine, Department of Human Genetics, McGill University
Prof. Ivo Gut	Spanish National Genome Analysis Center, Barcelona
Prof. Gert-Jan van Ommen	Leiden Center of Medical Genetics (chairman)
Prof. Eric Schadt	Mount Sinai School of Medicine, Institute for Genomics and Multiscale Biology, New York

Prof. Aarno Palotie	Wellcome Trust, Sanger Institute
Dr. Pauline C. Ng	Singapore Genome Institute
Prof. Harold Snieder	University of Groningen, The Netherlands
Dr. Andrew Morris	Oxford University, UK
Prof. Hans Lehrach	MPI Molecular Genetics, Berlin, FRG
Prof. Michael Snyder	Department of Genetics, Stanford University School of Medicine
Prof. Peter Vischer	Queensland Institute of Medical Research, 300 Herston
Prof. Greg Gibson	School of Biology and Center for Integrative Genomics, Atlanta, Georgia, USA.
Prof. David J. Hunter	Dean for Academic Affairs Harvard School of Public Health
Prof. Mark Gerstein	Department of Computer Science, Yale University
Prof. Muin J. Khoury	Director, Office of Public Health Genomics, CDC, Atlanta
Prof. Geoffrey S. Ginsburg	Duke Center for Personalized Medicine, North Carolina, USA
Prof. Stylianos Antonarakis	University Geneva Medical School
Prof. Jacques Beckman	University of Lausanne

IV Curriculum Vitae of the Scientific Organizer

Andres Metspalu, MD, PhD
Estonian Genome Center, University of Tartu
Tartu, Estonia
Tel +372 737 5066
www.geenivaramu.ee
andres.metspalu@ut.ee

PERSONAL
Born: March 11, 1951 in Estonia
Nationality: Estonian
Status: married, 4 children
Languages: Estonian, English, and Russian

EDUCATION
1979 Ph.D. in Molecular Biology, Structure and function of the eukaryotic ribosome. Institute of Molecular Genetics, Ukrainian Acad. of Sciences, Kiev, USSR. Advisor: Prof. Artur Lind.
1976 MD, University of Tartu (physician), Tartu, USSR.

CURRENT POSITION
2007 - … Director of Estonian Genome Center at University of Tartu, Tartu, Estonia

1992 - … Professor of Biotechnology at University of Tartu, Estonia

PAST POSITIONS

2000 - 2007 Founder and member of the managing board of the Estonian Genome Center, Tartu, Estonia
2000 WHO International Agency for Research of Cancer (IARC), Lyon, France, The Visiting Scientist Award for 12 months
1996 - 2007 Second appointment as Head of Molecular Diagnostics Centre at Tartu University Clinics, Tartu, Estonia
1993 - 1994 Sabbatical leave, visiting professor at Baylor College of Medicine, Dept. of Molecular and Human Genetics, Prof. C.T. Caskey lab, Houston, USA.
1986 - 1992 Research Director of the Estonian Biocentre, Tartu, Estonia
1985 - 1992 Head of Laboratory of Gene Expression, University of Tartu, Estonia
1982 - 1985 Senior scientists at the Laboratory of Molecular Biology, University of Tartu, USSR
1982 IREX fellow at Yale University, Prof. Joan Steitz laboratory, New Haven, USA.
1981 - 1982 IREX fellow at Columbia University, Prof. Alex Tzagaloff laboratory, New York, USA
1976 – 1982 specialist and junior scientist at University of Tartu. Estonia, USSR

RESEARCH PROJECTS

1. Opening Estonian Genome Project for European Research Area, FP7 – REGPOT – Research Potential, coordination and support action, SP4-Capacities Project 245536, Period: 01.12.2009 – 30.11.2012.
2. FP-7 collaborative project ENGAGE, 2008-2012, Univ. of Tartu EGC, (country PI)
3. Translational Genomics, University of Tartu, Develop. Fund grant nr: P1GV ARENG 2011-2015, (PI A. Metspalu, incl. 8 research groups), for 2012.
4. Estonian Ministry of Education and Research, grant NO SF0180142Cs08 (PI A. Metspalu), 2009-2013.
5. Centre of Excellence in Genomics grant for the EGC Univ. of Tartu.
6. Infrastructure for the Estonian Biobank (non-research activities) from the Estonian Government.
7. Estonian Research Infrastructure Roadmap, EGC UT grant (PI A. Metspalu), 2011-2015

RESEARCH INTERESTS

10-Year Track-record: I was over 40y old when Estonia became again an independent country and from 1993 normal system was in place how to do science: freedom to do science, competitive grant system based on peer review, free international communications and travel etc. Of course we did not have real funding, no patent laws etc. I was educated as M.D. and did my PhD on eucaryotic ribosome structure – 5S, 5.8S and tRNA interaction with ribosomal proteins. But when Human Genome Program was on the way I moved to human genetics and as a first thing thought about the new mutation analysis method which would be parallel and did not use gels. Result was Arrayed Primer Extension and 4-color CCD based detector «Genorama» Caskey, T.C., J.M. Shumaker and A. Metspalu. Parallel Primer Extension Approach to Nucleic Acid Sequence Analysis. US Patent No. PCT/US94/07086. In 1993-1994 I was at Baylor College of Medicine, Houston Texas, but decided to return to Estonia and build up human genetics/genomics here. Back at Univ. of Tartu, I developed APEX into real mutation analysis platform, published many papers and finally we had the diagnostic product commercialized by Asper Biotech Ltd. But then I was more interested in complex disese genetics and in a small country it twas very hard to get enough patients for any meaningful study. Therefore I decided to develop a lage population based biobank where we have enough people with medical records and biological material for many studies. It was hard to fund (€15M), it took 8 years to get 5% of the Estonian population (nearly 52 000 gene donors) into it, but today we have one of the best population based biobank in Europe. But as it turned finally out, it is still small and all biobanks have to collaborate. Therefore we have these huge meta-analysis papers. But we have done all by ourselves – samples, genotyping, analysis but it is hard to get first or last positions. For us it will still take time. Now, I hope to do science last ten years and would like to describe human genetic heterogeneity at the single cell level in all cells and phenotypes we can get from the patients and non-patients. We have studied the heterogeneity at the population level and individual level, but still do not have the answers, why it is so difficult to predict and diagnose the disease and treat the patients. Single cell omics analysis should move our knowledge far beyond the current state of the art. This will led us to the personalized medicine, finally.

PUBLICATIONS:

Author and co-author of more than 135 peer-reviewed articles, 2 reviews and 4 book chapters that have been cited over 3000 times. H-factor = 26. Inventor of 8 pending patent applications or patents. Speaker at more than 100 national and international conferences (including at the European Society of Human Genetics Conference in Paris in 1994, in Berlin in 1995, in Lisbon in 1998, Birmingham 2003; at International Congress of Human Genetics in Vienna in 2001 and Brisbaine 2006; at the HUGO Human Genome Meeting in Heidelberg in 1996, in Torino in 1998; at EMBO-EMBL meeting in Heidelberg in 2001; at the American Society of Human Genome Meeting in Honolulu, in 2009). Organiser of many international scientific meetings and workshops, in particular the annual conference of the European Society of Human Genetics as a member of the scientific program committee for 5 years, HUGO meeting in Helsinki 2006, Gene Forum in Tartu, Estonia (since 2000).

List of the top 10 publications:

1. Lango Allen … **Andres Metspalu,** … Hirschhorn JN., Hundreds of variants clustered ingenomic loci and biological pathways affect human height. **Nature,** 2010 Oct 14;467(7317):832-8.

2. Speliotes EK,..**Metspalu A**… Loos RJ., Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. **Nat Genet.** 2010 Nov;42(11):937-48.

3. Jacquemont S,…**Metspalu A**,… Froguel P. Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. **Nature**. 2011 Aug 31;478(7367):97-102.

4. Walters RG, …**Metspalu A**, …Beckmann JS. A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. **Nature. 2010** Feb 4;463(7281):671-5.

5. Mari Nelis, Tõnu Esko, **Andres Metspalu.** Genetic Structure of Europeans: a view from the North-East. ***PlosOne* 2009**, http://dx.plos.org/10.1371/journal.pone.0005472.

6. Kaarel Krjutškov, Reidar Andreson, Reedik Mägi, Tiit Nikopensius, Andrey Khrunin, Evelin Mihailov, Veronika Tammekivi, Helena Sork, Maido Remm and **Andres Metspalu**. Development of a single tube 640-plex genotyping method for detection of nucleic acid variations on microarrays. ***Nucleic Acids Res*. 2008** Jul;36(12):e75.
7. Montpetit A, Nelis M, Laflamme P, Magi R, Ke X, Remm M, Cardon L, Hudson TJ, **Metspalu A.** An evaluation of the performance of tag SNPs derived from HapMap in a Caucasian population. **PLoS Genet. 2006** Mar;2(3):e27.
8. Mueller JC, Lõhmussaar E, Mägi R, Remm M, Bettecken T, Lichtner P, Biskup S, Illig T, Pfeufer A, Luedemann J, Schreiber S, Pramstaller P, Pichler I, Romeo G, Gaddi A, Testa A, Wichmann HE, **Metspalu A**, Meitinger T. Linkage disequilibrium patterns and tagSNP transferability among European populations. **Am J Hum Genet. 2005** Mar;76(3):387-98.
9. Dawson E, Abecasis GR, Bumpstead S, Chen Y, Hunt S, Beare DM, Pabial J, Dibling T, Tinsley E, Kirby S, Carter D, Papaspyridonos M, Livingstone S, Ganske R, Lõhmussaar E, Zernant J, Tõnisson N, Remm M, Mägi R, Puurand T, Vilo J, Kurg A, Rice K, Deloukas P, Mott R, **Metspalu A**, Bentley DR, Cardon LR, Dunham I. A first-generation linkage disequilibrium map of human chromosome 22. **Nature. 2002** Aug 1;418(6897):544-8.
10. Tõnisson N, Zernant J, Kurg A, Pavel H, Slavin G, Roomere H, Meiel A, Hainaut P, **Metspalu A**. Evaluating the arrayed primer extension resequencing assay of TP53 tumor suppressor gene. **Proc Natl Acad Sci U S A. 2002**, 16;99(8):5503-8.

GRANTED PATENTS (in the last ten years)

Jevgeni Berik, Ants Kurg, Andres Metspalu. Method and apparatus for detection and analysis of biological materials through laser induced fluorescence. 03.11.2005. US2007091306, EP07105950.

Kaarel Krjutškov, Andres Metspalu. A method to determine single nucleotide polymorphisms and mutations in nucleic acid sequence. PCT/EE2007/000003. 06.03.2007

Anu Tammiste, Lili Milani, Krista Fischer, Tõnu Esko, Kristi Pettai, Kaarel Krjutskov, Reedik Mägi, Merike Leego, Eduard Maron, Andres Metspalu. A Method and a Kit to Predict Response to Antidepressant Treatment. US 61546607. 13.10.2011

Tarmo Annilo, Neeme Tõnisson, Tõnu Vooder, Kaie Kirotar, Urmo Võsa, Kristjan Välk, Raivo Kolde, Retlav Roosipuu, Jaak Vilo and Andres Metspalu. Methylation and MicroRNA markers of early-stage non-small cell lung cancer. US 6/1499578. 21.06.2011

OTHER RESEARCH RELATED ACTIVITIES

REFEREE FOR RESEARCH FUNDING ORGANISATIONS

2002 – 2004 The Wellcome Trust, UK Central and East European Fellowship program
2000 Helmholtz Society, Expert, Germany
2001– 2003 French Ministry of Research, Expert, France
2005 – 2006 EC FP6 SSA and CA projects Expert, Brussels
2008 ESFRI BMS Member from the Estonian Ministry of Research and Education
2008 EC ERC Senior Awards, Member of the Expert panel, Brussels
2009 – 2011 Chairman of the Lithuanian Ministry of Research and Education, Science Roadmap grant committee.
2011 Chairman for Netherlands Genomics Initiative of Centre for Medical Systems Biology site-visit

PhD THESIS COMMITTEES:

Riga Stradins University (RSU), Latvia; Leiden University, The Netherlands; Vilnius University, Lithuania.

EDITORIAL BOARDS

2003 – … Journal “Heredity Cancer in Clinical Practice” editorial board member
2004 – … Journal “Clinical Genetics” editorial board member
2006 – 2010 “Central European Science Journal” co-editor

SCIENTIFIC ADVISORY BOARDS

1988 – 1990 Member of the Expert Council of Soviet Union Human Genome program
1996 – … Member of the Scientific Council of the IMCB of University of Tartu
2000 –2010 European Society of Human Genetics as a board member and SPC member, member of Executive Board 2005-2007
2005 - 2007 President-elect, president and vice-president of the European Society of Human Genetics
1997 – 2000 Member of the Estonian Scientific Competence Council
2003 – 2009 Member of the Estonian Scientific Competence Council
2001 – 2011 European Science Foundation Functional Genomics Program: Member of steering committee
2003 – 2006 Biotechnology Centre of Excellence, Vilnius, Lithuania, Member of the International Advisory Board
2006 – 2007 Member of the Scientific Council of the Institute of Technology at University of Tartu
2007 - 2011 Veterinary and Food Board of the Estonian Government, Member of the Expert group for GMO- and Innovated Foods

INVITED PRESENTATIONS IN INTERNATIONAL CONFERENCES (> 50 in the last ten years)

"The Estonian Biobank and Human Genomics" Institut de Medicina Predictiva i Personalitzada del Cancer, 11.04.2011 Barcelona, Spain
Plenary session speaker "The Estonian Genome Project - 10 Years of Experience in Population Based Genome Analysis" 10th ScanBalt Forum 21.-24. 09. 2011, Heringsdorf, Germany
"Population based biobanks are for advancement of public health" 4th ESF Conference on Functional Genomics and Disease, April 14-17, 2010 Dresden, Germany
"The Estonian Biobank and Studies on Genetic Structure of Europeans" International Conference on Genomics (ICG-IV) Human and Beyond, Shenzhen, China 2-5.11.2009
"Genetic Structure of Europeans" Joint Congress Slovenian Biochemical Society and Slovenian Genetic Society, Otočec, Slovenia 20-23.09.2009
"A linkage disequilibrium map of human chromosome 22", 10th International Congress of Human Genetics ICHG 2001, Vienna, Austria, 15-19 May 2001
"DNA chips – methods and apparatus" in XVTH COURSE In MEDICAL GENETICS March 17-23, 2002 Bertinoro, Italy
Andres Metspalu. "Issues in setting up population DNA collections". European Human Genetics Conference 2003, Birmingham, UK, 3-6. mai 2003

INTERNATIONAL CONFERENCES – ORGANISING COMMITTEE (> 10 in the last ten years)

2003 – 2006 International Congress of Genetics (Brisbane, Australia) member of the SPC
2005–2010 Member of the SPC and organizational committee of the European Society Human Genetics Annual Conference
2005 – 2006 HUGO Genome Meeting in Helsinki, Member of the SPC
2000 – 2011 Chairman of SPC of the International Gene Forum, Tartu, Estonia http://www.geneforum.ee/GF2011/index.php?inc=lingid

INDUSTRIAL ACTIVITIES

1999 - ….. Asper Biotech Ltd., Tartu, Estonia. Founder and Chairman of the Board
2002 - 2004 EGeen International Inc., Foster City, CA, USA, Founder and Chief Scientific Officer

INTERNATIONAL PRIZES/AWARDS/ACADEMY MEMBERSHIPS

1980 Soviet Estonian Prize for Science
2001 3rd Class Order of the Estonian Red Cross
2002 Prix de la Garantie Medicale et Chirurgicale, France
2003 Estonian Science Award in Medicine

2003 L'Ordre des Palmes Académiques, Chevalier, France
2005 Member of the ScanBalt Academy
2010 Vilnius University *Dr. H. C.*
2010 Member of the Estonian Academy of Sciences
2011 Vice president of the Scan Balt Academy

TUTORED STUDENTS

18 PhD students: In University of Tartu, Estonia 1996: Illar Pata (senior sc. at Tallinn Technical University), Ants Kurg (professor at Univ. of Tartu); 1999: Tarmo Annilo (senior sc. At Univ. of Tartu), Ana Rebane (Swiss Institute of Allergy and Asthma Research, Davos); 2003: Jaana Tammur (senior sc. at Univ of Tartu); 2004: Tiina Kahre (Molecualr Diagnistic Centre at Tartu University Hospital); 2005: Neeme Tõnisson (senior sc. At Univ. of Tartu and residency at laboratory medicine), Krista Kaasik (UCSF, USA), Kati Koido; 2006: Elin Lõhmussaar (UCLA, USA); 2008: Janne Pullat (InBIo Ldt); 2010: Signe Altmäe (Karolinska Institute, Stockholm), Mari Nelis (Univ. Geneva Medical school), Kaarel Krjutškov (EGC, Univ. of Tartu) ; 2011: Tõnu Vooder (EGC Univ. of Tartu), Kristajan Välk (Competence Centre of Reproductive Medicine and Biology, Tartu, Estonia), Tiit Nikopensius (EGC Univ. of Tartu); 2012: Triin Jaagomägi (Dept. of Dental Medicine, Univ of Tartu).