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INTERNATIONAL EVALUATION OF RESEARCH AND DOCTORAL TRAINING AT THE UNIVERSITY OF HELSINKI 2005–2010

RC-Specific Evaluation of CSB – Cancer Systems Biology RC

Seppo Saari & Antti Moilanen (Eds.)



Evaluation Panel: Medicine, Biomedicine and Health Sciences

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Seppo Saari & Antti Moilanen

Title:

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International Evaluation of Research and Doctoral Training at the University of Helsinki 2005–2010 : RC-Specific Evaluation of CSB – Cancer Systems Biology RC

Summary:

Researcher Community (RC) was a new concept of the participating unit in the evaluation. Participation in the evaluation was voluntary and the RCs had to choose one of the five characteristic categories to participate.

Evaluation of the Researcher Community was based on the answers to the evaluation questions. In addition a list of publications and other activities were provided by the TUHAT system. The CWTS/Leiden University conducted analyses for 80 RCs and the Helsinki University Library for 66 RCs. Panellists, 49 and two special experts in five panels evaluated all the evaluation material as a whole and

discussed the feedback for RC-specific reports in the panel meetings in Helsinki. The main part of this report is consisted of the feedback which is published as such in the report. Chapters in the report:

- 1. Background for the evaluation
- 2. Evaluation feedback for the Researcher Community
- 3. List of publications

4. List of activities

5. Bibliometric analyses

The level of the RCs' success can be concluded from the written feedback together with the numeric evaluation of four evaluation questions and the category fitness. More conclusions of the success can be drawn based on the University-level report.

RC-specific information:

Main scientific field of research: Medicine, Biomedicine and Health Sciences

RC-specific keywords:

cancer, predisposition, familial, systems biology, regulatory, bioinformatics, molecular epidemiology

Participation category:

1. Research of the participating community represents the international cutting edge in its field

RC's responsible person:

Aaltonen, Lauri

Keywords:

Research Evaluation, Meta-evaluation, Doctoral Training, Bibliometric Analyses, Researcher Community

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Foreword

The evaluation of research and doctoral training is being carried out in the years 2010–2012 and will end in 2012. The steering group appointed by the Rector in January 2010 set the conditions for participating in the evaluation and prepared the Terms of Reference to present the evaluation procedure and criteria. The publications and other scientific activities included in the evaluation covered the years 2005–2010.

The participating unit in the evaluation was defined as a Researcher Community (RC). To obtain a critical mass with university-level impact, the number of members was set to range from 20 to 120. The RCs were required to contain researchers in all stages of their research career, from doctoral students to principal investigators (PIs). All in all, 136 Researcher Communities participated in this voluntary evaluation, 5857 persons in total, of whom 1131 were principal investigators. PIs were allowed to participate in two communities in certain cases, and 72 of them used this opportunity and participated in two RCs.

This evaluation enabled researchers to define RCs from the "bottom up" and across disciplines. The aim of the evaluation was not to assess individual performance but a community with shared aims and researcher-training activities. The RCs were able to choose among five different categories that characterised the status and main aims of their research. The steering group considered the process of applying to participate in the evaluation to be important, which lead to the establishment of these categories. In addition, providing a service for the RCs to enable them to benchmark their research at the global level was a main goal of the evaluation.

The data for the evaluation consisted of the RCs' answers to evaluation questions on supplied e-forms and a compilation extracted from the TUHAT – Research Information System (RIS) on 12 April 2011. The compilation covered scientific and other publications as well as certain areas of scientific activities. During the process, the RCs were asked to check the list of publications and other scientific activities and make corrections if needed. These TUHAT compilations are public and available on the evaluation project sites of each RC in the TUHAT-RIS.

In addition to the e-form and TUHAT compilation, University of Leiden (CWTS) carried out bibliometric analyses from the articles included in the Web of Science (WoS). This was done on University and RC levels. In cases where the publication forums of the RC were clearly not represented by the WoS data, the Library of the University of Helsinki conducted a separate analysis of the publications. This was done for 66 RCs representing the humanities and social sciences.

The evaluation office also carried out an enquiry targeted to the supervisors and PhD candidates about the organisation of doctoral studies at the University of Helsinki. This and other documents describing the University and the Finnish higher education system were provided to the panellists.

The panel feedback for each RC is unique and presented as an entity. The first collective evaluation reports available for the whole panel were prepared in July-August 2011. The reports were accessible to all panel members via the electronic evaluation platform in August. Scoring from 1 to 5 was used to complement written feedback in association with evaluation questions 1–4 (scientific focus and quality, doctoral training, societal impact, cooperation) and in addition to the category evaluating the fitness for participation in the evaluation. Panellists used the international level as a point of comparison in the evaluation. Scoring was not expected to go along with a preset deviation.

Each of the draft reports were discussed and dealt with by the panel in meetings in Helsinki (from 11 September to 13 September or from 18 September to 20 September 2011). In these meetings the panels also examined the deviations among the scores and finalised the draft reports together.

The current RC-specific report deals shortly with the background of the evaluation and the terms of participation. The main evaluation feedback is provided in the evaluation report, organised according to the evaluation questions. The original material provided by the RCs for the panellists has been attached to these documents.

On behalf of the evaluation steering group and office, I sincerely wish to thank you warmly for your participation in this evaluation. The effort you made in submitting the data to TUHAT-RIS is gratefully acknowledged by the University. We wish that you find this panel feedback useful in many ways. The bibliometric profiles may open a new view on your publication forums and provide a perspective for discussion on your choice of forums. We especially hope that this evaluation report will help you in setting the future goals of your research.

Johanna Björkroth Vice-Rector Chair of the Steering Group of the Evaluation

Steering Group of the evaluation

Steering group, nominated by the Rector of the University, was responsible for the planning of the evaluation and its implementation having altogether 22 meetings between February 2010 and March 2012.

Chair

Vice-Rector, professor Johanna Björkroth

Vice-Chair

Professor Marja Airaksinen

Chief Information Specialist, Dr Maria Forsman Professor Arto Mustajoki University Lecturer, Dr Kirsi Pyhältö Director of Strategic Planning and Development, Dr Ossi Tuomi Doctoral candidate, MSocSc Jussi Vauhkonen

Panel members

CHAIR

Professor Lorenz Poellinger Cancer biology, cell and molecular biology Karolinska Institute, Sweden

VICE-CHAIR

Professor Cornelia van Duijn Genetic epidemiology, Alzheimer's disease and related disorders Erasmus Medical Centre, the Netherlands

Professor Johanna Ivaska

Molecular cell biology, cell adhesion, cancer biology University of Turku, VTT Technical Research Centre, Finland

Professor Olli Lassila

Immunology, medical microbiology University of Turku, Finland

Professor Hans-Christian Pape

Neuroscience, neurophysiology University of Münster, Germany

Professor Thomas Ruzicka Dermatology, allergology Ludwig-Maximilians-Universität (LMU) München, Germany

Professor Lars Terenius

Experimental alcohol and drug dependence research, mental disorders, preventive medicine Karolinska Institute, Sweden

Professor Peter York

Physical pharmaceutics, pharmaceutical chemistry, pharmaceutical technology University of Bradford, Great Britain

The panel, independently, evaluated all the submitted material and was responsible for the feedback of the RC-specific reports. The panel members were asked to confirm whether they had any conflict of interests with the RCs. If this was the case, the panel members disqualified themselves in discussion and report writing.

Added expertise to the evaluation was contributed by two evaluators outside the panels and by three members from the other panels.

External Experts

Professor Olli Carpén Pathology, cancer cell metastasis University of Turku Finland

Professor Anders Linde

Oral biochemi Faculty of Odontology Göteborg University Sweden

Experts from the Other Panels

Professor Jan-Otto Carlsson, from the Panel of Natural Sciences Professor Danny Huylebroek, from the Panel of Biological, Agricultural and Veterinary Sciences

Professor Holger Stark, from the Panel of Natural Sciences

EVALUATION OFFICE

Dr Seppo Saari, Doc., Senior Adviser in Evaluation, was responsible for the entire evaluation, its planning and implementation and acted as an Editor-in-chief of the reports.

Dr Eeva Sievi, **Doc.**, Adviser, was responsible for the registration and evaluation material compilations for the panellists. She worked in the evaluation office from August 2010 to July 2011.

MSocSc Paula Ranne, Planning Officer, was responsible for organising the panel meetings and all the other practical issues like agreements and fees and editing a part the RC-specific reports. She worked in the evaluation office from March 2011 to January 2012.

Mr Antti Moilanen, Project Secretary, was responsible for editing the reports. He worked in the evaluation office from January 2012 to April 2012.

TUHAT OFFICE

Provision of the publication and other scientific activity data

Mrs Aija Kaitera, Project Manager of TUHAT-RIS served the project ex officio providing the evaluation project with the updated information from TUHAT-RIS. The TUHAT office assisted in mapping the publications with CWTS/University of Leiden.

MA Liisa Ekebom, Assisting Officer, served in TUHAT-RIS updating the publications for the evaluation. She also assisted the UH/Library analyses.

BA Liisa Jäppinen, Assisting Officer, served in TUHAT-RIS updating the publications for the evaluation.

HELSINKI UNIVERSITY LIBRARY

Provision of the publication analyses

Dr Maria Forsman, Chief Information Specialist in the Helsinki University Library, managed with her 10 colleagues the bibliometric analyses in humanities, social sciences and in other fields of sciences where CWTS analyses were not applicable.

Acronyms and abbreviations applied in the report

External competitive funding

AF - Academy of Finland TEKES - Finnish Funding Agency for Technology and Innovation EU - European Union ERC - European Research Council International and national foundations FP7/6 etc. /Framework Programmes/Funding of European Commission

Evaluation marks

- Outstanding (5)Excellent(4)Very Good(3)Good(2)
- Sufficient (1)

Abbreviations of Bibliometric Indicators

P - Number of publications

TCS - Total number of citations

MCS - Number of citations per publication, excluding self-citations

PNC - Percentage of uncited publications

MNCS - Field-normalized number of citations per publication

MNJS - Field-normalized average journal impact

THCP10 - Field-normalized proportion highly cited publications (top 10%)

INT_COV - Internal coverage, the average amount of references covered by the WoS

WoS - Thomson Reuters Web of Science Databases

Participation category

Category 1. The research of the participating community represents the international cutting edge in its field.

Category 2. The research of the participating community is of high quality, but the community in its present composition has yet to achieve strong international recognition or a clear break-through.

Category 3. The research of the participating community is distinct from mainstream research, and the special features of the research tradition in the field must be considered in the evaluation.

Category 4. The research of the participating community represents an innovative opening.

Category 5. The research of the participating community has a highly significant societal impact.

Research focus areas of the University of Helsinki

Focus area 1: The basic structure, materials and natural resources of the physical world

Focus area 2: The basic structure of life

Focus area 3: The changing environment – clean water

Focus area 4: The thinking and learning human being

Focus area 5: Welfare and safety

Focus area 6: Clinical research

Focus area 7: Precise reasoning

Focus area 8: Language and culture

Focus area 9: Social justice

Focus area 10: Globalisation and social change

1 Introduction to the Evaluation

1.1 RC-specific evaluation reports

The participants in the evaluation of research and doctoral training were Researcher Communities (hereafter referred to as the RC). The RC refers to the group of researchers who registered together in the evaluation of their research and doctoral training. Preconditions in forming RCs were stated in the Guidelines for the Participating Researcher Communities. The RCs defined themselves whether their compositions should be considered well-established or new.

It is essential to emphasise that the evaluation combines both meta-evaluation¹ and traditional research assessment exercise and its focus is both on the research outcomes and procedures associated with research and doctoral training. The approach to the evaluation is enhancement-led where self-evaluation constituted the main information. The answers to the evaluation questions formed together with the information of publications and other scientific activities an entity that was to be reviewed as a whole.

The present evaluation recognizes and justifies the diversity of research practices and publication traditions. Traditional Research Assessment Exercises do not necessarily value high quality research with low volumes or research distinct from mainstream research. It is challenging to expose the diversity of research to fair comparison. To understand the essence of different research practices and to do justice to their diversity was one of the main challenges of the present evaluation method. Understanding the divergent starting points of the RCs demanded sensitivity from the evaluators.

1.2 Aims and objectives in the evaluation

The aims of the evaluation are as follows:

- to improve the level of research and doctoral training at the University of Helsinki and to raise their international profile in accordance with the University's strategic policies. The improvement of doctoral training should be compared to the University's policy.²
- to enhance the research conducted at the University by taking into account the diversity, originality, multidisciplinary nature, success and field-specificity,
- to recognize the conditions and prerequisites under which excellent, original and high-impact research is carried out,
- to offer the academic community the opportunity to receive topical and versatile international peer feedback,
- to better recognize the University's research potential.
- to exploit the University's TUHAT research information system to enable transparency of publishing activities and in the production of reliable, comparable data.

1.3 Evaluation method

The evaluation can be considered as an enhancement-led evaluation. Instead of ranking, the main aim is to provide useful information for the enhancement of research and doctoral training of the participating RCs. The comparison should take into account each field of science and acknowledge their special character.

¹ The panellists did not read research reports or abstracts but instead, they evaluated answers to the evaluation questions, tables and compilations of publications, other scientific activities, bibliometrics or comparable analyses.

² <u>Policies on doctoral degrees and other postgraduate degrees at the University of Helsinki</u>.

The comparison produced information about the present status and factors that have lead to success. Also challenges in the operations and outcomes were recognized.

The evaluation approach has been designed to recognize better the significance and specific nature of researcher communities and research areas in the multidisciplinary top-level university. Furthermore, one of the aims of the evaluation is to bring to light those evaluation aspects that differ from the prevalent ones. Thus the views of various fields of research can be described and research arising from various starting points understood better. The doctoral training is integrated into the evaluation as a natural component related to research. Operational processes of doctoral training are being examined in the evaluation.

Five stages of the evaluation method were:

- 1. Registration Stage 1
- 2. Self-evaluation Stage 2
- 3. TUHAT³ compilations on publications and other scientific activities⁴
- 4. External evaluation
- 5. Public reporting

1.4 Implementation of the external evaluation

Five Evaluation Panels

Five evaluation panels consisted of independent, renowned and highly respected experts. The main domains of the panels are:

- 1. biological, agricultural and veterinary sciences
- 2. medicine, biomedicine and health sciences
- 3. natural sciences
- 4. humanities
- 5. social sciences

The University invited 10 renowned scientists to act as chairs or vice-chairs of the five panels based on the suggestions of faculties and independent institutes. Besides leading the work of the panel, an additional role of the chairs was to discuss with other panel chairs in order to adopt a broadly similar approach. The panel chairs and vice-chairs had a pre-meeting on 27 May 2011 in Amsterdam.

The panel compositions were nominated by the Rector of the University 27 April 2011. The participating RCs suggested the panel members. The total number of panel members was 50. The reason for a smaller number of panellists as compared to the previous evaluations was the character of the evaluation as a meta-evaluation. The panellists did not read research reports or abstracts but instead, they evaluated answers to the evaluation questions, tables and compilations of publications, other scientific activities, bibliometrics and comparable analyses.

The panel meetings were held in Helsinki:

- On 11–13 September 2011: (1) biological, agricultural and veterinary sciences, (2) medicine, biomedicine and health sciences and (3) natural sciences.
- On 18–20 September 2011: (4) humanities and (5) social sciences.

³ TUHAT (acronym) of Research Information System (RIS) of the University of Helsinki

⁴ Supervision of thesis, prizes and awards, editorial work and peer reviews, participation in committees, boards and networks and public appearances.

1.5 Evaluation material

The main material in the evaluation was the RCs' self-evaluations that were qualitative in character and allowed the RCs to choose what was important to mention or emphasise and what was left unmentioned.

The present evaluation is exceptional at least in the Finnish context because it is based on both the evaluation documentation (self-evaluation questions, publications and other scientific activities) and the bibliometric reports. All documents were delivered to the panellists for examination.

Traditional bibliometrics can be reasonably done mainly in medicine, biosciences and natural sciences when using the Web of Science database, for example. Bibliometrics, provided by CWTS/The Centre for Science and Technology Studies, University of Leiden, cover only the publications that include WoS identification in the TUHAT-RIS.

Traditional bibliometrics are seldom relevant in humanities and social sciences because the international comparable databases do not store every type of high quality research publications, such as books and monographs and scientific journals in other languages than English. The Helsinki University Library has done analysis to the RCs, if their publications were not well represented in the Web of Science databases (RCs should have at least 50 publications and internal coverage of publications more than 40%) – it meant 58 RCs. The bibliometric material for the evaluation panels was available in June 2011. The RC-specific bibliometric reports are attached at the end of each report.

The panels were provided with the evaluation material and all other necessary background information, such as the basic information about the University of Helsinki and the Finnish higher education system.

Evaluation material

- 1. Registration documents of the RCs for the background information
- 2. Self evaluation material answers to the evaluation questions
- 3. Publications and other scientific activities based on the TUHAT RIS:
 - 3.1. statistics of publications
 - 3.2. list of publications
 - 3.3. statistics of other scientific activities
 - 3.4. list of other scientific activities
- 4. Bibliometrics and comparable analyses:
 - 4.1. Analyses of publications based on the verification of TUHAT-RIS publications with the Web of Science publications (CWTS/University of Leiden)
 - 4.2. Publication statistics analysed by the Helsinki University Library mainly for humanities and social sciences
- 5. University level survey on doctoral training (August 2011)
- 6. University level analysis on publications 2005–2010 (August 2011) provided by CWTS/University of Leiden

Background material

University of Helsinki

- Basic information about the University of the Helsinki
- The structure of doctoral training at the University of Helsinki
- Previous evaluations of research at the University of Helsinki links to the reports: <u>1998</u> and <u>2005</u>

The Finnish Universities/Research Institutes

- Finnish University system
- Evaluation of the Finnish National Innovation System
- <u>The State and Quality of Scientific Research in Finland. Publication of the Academy of Finland</u> <u>9/09.</u>

The evaluation panels were provided also with other relevant material on request before the meetings in Helsinki.

1.6 Evaluation questions and material

The participating RCs answered the following evaluation questions which are presented according to the evaluation form. In addition, TUHAT RIS was used to provide the **additional material** as explained. For giving the feedback to the RCs, the panellists received the evaluation feedback form constructed in line with the evaluation questions:

1. Focus and quality of the RC's research

- Description of
 - the RC's research focus.
 - the quality of the RC's research (incl. key research questions and results)
 - the scientific significance of the RC's research in the research field(s)

 Identification of the ways to strengthen the focus and improve the quality of the RC's research The additional material: TUHAT compilation of the RC's publications, analysis of the RC's publications data (provided by University of Leiden and the Helsinki University Library)

A written feedback from the aspects of: scientific quality, scientific significance, societal impact, innovativeness

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

2. Practises and quality of doctoral training

- Organising of the doctoral training in the RC. Description of the RC's principles for:
- recruitment and selection of doctoral candidates
- supervision of doctoral candidates

- collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes

- good practises and quality assurance in doctoral training
- assuring of good career perspectives for the doctoral candidates/fresh doctorates
- Identification of the RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.

The additional material: TUHAT compilation of the RC's other scientific activities/supervision of doctoral dissertations

A written feedback from the aspects of: processes and good practices related to leadership and management

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

3. The societal impact of research and doctoral training

- Description on how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).
- Identification of the ways to strengthen the societal impact of the RC's research and doctoral training.

The additional material: TUHAT compilation of the RC's other scientific activities. A written feedback from the aspects of: societal impact, national and international collaboration, innovativeness

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

4. International and national (incl. intersectoral) research collaboration and researcher mobility

Description of

- the RC's research collaborations and joint doctoral training activities - how the RC has promoted researcher mobility
- Identification of the RC's strengths and challenges related to research collaboration and researcher mobility, and the actions planned for their development.
- A written feedback from the aspects of: scientific quality, national and international collaboration
 - Strengths
 - Areas of development
 - Other remarks
 - Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

5. Operational conditions

- Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).
- Identification of the RC's strengths and challenges related to operational conditions, and the
 actions planned for their development.

A written feedback from the aspects of: processes and good practices related to leadership and management

- Strengths
- Areas of development
- Other remarks
- Recommendations

6. Leadership and management in the researcher community

- Description of
 - the execution and processes of leadership in the RC
 - how the management-related responsibilities and roles are distributed in the RC
 - how the leadership- and management-related processes support
 - high quality research
 - collaboration between principal investigators and other researchers in the RC
 - the RC's research focus
 - strengthening of the RC's know-how
- Identification of the RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes

7. External competitive funding of the RC

- The RCs were asked to provide information of such external competitive funding, where:
 - the funding decisions have been made during 1.1.2005-31.12.2010, and
 - the administrator of the funding is/has been the University of Helsinki
- On the e-form the RCs were asked to provide:

1) The relevant funding source(s) from a given list (Academy of Finland/Research Council, TEKES/The Finnish Funding Agency for Technology and Innovation , EU, ERC, foundations, other national funding organisations), and

2)The total sum of funding which the organisation in question had decided to allocate to the RCs members during 1.1.2005–31.12.2010.

Competitive funding reported in the text is also to be considered when evaluating this point. A written feedback from the aspects of: scientific quality, scientific significance, societal impact, innovativeness, future significance

- Strengths
- Areas of development
- Other remarks
- Recommendations

8. The RC's strategic action plan for 2011–2013

RC's description of their future perspectives in relation to research and doctoral training.
 A written feedback from the aspects of: scientific quality, scientific significance, societal Impact, processes and good practices related to leadership and management, national and international collaboration, innovativeness, future significance

- Strengths
- Areas of development

- Other remarks
- Recommendations

9. Evaluation of the category of the RC in the context of entity of the evaluation material (1-8)

The RC's fitness to the chosen participation category

A written feedback evaluating the RC's fitness to the chosen participation category

- Strengths
- Areas of development
- Other remarks
- Recommendations

Numeric evaluation: OUTSTANDING (5), EXCELLENT (4), VERY GOOD (3), GOOD (2), SUFFICIENT (1)

10. Short description of how the RC members contributed the compilation of the stage 2 material Comments on the compilation of evaluation material

11. How the UH's focus areas are presented in the RC's research? Comments if applicable

12. RC-specific main recommendations based on the previous questions 1–11

13. RC-specific conclusions

1.7 Evaluation criteria

The panellists were expected to give evaluative and analytical feedback to each evaluation question according to their aspects in order to describe and justify the quality of the submitted material. In addition, the evaluation feedback was asked to be pointed out the level of the performance according to the following classifications:

•	outstanding	(5)
•	excellent	(4)
•	very good	(3)
•	good	(2)
•	sufficient	(1)

Evaluation according to the criteria was to be made with thorough consideration of the entire evaluation material of the RC in question. Finally, in questions 1-4 and 9, the panellists were expected to classify their written feedback into one of the provided levels (the levels included respective descriptions, 'criteria'). Some panels used decimals in marks. The descriptive level was interpreted according to the integers and not rounding up the decimals by the editors.

Description of criteria levels

Question 1 – FOCUS AND QUALITY OF THE RC'S RESEARCH

Classification: Criteria (level of procedures and results)

Outstanding quality of procedures and results (5)

Outstandingly strong research, also from international perspective. Attracts great international interest with a wide impact, including publications in leading journals and/or monographs published by leading international publishing houses. The research has world leading qualities. The research focus, key research questions scientific significance, societal impact and innovativeness are of outstanding quality.

In cases where the research is of a national character and, in the judgement of the evaluators, should remain so, the concepts of "international attention" or "international impact" etc. in the grading criteria above may be replaced by "international comparability".

Operations and procedures are of outstanding quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are in alignment with the documentation. The ambition to develop the community together is of outstanding quality.

Excellent quality of procedures and results (4)

Research of excellent quality. Typically published with great impact, also internationally. Without doubt, the research has a leading position in its field in Finland.

Operations and procedures are of excellent quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of excellent quality.

Very good quality of procedures and results (3)

The research is of such very good quality that it attracts wide national and international attention.

Operations and procedures are of very good quality, transparent and shared in the community. The improvement of research and other efforts are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of very good quality.

Good quality of procedures and results (2)

Good research attracting mainly national attention but possessing international potential, extraordinarily high relevance may motivate good research.

Operations and procedures are of good quality, shared occasionally in the community. The improvement of research and other efforts are occasionally documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of good quality.

Sufficient quality of procedures and results (1)

In some cases the research is insufficient and reports do not gain wide circulation or do not have national or international attention. Research activities should be revised.

Operations and procedures are of sufficient quality, shared occasionally in the community. The improvement of research and other efforts are occasionally documented and operations and practices are to some extent in alignment with the documentation. The ambition to develop the community together is of sufficient quality.

Question 2 – DOCTORAL TRAINING Question 3 – SOCIETAL IMPACT Question 4 – COLLABORATION

Classification: Criteria (level of procedures and results)

Outstanding quality of procedures and results (5)

Procedures are of outstanding quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are in alignment with the documentation. The ambition to develop the community together is of outstanding quality. The procedures and results are regularly evaluated and the feedback has an effect on the planning.

Excellent quality of procedures and results (4)

Procedures are of excellent quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of excellent quality. The procedures and outcomes are evaluated and the feedback has an effect on the planning.

Very good quality of procedures and results (3)

Procedures are of very good quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and

management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of very good quality.

Good quality of procedures and results (2)

Procedures are of good quality, shared occasionally in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are documented and operations and practices are to large extent in alignment with the documentation. The ambition to develop the community together is of good quality.

Sufficient quality of procedures and results (1)

Procedures are of sufficient quality, transparent and shared in the community. The practices and quality of doctoral training/societal impact/international and national collaboration/leadership and management are occasionally documented and operations and practices are to some extent in alignment with the documentation. The ambition to develop the community together is of sufficient quality.

Question 9 – CATEGORY

Participation category - fitness for the category chosen

The choice and justification for the chosen category below should be reflected in the RC's responses to the evaluation questions 1–8.

- 1. The research of the participating community represents the international cutting edge in its field.
- 2. The research of the participating community is of high quality, but the community in its present composition has yet to achieve strong international recognition or a clear break-through.
- 3. The research of the participating community is distinct from mainstream research, and the special features of the research tradition in the field must be considered in the evaluation. The research is of high quality and has great significance and impact in its field. However, the generally used research evaluation methods do not necessarily shed sufficient light on the merits of the research.
- 4. The research of the participating community represents an innovative opening. A new opening can be an innovative combination of research fields, or it can be proven to have a special social, national or international demand or other significance. Even if the researcher community in its present composition has yet to obtain proof of international success, its members can produce convincing evidence of the high level of their previous research.
- 5. The research of the participating community has a highly significant societal impact. The participating researcher community is able to justify the high social significance of its research. The research may relate to national legislation, media visibility or participation in social debate, or other activities promoting social development and human welfare. In addition to having societal impact, the research must be of a high standard.

An example of outstanding fitness for category choice (5) ⁵

The RC's representation and argumentation for the chosen category were convincing. The RC recognized its real capacity and apparent outcomes in a wider context to the research communities. The specific character of the RC was well-recognized and well stated in the responses. The RC fitted optimally for the category.

•	Outstanding	(5)
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- Excellent (4)
- Very good (3)
- Good (2)
- Sufficient (1)

The above-mentioned definition of outstanding was only an example in order to assist the panellists in the positioning of the classification. There was no exact definition for the category fitness.

⁵ The panels discussed the category fitness and made the final conclusions of the interpretation of it.

1.8 Timetable of the evaluation

The main timetable of the evaluation:

- 1. Registration
- 2. Submission of self-evaluation materials
- 3. External peer review
- 4. Published reports
 - University level public report
 - RC specific reports

November 2010 January-February 2011 May-September 2011 March-April 2012

The entire evaluation was implemented during the university's strategy period 2010–2012. The preliminary results were available for the planning of the following strategy period in late autumn 2011. The evaluation reports will be published in March/April 2012. More detailed time schedule is published in the University report.

1.9 Evaluation feedback – consensus of the entire panel

The panellists evaluated all the RC-specific material before the meetings in Helsinki and mailed the draft reports to the evaluation office. The latest interim versions were on-line available to all the panellists on the Wiki-sites. In September 2011, in Helsinki the panels discussed the material, revised the first draft reports and decided the final numeric evaluation. After the meetings in Helsinki, the panels continued working and finalised the reports before the end of November 2011. The final RC-specific reports are the consensus of the entire panel.

The evaluation reports were written by the panels independently. During the editing process, the evaluation office requested some clarifications from the panels when necessary. The tone and style in the reports were not harmonized in the editing process. All the reports follow the original texts written by the panels as far as it was possible.

The original evaluation material of the RCs, provided for the panellists is attached at the end of the report. It is essential to notice that the exported lists of publications and other scientific activities depend how the data was stored in the TUHAT-RIS by the RCs.

2 Evaluation feedback

2.1 Focus and quality of the RC's research

Description of

- the RC's research focus
- the quality of the RC's research (incl. key research questions and results)
- the scientific significance of the RC's research in the research field(s)

 Identification of the ways to strengthen the focus and improve the quality of the RC's research ASPECTS: Scientific quality, scientific significance, societal impact, innovativeness

This RC is an apparently tight and mutually beneficial collaborative network of two world-class established PIs and a third somewhat more junior PI who also has a very good track record. Together these PIs and their research groups master the multidisciplinary expertise necessary for top notch research in the rapidly developing field of cancer genomics.

The scientific excellence of this RC is obvious. The group has been shortlisted for an Academy of Finland Centre of Excellence (CoE), two of the PIs are Academy of Finland professors and have a European Reseach Council (ERC) advanced grant and their individual track records are outstanding. They also have shared publications. In addition to the obvious excellence of the individual PIs in the consortium, their collaboration brings the added benefit of combining expertise in different areas. Dr. Hautaniemi's team is expert in computational systems biology. Professor Aaltonen's group is expert in tumor genomics and Professor Taipale has set up world-class methodology for high-throughput screening and bioinformatics to allow a systems biology approach for the investigation of cancer cell biology.

Taken together, this is clearly an RC which is second to a few in Finland and brings together top PIs working with outstanding resources with complementary skills and expertise. The practical impact of the fact that one of the PIs is mainly working in Stockholm is not clearly discussed and the description that "the physical location next to each other provides excellent means to easily organize efforts" (5 Operational condition) considered by the panel as somewhat conflicting.

Numeric evaluation: 5 (Outstanding)

2.2 Practises and quality of doctoral training

- Organising of the doctoral training in the RC. Description of the RC's principles for:
 - recruitment and selection of doctoral candidates
 - supervision of doctoral candidates
 - collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes
 - good practises and quality assurance in doctoral training
 - assuring of good career perspectives for the doctoral candidates/fresh doctorates
- Identification of the RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.
- Additional material: TUHAT compilation of the RC's other scientific activities/supervision of doctoral dissertations

ASPECTS: Processes and good practices related to leadership and management

The RC has a good track record in training PhDs. Thus far 24 PhD theses have been supervised, ten of these have been approved with distinction and several have won prizes. The PIs are also involved in the doctoral training in Finland since all three have served in graduate school boards at the University of Helsinki (UH).

The RC finds it easy to recruit top students due to their excellent publication record and evidence for good practical hands-on supervision. Each student is supervised by a PI and a junior researcher, progress of the thesis is monitored in supervisory committees and regular meetings. A clear system has been set up where the demands for the student are gradually increased every year during their studies with the aim to support independence and maturity as scientists. The panel finds the doctoral training to be of very high quality.

For post-doctoral fellows, it is recognized that the aim of their training is to learn skills needed to become an independent PI. This is important.

One of the challenges of the RC is that due to the multidisciplinary nature of the research, it may be difficult to recruit students with expertise in biology, genomics and computational sciences. The aim of the RC is to train scientists in a cross-disciplinary manner. Based on the material submitted, the RC feels that the UH is not supportive enough of such training and this should be improved.

Numeric evaluation: 5 (Outstanding)

2.3 The societal impact of research and doctoral training

- Description on how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).
- Identification of the ways to strengthen the societal impact of the RC's research and doctoral training.
- Additional material: TUHAT compilation of the RC's other scientific activities.

ASPECTS: Societal impact, national and international collaboration, innovativeness

Outstanding research is always closely linked with high societal impact, good collaborations both nationally and internationally as well as a generation of innovations. This RC has clearly contributed in all of these areas.

The RC has a good track record in training PhDs. Thus far 24 PhD theses have been supervised. In addition, many post-docs have received training in these groups. The impact of training these top experts should not be underestimated. The PIs in this RC have also been active in policy making and have been involved in many positions responsible for development and management of the Finnish bioscience research and legislation.

The group has actively communicated their scientific discoveries to the public.

Most importantly, their science has generated innovations and breakthroughs with clinical and commercial impact.

The RC plans to continue their active role in communicating science to the general public and in engaging in discussion with experts in different forums.

Numeric evaluation: 5 (Outstanding)

2.4 International and national (incl. intersectoral) research collaboration and researcher mobility

- Description of
 - the RC's research collaborations and joint doctoral training activities
 - how the RC has promoted researcher mobility
- Identification of the RC's strengths and challenges related to research collaboration and researcher mobility, and the actions planned for their development.

ASPECTS: Scientific quality, national and international collaboration

The members of this RC have strong national and international collaborations. This is evident both from their publication record as well as from their involvement in many multi-national consortia. The top-class research of these groups has facilitated their ability to establish necessary international collaborations. At present they are involved in two of EU's Seventh Framework Programme (FP7) consortia and have an impressive list of international collaborators.

The strong existing international links and the commitment to promote student mobility guarantee that students and post-docs working in the groups will be offered with possibilities for research exchange and international meetings. It is important that this kind of international training and gaining of experience is supported by the RC and students are actively encouraged to participate in such activities.

Numeric evaluation: 5 (Outstanding)

2.5 Operational conditions

- Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).
- Identification of the RC's strengths and challenges related to operational conditions, and the actions
 planned for their development.

ASPECTS: Processes and good practices related to leadership and management

All of the three groups of this RC are very well funded. They work in environments with the best infrastructure and resources. In addition to the standard research requirements, the ability to exploit the unique Finnish resource, the Finnish Cancer Registry, is key to their research. They also have good national connections to clinical pathology departments.

At present, all the resources necessary for their continued success seem in place. However, it is important to emphasize that continued investment in research infrastructure in the UH and Finland is absolutely critical. The fact that one of the groups is predominantly working in Stockholm might hamper the close collaboration described by the RC in their documents.

2.6 Leadership and management in the researcher community

- Description of
 - the execution and processes of leadership in the RC
 - how the management-related responsibilities and roles are distributed in the RC
 - how the leadership- and management-related processes support
 - high quality research
 - collaboration between principal investigators and other researchers in the RC
 - the RC's research focus
 - strengthening of the RC's know-how
- Identification of the RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes

ASPECTS: Processes and good practices related to leadership and management

Rather than presenting a clear hierarchical management structure, the RC relies on PI driven management of the individual groups. In addition, they rely on the science and the efforts needed for scientific discoveries in guiding the necessary actions. This seems like the best possible management structure since science, not predetermined structures or goals, should be the primary director of research efforts also elsewhere. Thus the RC is composed of three independent groups working together.

Within the groups the supervision has been organized by sharing the responsibility between the PIs and other senior scientists in the group. Know-how is actively shared between the groups. In their material the RC emphasizes how close physical location of the groups is key to collaboration. This seems a bit odd as one of the groups is predominantly located in Sweden.

The current organization is clearly functioning well and no major changes are needed.

2.7 External competitive funding of the RC

• The RCs were asked to provide information of such external competitive funding, where:

• the funding decisions have been made during 1.1.2005-31.12.2010, and

- the administrator of the funding is/has been the University of Helsinki
- On the e-form the RCs were asked to provide:

1) The relevant funding source(s) from a given list (Academy of Finland/Research Council, TEKES/The Finnish Funding Agency for Technology and Innovation, EU, ERC, foundations, other national funding organisations, other international funding organizations), and

2) The total sum of funding which the organisation in question had decided to allocate to the RCs members during 1.1.2005–31.12.2010.

Competitive funding reported in the text is also to be considered when evaluating this point. ASPECTS: Scientific quality, scientific significance, societal impact, innovativeness and future significance

The funding of the RC is exceptional in volume and in versatility. Significant funding has been secured from many national as well as international sources. The RC has been very successful in winning highly competitive funding, such as ERC advanced grants.

2.8 The RC's strategic action plan for 2011–2013

• RC's description of their future perspectives in relation to research and doctoral training. ASPECTS: Scientific quality, scientific significance, societal Impact, processes and good practices related to leadership and management, national and international collaboration, innovativeness, future significance

The RC has been shortlisted for an Academy of Finland CoE position for the next five years. These plans are likely to shape the future collaboration. The groups plan to collaborate in research falling under three main goals: 1) Identification of cancer predisposition genes, 2) Characterization of common cancer susceptibility variant and 3) Characterization of mutatomes.

These are all important goals with significant clinical and societal impact. It is likely that world-class science will be achieved also in the future related to the goals and others.

The training provided by the RC has been excellent in the past and the plans are to continue providing excellent PhD student training. Also international student training will be an emphasis in the future as well. International collaborations will be continued for example in the form of EU consortia.

2.9 Evaluation of the category of the RC in the context of entity of the evaluation material (1-8)

The RC's fitness to the chosen participation category. Category 1. The research of the participating community represents the international cutting edge in its field.

The category chosen is particularly suitable for this RC. It is clear that their research and doctoral training represent the international cutting edge in its field.

This is evident based on all the evaluation criteria. The scientific track-record is outstanding. The secured funding is exceptionally high, the research environment is the best in Finland and the training of young researchers is of a high standard.

Numeric evaluation: 5 (Outstanding)

2.10 Short description of how the RC members contributed the compilation of the stage 2 material

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2.11 How the UH's focus areas are presented in the RC's research

Focus area 2: The basic structure of life

2.12 RC-specific main recommendations

The RC is clearly world class and the collaboration of the three groups with distinct expertise is clearly mutually beneficial. The groups have clearly found their own niche which is combining clinical and epidemiological data with modern genomics, systems biology approaches and computational science. The recommendation is: "Keep up the good work!"

2.13 RC-specific conclusions

This RC is clearly world-class and an example to others in setting high standards and achieving them. All the aspects to be evaluated: Science, collaborations, training of students and post-docs, societal impact and fitting with the UH strategy are all at their highest levels. No major changes are suggested.

2.14 Preliminary findings in the Panel-specific feedback

Everything is excellent.

2.15 Preliminary findings in the University-level evaluation

Again, everything is as good as it gets. The groups have found their own research tracks where they clearly excel and the collaborations between the groups seem to be mutually beneficial and clearly bring added value.

The groups have a great track record in doctoral training with over 20 supervised PhD theses and many of them approved with honors. In addition, many of the post-docs trained by these RCs have found suitable work after leaving the laboratories, warranting commendation.

The world-class medical research carried out by the groups is likely to have an important societal impact in facilitating the development of future cures. In addition, the groups have been actively communicating their findings to the general public. The PIs have also been actively involved in policy making.

The groups are predominantly managed by the shared commitment to pursue top science. Supervision in the individual groups appears to be relatively well organized and no suggestions for improvement are suggested by the panel.

The RC has been able to attract high levels of internationally competitive funding like ERC advanced grants. Their external funding is thus in very good shape and could hardly be better.

The plans of the RC for future collaboration and research are convincing and exciting, and the panel is unanimously convinced that success will be achieved also in the future.

3 Appendices

- A. Original evaluation material
 - a. Registration material Stage 1
 - b. Answers to evaluation questions Stage 2
 - c. List of publications
 - d. List of other scientific activities
- B. Bibliometric analyses
 - a. Analysis provided by CWTS/University of Leiden
 - b. Analysis provided by Helsinki University Library (66 RCs)



International evaluation of research and doctoral training at the University of Helsinki 2005-2010

RC-SPECIFIC MATERIAL FOR THE PEER REVIEW

NAME OF THE RESEARCHER COMMUNITY: Cancer Systems Biology RC (CSB)

LEADER OF THE RESEARCHER COMMUNITY: Academy Professor Lauri Aaltonen, Research Programs Unit, Faculty of medicine

RC-SPECIFIC MATERIAL FOR THE PEER REVIEW:

- Material submitted by the RC at stages 1 and 2 of the evaluation
 STAGE 1 material: RC's registration form (incl. list of RC participants in an excel table)
 STAGE 2 material: RC's answers to evaluation questions
- TUHAT compilations of the RC members' publications 1.1.2005-31.12.2010
- TUHAT compilations of the RC members' other scientific activities 1.1.2005-31.12.2010
- Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010 (analysis carried out by CWTS, Leiden University)

NB! Since Web of Science(WoS)-based bibliometrics does not provide representative results for most RCs representing humanities, social sciences and computer sciences, the publications of these RCs will be analyzed by the UH Library (results available by the end of June, 2011)



RC-SPECIFIC STAGE 1 MATERIAL (registration form)

1 Responsible person

Name: Aaltonen, Lauri E-mail: Phone: 09-19125595 Affiliation: Academy Professor Street address: Haartmaninkatu 8

2 DESCRIPTION OF THE PARTICIPATING RESEARCHER COMMUNITY (RC)

Name of the participating RC (max. 30 characters): Cancer Systems Biology RC

Acronym for the participating RC (max. 10 characters): CSB

Description of the operational basis in 2005-2010 (eg. research collaboration, joint doctoral training activities) on which the RC was formed (MAX. 2200 characters with spaces): The practical motivation for forming the research community (RC) originates from the vast methodological progress in genomics. The availability of multiple high-throughput platforms has resulted in massive accumulation of multidimensional and heterogeneous molecular data , which needs to be interpreted with approaches requiring wide range of expertise from genetics to biochemisty and computer science. Thus the Aaltonen / Tumor Genomics Group has formed a strong partnership with the Taipale / Medical Systems Biology Group and Hautaniemi / Computational Systems Biology Group to achieve a multidisclipinary community that focuses on gaining comprehensive understanding of cancer progression and identifying targets for effective anti-cancer therapeutics.

The three groups have strong history as working within the same unit within the University of Helsinki; Genome-Scale Biology research program / Research program Unit of the UH Faculty of Medicine. The three groups also have a common future, not only as part of the research program but the three PIs together with two others form the proposed Academy of Finland's Center of Excellence in Cancer Genetics; this proposal has progressed to the shortlist of 36 candidate CoEs with maximum scores.

Like any scientifically strong collaboration, the RC has significantly supported the doctoral training in the three participating groups. While all three PIs have had a significant impact for UH doctoral training e.g. by serving in Graduate School Boards - Biomedical Graduate School, Helsinki Graduate School in Biotechnology and Molecular Biology, and Finnish National Graduate School in Computational Biology - the impact of the RC to internal doctoral training is also an important focus of this review. Here the simple and strong scientific synergy has led to multidisclipinary joint efforts and joint publications, forming a solid basis to prize-winning PhD thesis works originating from this RC.

3 SCIENTIFIC FIELDS OF THE RC

Main scientific field of the RC's research: medicine, biomedicine and healt sciences

RC's scientific subfield 1: Genetics and Heredity



RC-SPECIFIC STAGE 1 MATERIAL (registration form)

RC's scientific subfield 2: Medical Informatics

RC's scientific subfield 3: Oncology

RC's scientific subfield 4: --Select--

Other, if not in the list:

4 RC's PARTICIPATION CATEGORY

Participation category: 1. Research of the participating community represents the international cutting edge in its field

Justification for the selected participation category (MAX. 2200 characters with spaces): Simply a glance at the list of publications should be sufficient to conclude that the "cutting edge research" category best fits our RC. The many key discoveries made within this RC including identification of multiple cancer predisposition genes and innovative use of high throughput technologies and computational methods leave little room for other choices.

2 of 3 PIs are Academy professors – the highest rank in the Finnish research hierarchy - and have been selected for an ERC advanced grant which is probably the most competed funding instrument directed to single PIs. One could continue the justification with many other merits but clearly of the five available categories only number one, "cutting edge research", would be an appropriate choice and any other choice would appear odd.

5 DESCRIPTION OF THE RC'S RESEARCH AND DOCTORAL TRAINING

Public description of the RC's research and doctoral training (MAX. 2200 characters with spaces): Cancer is a disease involving two unique genomes – germline, and that of the respective tumor. The rapid advances in genomic technologies are now enabling whole genome analysis of individuals and cancers. This will finally allow thorough dissection of germline and somatic genetic variation contributing to neoplasia. The RC has made seminal discoveries in this field on the highest international level, thriving on the powerful synergistic combination of advancing technologies, unique national materials and infrastructure, and sophisticated data analyses.

This multidisciplinary RC integrates clinical characteristics and molecular epidemiology of cancer predisposition into genomics and medical and computational systems biology. Within the RC the participating groups have invested heavily to ensure that the young PhD students get the best possible supervision. Often two supervisors have been appointed for a thesis project, the PI and one of the other senior team members. This has ensured that also day-to-day hands-on supervision is available to all students. The RC groups have also been extremely supportive towards courses, visits, and similar activities which enable the students to gain a broader understanding of the field, and science in general.

The three PIs in the RC are from different backgrounds (medicine, biology, engineering). This, together with the interdisciplinary nature of the consortium results in formation of supportive, creative and inspirational research environment that enables both top-level research, and training of a new generation of scientists who will have the necessary skills to excel in their future careers and contribute to the ongoing transformation of biomedicine from largely qualitative and descriptive approaches towards more exact,



<u>RC-SPECIFIC STAGE 1 MATERIAL</u> (registration form)

quantitative and predictive science. The Aaltonen laboratory received the UH's Occupational Safety and Health Award 2005 as a formal acknowledgement of excellent working atmosphere, and this reflects the spirit of the whole RC.

Significance of the RC's research and doctoral training for the University of Helsinki (MAX. 2200 characters with spaces): The core contribution of the RC's research – for UH and others - is its impact into scientific progress. This is best documented by the high quality publications the PIs have coauthored during the evaluation period; please see the lists of publications. Additional prestige for the University has been provided through the 5EU fwp grants coordinated by the PIs during this time window, many science prizes, and in particular the two ERC advanced grants won by the RC PIs.

In doctoral training the RC PIs have supervised 24 PhD thesis projects. The results have been excellent; e.g. UH Medical Faculty thesis prize 1999, U of Oulu Medical Faculty thesis prize 2007, the UH thesis prize 2004 and 2008. Several thesis works have been approved with distinction, by far exceeding the average <10% receiving this mark. The CoE PIs are represented in Boards of all the relevant Graduate Schools; Helsinki Biomedical Graduate School, Helsinki Graduate School in Biotechnology and Molecular Biology, and Finnish National Graduate School in Computational Biology. Thus the commitment of the RC to the curricula of the Graduate Schools is firm and the link between the RC and the Graduate Schools couldn't be tighter.

The groups in this RC are active in technology transfer, and, for instance, computational system biology group has hosted three PhD students from Italy, Norway and Finland in 2010 alone and has sent students to Netherlands and MIT for longer research visits. The goal for these students was to learn to use and apply herein developed computational tools to their data. Bilateral research exchange is significant to Univ. of Helsinki in pragmatic view of getting opportunities for our students to collaborate with cutting-edge groups as well as branding view of demonstrating that research conducted in the UH attracts the scientific community.

Keywords: cancer, predisposition, familial, systems biology, regulatory, bioinformatics, molecular epidemiology

6 QUALITY OF RC'S RESEARCH AND DOCTORAL TRAINING

Justified estimate of the quality of the RC's research and doctoral training at national and international level during 2005-2010 (MAX. 2200 characters with spaces): The RC represents scientific stature in research, which is second to few in Finland. This is exemplified by some of our first/last author papers 2005-10: Hallikas O, et al. Genome-wide prediction of mammalian enhancers based on high-throughput analysis of transcription factor binding affinity. Cell 124, 47-59, 2006. Vierimaa O, et al. Pituitary adenoma predisposition caused by germline mutations in the AIP gene. Science 312, 1228-1230, 2006. Björklund M, et al. Identification of pathways regulating cell size and cell cycle progression by RNAi. Nature 439, 1009-1013, 2006. Varjosalo M, et al. Application of active and kinase-deficient kinome collection for identification of kinases regulating Hedgehog signaling. Cell 133, 537-548, 2008. Wu J, et al. Integrated network analysis platform for protein-protein interactions. Nature Methods 6, 75-7, 2009 Tuupanen S, et al. The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. Nature Genetics 41, 885-890, 2009.



RC-SPECIFIC STAGE 1 MATERIAL (registration form)

Our track record in researcher training is excellent, with 24 thesis works supervised, 10 of which have been approved by distinction. The 2005-2010 results have been outstanding; 17 completed thesis work, 8 of which have been approved with distinction, and e.g. U of Oulu Medical Faculty thesis prize 2007, the best bioinformatics/systems biology PhD thesis of the year 2008 in Finland by the Finnish Bioinformatics Society, and the UH thesis prize 2008.

The RC has been active as organiser of courses and other forms of training, such as graduate school activities: Aaltonen being in the Board of the Helsinki Biomedical Graduate School and serving as a tutor for the MD PhD program, U of Helsinki; Taipale being in the boards of Graduate Schools in Biotechnology and Molecular Biology and Computational biology. Hautaniemi is active in teaching computational methods to new generations of bioinformaticians as well as to biologists, and has organized two seminar courses and two lecture courses on using computational methods in biological data in the past three years.

Comments on how the RC's scientific productivity and doctoral training should be evaluated (MAX. 2200 characters with spaces): 1. Quality of publications

- 2. Prizes won by PIs
- 3. Success in deriving competed funding
- 4. Success in competing for positions
- 5. Number and quality of PhD thesis
- 6. Prizes and distinctions won by the PhD students

RC's publication strategy is to publish the work in high quality journals as with as little delay as possible. Data is also shared before publication in some of the consortia, in particular COGENT (aim to take genomewide association studies forward as a multicenter international consortium) and SYNERGY (Systems approach to gene regulation biology through nuclear receptors). Communicating key results to the public after successful scientific peer-review has also been important. This, however, does not mean that the materials, methods and results would not be discussed before the publication. The students in this RC are encouraged to actively discuss and present on-going work to fellow students in the two other labs. The fact that Aaltonen, Hautaniemi and Taipale labs are physically next to each other facilitates significantly collaboration between the groups.

LIST OF RC MEMBERS

NAM	E OF THE RESEARCHER COI	MMUNITY:	Cancer System	s Biology RC	
RC-LEADER		L. Aaltonen	-		
Category		1			
,			PI-status (TUHAT,	Title of research and	
	Last name	First name	29.11.2010)	teaching personnel	Affiliation
1	Aaltonen	Lauri	X	professor	Faculty of medicine/Research Programs Unit
2	Aavikko	Mervi		doctoral candidate	Faculty of medicine/Research Programs Unit
3	Alhopuro	Pia		postdoctoral researcher	Faculty of medicine/Research Programs Unit
4	Gylfe	Alexandra		doctoral candidate	Faculty of medicine/Research Programs Unit
5	Heliövaara	Elina		doctoral candidate	Faculty of medicine/Research Programs Unit
6	Kaasinen	Eevi		doctoral candidate	Faculty of medicine/Research Programs Unit
7	Karhu	Auli		senior researcher	Faculty of medicine/Research Programs Unit
8	Kondelin (os. Sirkiä)	Johanna		doctoral candidate	Faculty of medicine/Research Programs Unit
9	Launonen	Virpi	X	senior researcher	Faculty of medicine/Research Programs Unit
10	Lehtonen	Heli		postdoctoral researcher	Faculty of medicine/Research Programs Unit
11	Mehine	Miika		doctoral candidate	Faculty of medicine/Research Programs Unit
12	Niittymäki	lina		doctoral candidate	Faculty of medicine/Research Programs Unit
13	Saarinen	Silva		doctoral candidate	Faculty of medicine/Research Programs Unit
14	Tuupanen	Sari		postdoctoral researcher	Faculty of medicine/Research Programs Unit
15	Vahteristo	Pia		senior researcher	Faculty of medicine/Research Programs Unit
16	Hautaniemi	Sampsa	X	research director	Faculty of Medicine/Research Programs Unit
17	Chen	Ping		doctoral candidate	Faculty of Medicine/Research Programs Unit
18	Karinen	Sirkku		doctoral candidate	Faculty of Medicine/Research Programs Unit
19	Laakso	Marko		doctoral candidate	Faculty of Medicine/Research Programs Unit
20	Lahesmaa-Korpinen	Anna-Maria		doctoral candidate	Faculty of Medicine/Research Programs Unit
21	Louhimo	Riku		doctoral candidate	Faculty of Medicine/Research Programs Unit
22	Núñez Fontarnau	Javier		postdoctoral researcher	Faculty of medicine/Research Programs Unit
23	Ovaska	Kristian		doctoral candidate	Faculty of Medicine/Research Programs Unit
24	Rantanen	Ville		doctoral candidate	Faculty of Medicine/Research Programs Unit
25	Rogojin	Vladimir		postdoctoral researcher	Faculty of Medicine/Research Programs Unit
26	Wu	Jianmin		postdoctoral researcher	Faculty of Medicine/Research Programs Unit
27	Taipale	Jussi	X	professor	Faculty of Medicine/Research Prgrams Unit
28	Bonke	Albertus		postdoctoral researcher	Faculty of Medicine/Research Prgrams Unit
29	Hallikas	Outi		postdoctoral researcher	Faculty of Medicine/Research Prgrams Unit
30	Jolma	Arttu		doctoral candidate	Faculty of Medicine/Research Prgrams Unit
31	Li	Song-Ping		postdoctoral researcher	Faculty of Medicine/Research Prgrams Unit
32	Sokolova	Maria		postdoctoral researcher	Faculty of Medicine/Research Prgrams Unit
33	Turunen	Mikko		doctoral candidate	Faculty of Medicine/Research Prgrams Unit
34	Vähärautio (os. Saramäki)	Anna		senior researcher (academy research fellow)	Faculty of Medicine/Research Prgrams Unit
35	Wei	Gonghong		postdoctoral researcher	Faculty of Medicine/Research Prgrams Unit
36	Yan	Jian		doctoral candidate	Faculty of Medicine/Research Prgrams Unit



RC-SPECIFIC STAGE 2 MATERIAL

BACKGROUND INFORMATION

Name of the RC's responsible person: Aaltonen, Lauri

E-mail of the RC's responsible person:

Name and acronym of the participating RC: Cancer Systems Biology, CSB

The RC's research represents the following key focus area of UH: 2. Elämän perusrakenne – The basic structure of life

Comments for selecting/not selecting the key focus area: This RC represents a coalition of three active, world-class research groups, whose main focus is to characterize genetic mechanisms of human cancer predisposition and progression using a holistic, systems biology approach. The published results of the RC members have revealed a number of important and novel discoveries on genetics and systems biology, which belong to the key focus area of "2. The basic structure of life" by the University of Helsinki strategy. Furthermore, Jussi Taipale and Lauri Aaltonen are members of one of the Centers of Excellence listed in the strategy plan at this focus area, and our new Center of Excellence (CoE) proposal "Cancer Genetics" (now shortlisted for final decision) which involves all the three RC groups (Aaltonen, Taipale & Hautaniemi) clearly falls under this focus area as well.

1 FOCUS AND QUALITY OF RC'S RESEARCH (MAX. 8800 CHARACTERS WITH SPACES)

Description of the RC's research focus, the quality of the RC's research (incl. key research questions and results) and the scientific significance of the RC's research for the research field(s).

The RC's research focus

The focus of the Cancer Systems Biology (CSB) RC is to examine cancer patient samples - carefully selected from the Finnish population - using state-of-the-art technologies to comprehensively characterize the genetic profiles and their connections to cancer using holistic systems biology approach. Cancer is a disease involving two unique genomes; germline, and that of the respective tumor. These genomes can now be read using high throughput analysis technologies. This allows a thorough dissection of germline and somatic genetic variation contributing to neoplasia. Accordingly, systematic large-scale efforts to characterize tumor genomes are underway under the umbrella of the International Cancer Genome Consortium (ICGC) and elsewhere. The members of RC are contributors in these efforts. Key discoveries are anticipated in the following fields of research relevant for CSB Research Community (for more details see section 8):

i) Identification of high/moderate-penetrance cancer predisposition conditions, and the respective susceptibility genes.

ii) Characterization of common cancer susceptibility variants. In particular, little is known about the biology underlying these variants – starting from identification of the causative variants from the associated genomic regions.

iii) Characterization of mutatomes of all clinically relevant tumor types; in CSB colorectal cancer and uterine leiomyomas are in focus.

The quality and the scientific significance of the RC's research

Genetics of cancer is a key field of the medical research, in which Finland and the members of the CSB have excellent traditions in advancing the field at the highest international level. CSB thrives on the



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powerful synergistic combination of advancing technologies, unique national materials and infrastructure, and sophisticated data analyses, to take human cancer genetics research to a new level, and finally to translate the molecular findings into clinical benefits, such as novel approaches in risk prediction, prevention, diagnosis, and treatment.

CSB represents scientific stature in health research, which is second to few or none in Finland. The three groups in CSB have made seminal discoveries in the field of cancer research on the highest international level. This is best documented by the high quality publications he PIs have published during the evaluation period. Additional prestige for the University has been provided through the 4 EU grants coordinated by the PIs during this time window, many science prizes, and in particular the two ERC Advanced Grants won by the RC PIs (Taipale 2008 Aaltonen 2010).

Some of our last author papers 2005-2010:

Hallikas O, et al. Genome-wide prediction of mammalian enhancers based on high-throughput analysis of transcription factor binding affinity. Cell 124, 47-59, 2006.

Vierimaa O, et al. Pituitary adenoma predisposition caused by germline mutations in the AIP gene. Science 312, 1228-1230, 2006.

Björklund M, et al. Identification of pathways regulating cell size and cell cycle progression by RNAi. Nature 439, 1009-1013, 2006.

Varjosalo M, et al. Application of active and kinase-deficient kinome collection for identification of kinases regulating Hedgehog signaling. Cell 133, 537-548, 2008.

Wu J, et al. Integrated network analysis platform for protein-protein interactions. Nature Methods 6, 75-77, 2009.

Tuupanen S, et al. The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. Nature Genetics 41, 885-890, 2009.

A brief account on the Groups:

Tumor genomics group / Aaltonen (CSB coordinator) Aaltonen, is a second term Academy Professor; received this position in 2002 at the age of 39. The main current administrative positions; Director, Genome-Scale Biology research program, University of Helsinki & Director, Biocentrum Helsinki. Publications have been cited >17.500 times. Of other merits e.g. the EMBO membership in 2000, Anders Jahre Prize for Young Researchers 2000, being a nominee for the Descartes prize 2005 with Prof. Tomlinson and ERC Adv Grant 2010, are useful indicators. The core field of research is tumor genomics, in particular molecular basis of cancer susceptibility. The track record in cancer genetics speaks for itself.

Medical systems biology group / Taipale Taipale holds a joint appointment as an Academy of Finland Professor (40%) and Professor of Medical Systems Biology at the Karolinska Institutet, Sweden (60%). Focus of the Taipale group is on systems biology of growth control and cancer. Prof. Taipale has published 40 articles of which nine are in the most prestigious scientific journals (Nature, Science, Cell). He has won numerous awards and grants (e.g. Anders Jahre Prize for Young Researchers, EMBO Young Investigator, ERC Advanced Grant), and is internationally recognized as a leader in the field of genomics and systems biology. The Taipale group brings to the RC expertise on high-throughput screening using cDNA and RNA interference, and computational and experimental methods to identify causative regulatory mutations in non-protein coding DNA and to analyze genetic networks. In addition, Taipale group has extensive expertise on mouse models of gene and regulatory region function. See publications.



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Computational systems biology group / Hautaniemi Hautaniemi is an Academy Research Fellow and started his group at spring 2006 after extremely successful post-doc period in the Douglas A. Lauffenburger laboratory at MIT. With 46 publications cited >1300 times and h-index of 18 Hautaniemi is a recognized systems biologist who has served in a number of conference program committees, such as ISMB/ECCB 2009 (the largest systems biology annual meeting), reviewed systems biology grant applications in EU and USA, such as NIH R01 supplement grants, and coordinates national and international bioinformatics/systems biology consortiums. The main objective of the Hautaniemi group is to develop and apply novel computational methodologies to characterize and control cell decision processes in cancer.

• Ways to strengthen the focus and improve the quality of the RC's research.

Cancer is a complex group of diseases and to characterize origin of tumors using high-quality data requires a truly cross-disciplinary team of researchers. This RC brings together expertise in medicine (LA), biochemistry (JT) and engineering (SH), which are the key disciplines enabling systematic and innovative cancer research. A major strength of this RC is that the groups are physically close in the excellent research infrastructure in Biomedicum. CSB is a prime example of multidisciplinary research team that has a clearly defined and innovative vision.

To upgrade our efforts we have now submitted an application for an Academy of Finland's Center of Excellence status, and further broadened our approach by including computer scientists to facilitate sequence analysis (Professor Veli Mäkinen), as well as epidemiologists to facilitate patient selection (the unique Finnish Cancer Registry, Professor Timo Hakulinen as Director). This application has been shortlisted for final funding decisions (36 out of 135 proposals were shortlisted after international review), expected in June 2011.

2 PRACTISES AND QUALITY OF DOCTORAL TRAINING (MAX. 8800 CHARACTERS WITH SPACES)

 How is doctoral training organised in the RC? Description of the RC's principles for recruitment and selection of doctoral candidates, supervision of doctoral candidates, collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes, good practises and quality assurance in doctoral training, and assuring good career perspectives for the doctoral candidates/fresh doctorates.

Recruitment and selection of doctoral candidates.

Our basic principle of doctoral training we provide is that excellence in research and hands-on supervision attracts the best students from Finland and abroad, and results in very competitive PhDs who can continue conducting high impact research in academia or industry. We are open for applications year round and the background of the students vary from pure mathematics to medicine and the procedure of handling the applications varies. For students already enrolled to the MD PhD program of the medical faculty the procedure proceeds as follows. The MD PhD Program is a joint program between the Biomedical Graduate School, Faculty of Medicine and other Meilahti Campus Graduate Schools. The students are selected amongst the first year MD students, and they start their graduate education by doing rotations in research groups during their first two summers, including one clinical rotation. The CSB almost always can provide a rotation slot for an interested student. Following this the students choose their research group and thesis project and continue doing their research side by side with their medical studies for approximately three years; however, the prerequisite for this step is that the student's performance during the CSB rotation period has met our standards, in view of scientific potential as well as ability for team work. Having finished their MD degree, the students continue with full-time research with their thesis project until the defense of their thesis. The selection of students from other fields, such as biology, mathematics and computer science, is performed based



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on transcripts, interview and recommendations of the applicants. In the interview there is always at least one post-doc and PhD student included. The number of applications we receive per year is around 300. Of these, on average 30 are invited to an interview and around 20 are hired.

Supervision of doctoral candidates.

A doctoral student is exposed to the four essential fields of research (research, grant application writing, peer-review, teaching) in a controlled and gradual manner. During the first two years, a PhD student is expected to accomplish studies and the research projects are clearly defined. Projects are supervised by late-stage PhD students and post-docs in addition to PI. After successfully contributing two projects, the independence level of the student is increased with delineating the objective of the research and expecting him/her to suggest a plan how to achieve this goal. The plan is discussed with PI and post-docs and after approval, the student starts executing the plan. Simultaneously, student is exposed to peerreview journal paper the PI is asked to review. While PI writes the review, the student writes his/her own critique and the points of the paper are discussed together; confidentiality requirements of peerreview are strictly adhered to. This way our students get a better understanding of the scientific peerreview procedure, and can plan their own papers accordingly. The students are also encouraged to apply for personal grants, as well as contribute small portions of large EU grant proposals/reports the PIs are writing. Exposure to grant writing and reporting helps students to express their results and plans in a clear fashion, which facilitates dissemination of the results to public and newspapers/magazines. In addition, students are expected to teach at least in one graduate school course, or a course or lecture at the undergraduate level. This way we expect to train senior researchers who have both a strong publication record as well as solid skills in teaching and grant writing, to be able to successfully compete for tenure track positions in the UH and elsewhere; currently the most difficult bottleneck in research careers.

Postdoctoral training During post-doctoral fellowship one should learn the skills required to become a professional scientist, with a clear, independent research interest and direction. The only way of learning to become independent is to have responsibility and say in one's own projects. It is also critical to learn how to direct students, and teach at the university level. These skills are also required for receiving a docentship, a key requirement for career advancement, and an important aim of a successful post-doc period. Therefore, after 1-1.5 years, when the post-doctoral scientist is familiar with the research topic, and has developed scientific self-confidence, s/he will also take the responsibility of directing one or two graduate students.

Collaboration with faculties, departments/institutes, and potential graduate schools/doctoral programmes.

Given the multidisciplinary nature of our RC, we contribute to several graduate schools and doctoral programs at levels from lecturing to board memberships. The CSB PIs have supervised more than 50 PhD thesis projects. The results have been excellent; e.g. UH Medical Faculty thesis prize 1999, U of Oulu Medical Faculty thesis prize 2007, best population sciences thesis 2006, the UH thesis prize 2004 and 2008. Several thesis works have been approved with distinction, by far exceeding the average <10% receiving this mark. The CSB PIs are represented in Boards of all the relevant Graduate Schools; Helsinki Biomedical Graduate School, Helsinki Graduate School in Biotechnology and Molecular Biology, and Finnish National Graduate School in Computational Biology. The PIs are also active in teaching genetics, bioinformatics and systems biology to MD and TRANSMED (MSc program in translation medicine) students. Thus the commitment of the CSB to the curricula of the Graduate Schools is firm and the link between the CSB and the Graduate Schools couldn't be tighter.

The PIs of CSB are also closely collaborating with Faculty of Science (Kumpula campus), and Institute Biotechnology (Viikki campus) in addition to obviously close relations to Faculty of Medicine and HUCH



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in Meilahti. While our collaboration is focused on research, we also teach frequently in the other campuses and a large portion of our summer interns are from Kumpula or Viikki campuses.

Good practices and quality assurance in doctoral training.

Typically each student in CSB has two PI-level supervisors and PhD committee comprising 2-3 PI-level researchers. The committee gets together at least once in a year and the student explains his/her research and results. This assures that external but skilled panel of researchers comments periodically students' progress. While students in the CSB have day-to-day hands-on guidance from post-docs, the students meet regularly with PIs and discuss plans, progress and results. This ensures that quality of the research remains high and possible issues can be solved early so that they do not grow to a crisis. As an indication of high quality and good practice in doctoral training is that the Aaltonen lab received the UH's Occupational Safety and Health Award 2005 as a formal acknowledgement of excellent working atmosphere.

Assuring good career perspectives for the doctoral candidates/fresh doctorates.

We strongly believe that the best possible prospect for a PhD or post-doc stem from excellent research results, experience in supervising students and contributing to grant applications. The most important from these is research as a typical career option for researchers graduating from CSB is academia or research and development in industry. We have been extremely successful in educating generations of researchers who have landed excellent post-doc and PI positions in academia as well as high R&D positions in industry in Finland and abroad.

• RC's strengths and challenges related to the practises and quality of doctoral training, and the actions planned for their development.

The major challenge for the holistic cancer research is getting talented students into research groups. Given multidisciplinary nature of our research, it is not enough to master one's own field but also have at least some understanding on the basic concepts of the other, relevant fields. It is increasingly difficult to recruit students and post-docs who have strong skills in their own field and who are open to cross-disciplinary research. Our actions have been to be active in both teaching in the curriculum cross-disciplinary topics, as well as supervising students so that they experience cross-disciplinary research from the beginning of their career. However, we feel that it is important that the University clearly commits to support cross-disciplinary research and education not just in speeches but in practice with clear directed funding for these activities. Equally well we expect our proposal for an Academy of Finland's Center of Excellence to be funded, to facilitate the further broadening of our approaches by adding computer sciences and epidemiology to the consortium.

3 SOCIETAL IMPACT OF RESEARCH AND DOCTORAL TRAINING (MAX. 4400 CHARACTERS WITH SPACES)

• Description of how the RC interacts with and contributes to the society (collaboration with public, private and/or 3rd sector).

The societal impact of CSB relates mostly to expected advancements in management of cancer and cancer predisposition. We actively seek for opportunities to promote well-being in Finland and elsewhere, whether in form of breakthroughs of scientific, clinical, or commercial value. Of note, in particular prevention of cancer in predisposed individuals is an exceptionally valuable goal as the benefits focus on individuals on their most productive period of life, and already the results thus far achieved are world-leading with many lives saved. This greatly increases the impact of our research on health and wellbeing. It is also important to see that the data analysis platforms and approaches developed by the CSB are not only valid for cancer research, but in many cases also for medical genetics in general. The CSB thus is an important player in ensuring that the proud traditions of this most



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successful field of Finnish medical research are maintained, and the discipline continues to flourish on the highest international level. We have also been active in advising policy makers. Examples include active contributions in the preparations for the Academy law 2009 and Biobank legislation 2010, including several personal discussions with members of the parliament and Government Ministers. Aaltonen's recent nominations as the Director of one of the Finnish Biocenters Biocentrum Helsinki, will position CSB well in the National network of policymaking in biosciences. Another prominent forum where CSB is represented by Aaltonen is the Advisory Board on Biotechnology (Biotekniikan neuvottelukunta). This is a consultative body of experts in issues related to bio- and gene technology appointed by the Government for a term of three years. It aims to promote cooperation between authorities, researchers and operators in biotechnology and in particular gene technology, and it monitors developments and research in gene technology as well as its health and environmental impacts. In addition, it is the Board's task to monitor and promote international cooperation in biotechnology and take into account the ethical considerations of gene technology.

The dialog with laymen as well as professionals about the possibilities and risks in genetics is a field where CSB indeed has proud traditions, to be maintained.

In this environment our doctoral student learn the ways to promote societal impact of research in their own work. High-quality researcher training on its own of course has significant societal impact, in accordance to the National strategies. Finland as a country of knowledge could not thrive without doctoral training programs aiming at – and reaching – levels of expertise comparable to the very best similar programs worldwide.

• Ways to strengthen the societal impact of the RC's research and doctoral training.

Continuing excellence in field that has a major impact on public health is the key in strengthening the impact of our RC to society. We are already active in public forums (Vieraskynä column and interviews to the major newspaper Helsingin Sanomat, and other similar forums such as Aamulehti) as well as contributions to specialized magazines, such as Duodecim, which is subscribed by almost every Finnish medical doctor. For instance, SH has contributed to two articles to Duodecim on systems biology and computational analysis of cancer, as well as interviewed as the Colleague of the Week. Outreach, i.e., our efforts to contribute the discussion with "layman", high-school students and specialists is an important aspect of research and promote both University of Helsinki in particular and excellent cross-disciplinary research in general. We shall continue our efforts to support our students in their future careers, in Finland and elsewhere. As with other items in this review, one important way to step up CSB efforts is the upgrade to a Center of Excellence; proposal submitted 10/2010.

4 INTERNATIONAL AND NATIONAL (INCL. INTERSECTORAL) RESEARCH COLLABORATION AND RESEARCHER MOBILITY (MAX. 4400 CHARACTERS WITH SPACES)

• Description of the RC's research collaborations and joint doctoral training activities and how the RC has promoted researcher mobility.

The RC's research collaborations and joint doctoral training activities.

CSB as well as the research problem it is tackling are multidisciplinary at core, integrating medical genetics and molecular epidemiology (Aaltonen) with systems biology research in biomedicine (Taipale) and bioinformatics (Hautaniemi). All the PIs are from different backgrounds, are highly ambitious, energetic and at an active career stage; the passion for breakthrough science is strongly present. This, together with the interdisciplinary nature of the consortium results in formation of supportive, creative and inspirational research environment that enables both top-level research, and training of a new generation of scientists who will have the necessary skills to excel in their future careers and contribute



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to the ongoing transformation of biomedicine from largely qualitative and descriptive approaches towards more exact, quantitative and predictive science. Regular meetings, at least twice per year on the whole CSB level, are seen as an important tool for evaluating past achievements and problems, exchanging ideas and experiences and planning for the future.

An excellent and objective measure of networking nationally and internationally is the number and spectrum of coauthors in papers of the CSB PIs; please see the respective lists of publications.

The members of this CSB have extensive national collaboration network. In bioinformatics and systems biology close collaboration with HIIT (Helsinki Institute for Information Technology; our collaborator Mäkinen as member), FIMM (Taipale, Aaltonen as associate members) and Biocenter Finland Bioinformatics Infrastructure Network (Hautaniemi as chairman) together with personal collaborations mean that all Finnish major groups who develop and apply computational methods to biomedical data are periodically met by the members of the CSB. This ensures that methods that are possibly provided by other groups in Finland and could be applicable in CSB can be deployed rapidly. Biocentrum Helsinki is an umberella organization supporting the networking of the best biotechnology groups in UH and Aalto University (currently 29 groups, all selected after peer review), and Aaltonen, Taipale, and Hautaniemi groups all are members of this key local organization, Aaltonen being the newly appointed Director of this organization. Clinical team members in CSB research and medical genetics units from all university hospitals are valuable collaborators for our nation-wide efforts.

How the RC has promoted researcher mobility.

Collaboration and mobility are ones of the strengths of CSB. We strongly encourage our PhD students to spend at least three months in a laboratory abroad. For instance, students from CSB have conducted research visits in MIT (USA), Nijgemen (Netherlands), KI (Sweden) and Oxford (UK), to mention a few. Furthermore, the PIs of this RC are mobile and frequent speakers in conferences and workshops. This brings forward novel collaboration possibilities with the other leading scientists. Indeed, this is seen as a number of EU, ERC and NIH projects we are committed and coordinating.

RC's strengths and challenges related to research collaboration and researcher mobility, and the
actions planned for their development.

In studies on cancer genetics the proposed CSB is second to few or none in the world. It has thus been easy for us to establish the necessary international collaborations. In systems biology SH coordinates a FP7 ERANET SysBio consortium SYNERGY that contains partners from four EU countries, and JT a FP7 project SYSCOL that has 11 partners from nine countries. In somatic colon cancer genetics CSB is in key position as part of the ICGC (official affiliation pending) effort to characterize colorectal cancer through genomic sequencing and other omics platforms. This consortium which was initiated by the CSB coordinator includes the top CRC research groups in the world, Ian Tomlinson (U of Oxford), Victor Velculescu and Bert Vogelstein (Johns Hopkins). In systems biology our major collaborators are Douglas Lauffenburger (MIT), Eran Segal (Weizmann) and Timothy Hughes (U. Toronto). We have not identified major challenges in our research collaboration network and we receive frequently invitations to FP7 and NIH proposals.

5 OPERATIONAL CONDITIONS (MAX. 4400 CHARACTERS WITH SPACES)

• Description of the operational conditions in the RC's research environment (e.g. research infrastructure, balance between research and teaching duties).

The description of research environment can be broken into two; Description of the National infrastructure relevant for the RC, and description of the local research environment.



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1) National infrastructure relevant for CSB

The availability of unique national infrastructures and favorable attitudes and legislation form the basis of this research effort. Two key resources for the future are the Finnish Cancer Registry and the population registry center. Combination of cancer registry data and population registry data, as well as the genetic homogeneity of the population, provides powerful tools for detection of aberrant clustering of cancer cases. Use of resources such as systematic collections of paraffin embedded tissue specimens – particularly useful in cancer research – is enabled by legislation, as long as appropriate authorization has been granted. We have 16 years of experience in collaborating with Finnish pathology departments, and specimens can be found with great accuracy. The centralized national health care system enables also the efficient gathering of relevant patient record data from hospitals, when appropriately authorized.

2) Local research environment

The position of the Biomedicum campus as the most impressive cluster of biomedical research infrastructure in the country is well established. As depicted above the unique materials can be efficiently utilized based on advanced but well-working genome-wide tools such as high throughput sequencing, SNP microarrays on normal as well as tumor DNA, CGH array on normal and tumor DNA, as well as transcriptome analyses on normal and tumor RNA (being replaced by RNA-sequencing). All these platforms are at routine use within CSB and the required technology is conveniently available through the local core facilities Biomedicum Genomics and FIMM Technology Center. Several other campus core facilities such as Virus Core Facility, Molecular Imaging Unit, Biomedicum High Throughput Center, and Biomedicum Experimental Animal Center may also be utilized providing excellent assistance in carrying out sophisticated functional experiments. The Research and Innovation Services of the University of Helsinki gives high quality support services for the CSB researchers from project planning to proposal preparation, contract negotiation, implementation and exploitation of the results. All the CSB laboratory and office space are located next to each other at the Biomedicum 5th floor enabling easy communication between the groups at all levels.

RC efforts are strongly supported by the University of Helsinki / Medical Faculty as well as the Academy of Finland. All CSB PIs are members of the Faculty's 5-year research program "Genome-Scale Biology" (2007-2011, the coordinator as the current director) and two PIs belong to the Academy of Finland's Center of Excellence in Translational Genome-Scale Biology (2006-2011).

Research and teaching duties

The main focus of the CSB is on highest level research. Teaching focuses on pre- and postgraduate students within the groups, with the aim of providing them the best possible supervision both at the strategic as well as day-to-day hands-on level. Naturally, the senior members of the team are involved in various training activities on the campus and elsewhere.

 RC's strengths and challenges related to operational conditions, and the actions planned for their development.

The biggest strength of the CSB is the well-established collaboration between the three world-leading groups who all have their well-defined roles and expertise that complement each other. The close collaboration will likely continue in the framework of Academy's CoE as well as through Faculty's research program. Physical location next to each other at the leading biomedical research institute in Finland provides excellent means to easily organize collaborative efforts, to utilize cutting edge technologies, and to continue pursuing scientific research at the highest level. A major challenge related to operational conditions is the continuous need to upgrade the supporting research infrastructure. Without expensive state-of-the-art technologies CSB research cannot be competitive, and the



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commitment of the University to our field of research is not only welcome but essential for future success on the highest international level.

6 LEADERSHIP AND MANAGEMENT IN THE RESEARCHER COMMUNITY (MAX. 4400 CHARACTERS WITH SPACES)

Description of the execution and processes of leadership in the RC, how the management-related
responsibilities and roles are distributed in the RC and how the leadership- and management-related
processes support high quality research, collaboration between principal investigators and other
researchers in the RC, the RC's research focus and strengthening of the RC's know-how.

CSB is a research community formed by three independent research groups and as such does not have a particular management structure. Efforts towards scientific breakthroughs dictate the actions, rather than decisions of a governing body or director. All groups are members of Genome-Scale Biology Research Program, where Aaltonen happens to be the director. Similarly, the Center of Excellence proposal which is the key for future actions is coordinated by Aaltonen. Thus it seemed conceivable that Aaltonen also acts as the contact person of the Research Community, but this could have equally well been any other PI. Our mode of administration is network management. The CSB is a focused and simple Research Community with a flat organizational profile; 3 teams with clear roles and strengths, working together.

Within the groups the structure needs to be more organized. Senior scientists have significant roles e.g. in supervising PhD students, organizing day-to-day work in the laboratories and participating in writing the grant applications and reports. This enables efficient advancement of the projects and at the same time is extremely useful for the senior scientists themselves as they gain valuable experience in administrative, tutorial and organizational matters which come most valuable when starting their own and independent research groups.

Collaboration between the three CSB research groups is close at all possible levels. The PIs vision the future strategies and discuss their implementation and study design with senior scientists, who with doctoral students organize and execute them in practice. Also the laboratory personnel frequently asks tips and guidance from other CSB groups who already have experience in specific practical issues. One major advantage is naturally the physical location of all CSB groups next to each other. As people from the three groups see each other at the basically daily basis, they have gotten to know each other rather well, making it very easy to approach members from other groups and also organize meetings often and on a short notice or spontaneously. Relaxed atmosphere and informal relationships between and within the groups also allows and encourages students to express their ideas and suggestions for open discussion; indeed this is what one calls a "creative research environment".

As all three groups are world-leaders in their own field, they naturally have other collaborations also outside the context of CSB. New insights and innovative ideas from these collaborations, naturally within the limit that confidentiality provides, is distributed between the CSB and within the individual research groups. Also students of this RC are encouraged to actively discuss and present their on-going work to fellow students in the other two labs.

• RC's strengths and challenges related to leadership and management, and the actions planned for developing the processes.

An obvious strength is that all actions arise from needs of scientific excellence, and this will not change in the future. In management the RC aims to adopt useful tools such as yearly risk analysis, already in use in the coordinators group with excellent results. This analysis covers issues ranging from ergonomics



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to other aspects of well-being at work, such as stress related issues. Monitoring these allow efficient and early interventions and help in maintaining a pleasant, relaxed, and creative working atmosphere.

A clear challenge is the growing administrative workload of the PIs. To cope with this many tasks have been delegated to the other senior scientists to ensure fluent advancement of the projects. There is a risk that new paperwork related duties are implemented whenever useful from the administrative point of view, with little interest to monitor the cumulative impact of these measures to creative scientific work. We shall continue our efforts to communicate these concerns and observations to the UH governing bodies.

7 EXTERNAL COMPETITIVE FUNDING OF THE RC

- Listing of the RCs external competitive funding, where:
 the funding decisions have been made during 1.1.2005-31.12.2010, and
 the administrator of the funding is/has been the University of Helsinki
- Academy of Finland (AF) total amount of funding (in euros) AF has decided to allocate to the RC members during 1.1.2005-31.12.2010: 4490000
- Finnish Funding Agency for Technology and Innovation (TEKES) total amount of funding (in euros) TEKES has decided to allocate to the RC members during 1.1.2005-31.12.2010: 0
- European Union (EU) total amount of funding (in euros) EU has decided to allocate to the RC members during 1.1.2005-31.12.2010: 2910000
- European Research Council (ERC) total amount of funding (in euros) ERC has decided to allocate to the RC members during 1.1.2005-31.12.2010: 4680000
- International and national foundations names of international and national foundations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
 - names of the foundations: Sigrid Juselius, AICR, Finnish Cancer Organizations
 - total amount of funding (in euros) from the above-mentioned foundations: 2210000
- Other international funding names of other international funding organizations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
 - names of the funding organizations:
 - total amount of funding (in euros) from the above-mentioned funding organizations: 0
- Other national funding (incl. EVO funding and Ministry of Education and Culture funded doctoral programme positions) names of other national funding organizations which have decided to allocate funding to the RC members during 1.1.2005-31.12.2010, and the amount of their funding (in euros).
 - names of the funding organizations: EVO, Ministry of Education, U of Helsinki, Biocentrum Helsinki, VTT, CIMO, FIMM
 - total amount of funding (in euros) from the above-mentioned funding organizations: 2910000



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8 RC's strategic action plan for 2011–2013 (MAX. 4400 CHARACTERS WITH SPACES)

Description of the RC's future perspectives in respect to research and doctoral training.

As indicated above, key discoveries are anticipated in the following fields of research relevant for CSB Research Community:

i) Identification of high/moderate-penetrance cancer predisposition conditions, and the respective susceptibility genes.

ii) Characterization of common cancer susceptibility variants. In particular, little is known about the biology underlying these variants – starting from identification of the causative variants from the associated genomic regions.

iii) Characterization of mutatomes of all clinically relevant tumor types.

To discover high/moderate penetrance loci (i) our main tool will be the Finnish Cancer Registry (FCR). The combined use of the National Population Registries (NPR) and FCR will enable systematic detection of clusters of specific types of cancer based on geographical clustering and clustering by family name at birth. This resource of 1 000 000 cancer probands will provide us with a significant advantage as compared with competitors, in recruiting key patients, as well as in subsequent stages in validation such as detailed characterization of new susceptibility phenotypes. For colorectal cancer (CRC) we are in good position with already existing materials and we shall create whole genome data from our familial CRC patient materials, to discover predisposition changes in particular in genomic regions predicted to function in gene regulation.

For characterization of common cancer susceptibility variants (ii) we will focus on CRC, where our key collaborator in view of GWAS data is EU COGENT consortium. COGENT has created data on 50 000 CRC cases and 50 000 cancer free controls to identify the key loci predisposing to common CRC. Such data is often descriptive, and much work needs to be done before the causative changes are identified and mechanisms of disease unraveled. The associated regions will be scrutinized in CSB through our innovative in-silico and experimental approaches as exemplified in our recent article (Tuupanen et al, Nature Genetics 2009).

The CSB will contribute to systematic analysis of tumor mutatomes (iii) focusing on CRC and uterine leiomyomas. The number of well documented CRC/normal tissue fresh sample pairs in our unit exceeds 1500. 200 fresh frozen uterine leiomyoma/normal myometrium pairs are available. These efforts will result in a comprehensive catalogue of somatic mutations in a large number of tumors.

CSB scrutinizes patient samples by genome-wide approaches, such as genomic sequencing, RNA-seq, and copy number variation arrays. Advanced computer science methods are used to translate the raw data from these approaches into knowledge. In addition, sophisticated system biology approaches will be developed and utilized to model the effects of the identified variants: e.g. computational methods, functional approaches, and high-throughput screening methods.

A new CoE proposal (now shortlisted for final decision) which involves all the three RC groups is a key for the future research actions.

CSB aims to continue providing doctoral training on the highest national and international level. The good practices implemented will be carefully maintained and developed.

The major advance would be the promotion of student exchange both within the CSB groups and on an international level. For students with a biological/medical background, this could include getting familiar with computational tools and respectively for bioinformatics/computing students the possibility to work



RC-SPECIFIC STAGE 2 MATERIAL

in a wetlab environment. The CSB network would improve the standard of international visits through the formation of a broader contact network through the group leaders. PhD research training could also be improved by systematically offering students not enrolled in Graduate Schools the possibility for comparable benefits, such as a thesis committee. Regular research discussions or journal clubs creating a relaxed setting where students could practice and improve their scientific thinking are also desired. The CSB network could promote personal development, which is rarely targeted in the currently available programs. The CSB could organize courses related to leadership, networking and presentation skills. A CSB alumni-network could be formed where alumni members of the groups presented their current research and provided career advice to current students in an informal environment.

9 SHORT DESCRIPTION OF HOW THE RC MEMBERS HAVE CONTRIBUTED TO THE COMPILATION OF THE STAGE 2 MATERIALS (MAX. 1100 CHARACTERS WITH SPACES).

The CSB group leaders Aaltonen, Hautaniemi & Taipale first drafted the document in several successive rounds. Subsequently the document was further expanded and polished by postdoctoral members of the RC, in particular Docents Auli Karhu and Pia Vahteristo. The views of the predoctoral students were collected by Anna-Maria Lahesmaa-Korpinen, and all predocs of the RC had the opportunity – and were encouraged to - express their views, and many of them indeed did. Technical assistance was provided by Sirpa Soisalo, Tiia Pelkonen, and Ritva Lautala.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

1 Analysis of publications

 Associated person is one of Lauri Antti Aaltonen , 		Mervi Aavikko,	Pia Pauliina Alhopuro,		
	Alexandra Gylfe,		Elina Heliövaara,	Eevi Kaasinen ,	Auli
Inkeri Karhu,	keri Karhu, Johanna Kondelin,		3	Virpi Launonen ,	Heli
Lehtonen,	Miil	a Mehine,	lina Niittymäki,	Silva Saarinen ,	
Sari Tuupanen,	ari Tuupanen, Pia Marita Vahteristo,		Sampsa Hautar	niemi,	Ping Chen ,
Sirkku Helena Karinen ,		Marko Kalevi Laakso ,	Anna-Maria Kristiina La	Anna-Maria Kristiina Lahesmaa-Korpinen,	
	Riku Lou	himo ,	Kristian Ovaska,	Ville Rantanen,	
Vladimir Rogojin,	Vladimir Rogojin, Jianmin Wu,		Jussi Taipale ,	Albertus Wilhelm Martinus Bonk	
	Outi Hallikas ,	,	Arttu Jolma,	Songping Li,	
Maria Sokolova , Mikko Turunen ,		Mikko Turunen ,	Anna Valpuri Vähär	autio, Gr	onghong Wei,
	Jian Yan				

Publication year

Publication type	2005	2006	2007	2008	2009	2010	Total Count 2005 - 2010
A1 Refereed journal article	24	25	26	21	21	25	142
A2 Review in scientific journal		1	2	2		1	6
A3 Contribution to book/other compilations (refereed)			1		1		2
B1 Unrefereed journal article		4					4
B3 Unrefereed article in conference proceedings						1	1
E1 Popular article, newspaper article				1			1

1



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

2 Listing of publications

A1 Refereed journal article

2005

Alazzouzi, H, Davalos, V, Kokko, A, Domingo, E, Woerner, SM, Wilson, AJ, Konrad, L, Laiho, P, Espin, E, Armengol, M, Imai, K, Yamamoto, H, Mariadason, JM, Gebert, JF, Aaltonen, LA, Schwartz, S, Arango, D **2005**, 'Mechanisms of inactivation of the receptor tyrosine kinase EPHB2 in colorectal tumors', **Cancer Research**, vol 65, no. 22, pp. 10170-10173.

Alazzouzi, H, Alhopuro, P, Salovaara, R, Sammalkorpi, H, Järvinen, HJ, Mecklin, J, Hemminki, A, Schwartz, S, Aaltonen, LA, Arango, D 2005, 'SMAD4 as a prognostic marker in colorectal cancer', Clinical Cancer Research, vol 11, no. 7, pp. 2606-2611.

Alhopuro, P, Alazzouzi, H, Sammalkorpi, H, Davalos, V, Salovaara, R, Hemminki, A, Järvinen, HJ, Mecklin, J, Schwartz, S, Aaltonen, LA, Arango, D 2005, 'SMAD4 levels and response to 5-fluorouracil in colorectal cancer', Clinical Cancer Research, vol 11, no. 17, pp. 6311-6316.

Alhopuro, P, Ylisaukko-oja, S, Koskinen, W, Bono, P, Arola, J, Järvinen, HJ, Mecklin, J, Atula, T, Kontio, R, Mäkitie, A, Suominen, S, Leivo, I, Vahteristo, P, Aaltonen, L, Aaltonen, LA, Suominen, S **2005**, 'The MDM2 promoter polymorphism SNP309T->G and the risk of uterine leiomyosarcoma, colorectal cancer, and squamous cell carcinoma of the head and neck', **Journal of Medical Genetics**, vol 42, no. 9, pp. 694-698.

Alhopuro, P, Katajisto, P, Lehtonen, RJ, Ylisaukko-oja, S, Näätäsaari, L, Karhu, A, Westerman, AM, Wilson, JHP, de Rooij, FWM, Vogel, T, Moeslein, G, Tomlinson, IPM, Aaltonen, LA, Mäkelä, TP, Launonen, V **2005**, 'Mutation analysis of three genes encoding novel LKB1-interacting proteins, BRG1, STRADalpha, and MO25alpha, in Peutz-Jeghers syndrome', **British Journal of Cancer**, vol 92, no. 6, pp. 1126-1129.

Alhopuro, P, Parker, AR, Lehtonen, R, Enholm, S, Järvinen, HJ, Mecklin, J, Karhu, A, Eshleman, JR, Aaltonen, LA **2005**, 'A novel functionally deficient MYH variant in individuals with colorectal adenomatous polyposis', **Human Mutation**, vol 26, no. 4, pp. 393.

Arango, D, Laiho, P, Kokko, A, Alhopuro, P, Sammalkorpi, H, Salovaara, R, Nicorici, D, Hautaniemi, S, Alazzouzi, H, Mecklin, J, Järvinen, HJ, Hemminki, AE, Astola, J, Schwartz, S, Aaltonen, LA 2005, 'Gene-expression profiling predicts recurrence in Dukes' C colorectal cancer', Gastroenterology, vol 129, no. 3, pp. 874-884.

Birkenkamp-Demtröder, K, Olesen, SH, Sørensen, FB, Laurberg, S, Laiho, P, Aaltonen, LA, Ørntoft, TF **2005**, 'Differential gene expression in colon cancer of the caecum versus the sigmoid and rectosigmoid', **Gut**, vol 54, no. 3, pp. 374-384.

Calabrese, P, Mecklin, J, Järvinen, HJ, Aaltonen, LA, Tavare, S, Shibata, D 2005, 'Numbers of mutations to different types of colorectal cancer', BMC Cancer, vol 5, no. 126.

Domingo, E, Niessen, R, Oliveira, C, Alhopuro, P, Moutinho, C, Espin, E, Armengol, M, Sijmons, RH, Kleibeuker, JH, Seruca, R, Aaltonen, LA, Imai, K, Yamamoto, H, Schwartz, S, Hofstra, RMW **2005**, 'BRAF-V600E is not involved in the colorectal tumorigenesis of HNPCC in patients with functional MLH1 and MSH2 genes', **Oncogene**, vol 24, no. 24, pp. 3995-3998.

Eschrich, S, Yang, I, Bloom, G, Kwong, KY, Boulware, D, Cantor, A, Coppola, D, Kruhøffer, M, Ørntoft, TF, Quackenbush, J, Yeatman, TJ, Aaltonen, LA **2005**, 'Molecular staging for survival prediction of colorectal cancer patients', **Journal of Clinical Oncology**, vol 23, no. 15, pp. 3526-3535.

Hampel, H, Stephens, JA, Pukkala, E, Sankila, R, Aaltonen, LA, Mecklin, J, Chapelle, ADL 2005, 'Cancer risk in hereditary nonpolyposis colorectal cancer syndrome: later age of onset', Gastroenterology, vol 129, no. 2, pp. 415-421.

Hienonen, T, Sammalkorpi, H, Enholm, S, Alhopuro, P, Barber, TD, Lehtonen, R, Nupponen, NN, Lehtonen, H, Salovaara, R, Mecklin, J, Järvinen, HJ, Koistinen, R, Arango, D, Launonen, V, Vogelstein, B, Karhu, A, Aaltonen, LA **2005**, 'Mutations in two short noncoding mononucleotide repeats in most microsatellite-unstable colorectal cancers', **Cancer Research**, vol 65, no. 11, pp. 4607-4613.

Hienonen, T, Sammalkorpi, H, Isohanni, P, Versteeg, R, Karikoski, R, Aaltonen, LA 2005, 'A 17p11.2 germline deletion in a patient with Smith-Magenis syndrome and neuroblastoma', Journal of Medical Genetics, vol 42, no. 1, pp. e3.

Kilpivaara, O, Bartkova, J, Eerola, H, Syrjäkoski, K, Vahteristo, P, Lukas, J, Blomqvist, C, Holli, K, Heikkilä, P, Sauter, G, Kallioniemi, O, Bartek, J, Nevanlinna, H **2005**, 'Correlation of CHEK2 protein expression and c.1100delC mutation status with tumor characteristics among unselected breast cancer patients', **International Journal of Cancer**, vol 113, no. 4, pp. 575-580.

Kiuru, MHT, Lehtonen, R, Eerola, H, Aittomäki, K, Blomqvist, C, Nevanlinna, H, Aaltonen, LA, Launonen, V 2005, 'No germline FH mutations in familial breast cancer patients', European Journal of Human Genetics, vol 13, no. 4, pp. 506-509.

Kruhøffer, M, Jensen, JL, Laiho, P, Dyrskjot, L, Salovaara, R, Arango, D, Birkenkamp-Demtröder, K, Sørensen, FB, Christensen, LL, Buhl, L, Mecklin, J, Järvinen, HJ, Thykjaer, T, Wikman, F, Bech-Knudsen, F, Juhola, M, Nupponen, N, Laurberg, S, Andersen, CL, Aaltonen, LA, Ørntoft, TF **2005**, 'Gene expression signatures for colorectal cancer microsatellite status and HNPCC', **British Journal of Cancer**, vol 92, no. 12, pp. 2240-2248.

Launonen, V 2005, 'Mutations in the human LKB1/STK11 gene', Human Mutation, vol 26, no. 4, pp. 291-297.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Rasinpera, H, Forsblom, C, Enattah, NS, Halonen, P, Salo, K, Victorzon, M, Mecklin, JP, Jarvinen, H, Enholm, S, Sellick, G, Alazzouzi, H, Houlston, R, Robinson, J, Groop, PH, Tomlinson, I, Schwartz, S, Aaltonen, LA, Jarvela, I, FinnDiane Study Grp **2005**, 'The C/C-13910 genotype of adult-type hypolactasia is associated with an increased risk of colorectal cancer in the Finnish population', **Gut**, vol 54, pp. 643-647.

Sotamaa, K, Liyanarachchi, S, Mecklin, J, Järvinen, HJ, Aaltonen, LA, Peltomäki, P, Chapelle, ADL 2005, 'p53 codon 72 and MDM2 SNP309 polymorphisms and age of colorectal cancer onset in Lynch syndrome', Clinical Cancer Research, vol 11, no. 19, pp. 6840-6844.

Sweet, K, Willis, J, Zhou, X, Gallione, C, Sawada, T, Alhopuro, P, Khoo, SK, Patocs, A, Martin, C, Bridgeman, S, Heinz, J, Pilarski, R, Lehtonen, R, Prior, TW, Frebourg, T, Teh, BT, Marchuk, DA, Aaltonen, LA, Eng, C 2005, 'Molecular classification of patients with unexplained hamartomatous and hyperplastic polyposis', JAMA : The Journal of the American Medical Association, vol 294, no. 19, pp. 2465-2473.

Vahteristo, P, Yliannala, K, Tamminen, A, Eerola, H, Blomqvist, C, Nevanlinna, H 2005, 'BACH1 Ser919Pro variant and breast cancer risk', BMC Cancer, vol 6, no. 19, pp. 1-7.

Vahteristo, P, Ojala, S, Tamminen, A, Tommiska, J, Sammalkorpi, H, Kiuru-Kuhlefelt, S, Eerola, H, Aaltonen, LA, Aittomäki, K, Nevanlinna, H 2005, 'No MSH6 germline mutations in breast cancer families with colorectal and/or endometrial cancer', Journal of Medical Genetics, vol 42, no. 4, pp. e22.

Vanharanta, S, Wortham, NC, Laiho, P, Sjöberg, J, Aittomäki, K, Arola, J, Tomlinson, IPM, Karhu, A, Arango, D, Aaltonen, LA 2005, '7q deletion mapping and expression profiling in uterine fibroids', Oncogene, vol 24, no. 43, pp. 6545-6554.

2006

Björklund, M, Taipale, MK, Varjosalo, MT, Saharinen, J, Lahdenperä, J, Taipale, J 2006, 'Identification of pathways regulating cell size and cell-cycle progression by RNAi', Nature, vol 439, pp. 1009-1013.

Bond, GL, Menin, C, Bertorelle, R, Alhopuro, P, Aaltonen, LA, Levine, AJ 2006, 'MDM2 SNP309 accelerates colorectal tumour formation in women', Journal of Medical Genetics, vol 43, no. 12, pp. 950-952.

Davalos, V, Dopeso, H, Castano, J, Wilson, AJ, Vilardell, F, Romero-Gimenez, J, Espin, E, Armengol, M, Capella, G, Mariadason, JM, Aaltonen, LA, Schwartz, S, Arango, D 2006, 'EPHB4 and survival of colorectal cancer patients', Cancer Research, vol 66, no. 18, pp. 8943-8948.

Hallikas, O, Taipale, J 2006, 'High-throughput assay for determining specificity and affinity of protein-DNA binding interactions', Nature Protocols, vol 1, no. 1, pp. 215-222.

Hallikas, O, Palin, K, Sinjushina, N, Rautiainen, R, Partanen, JM, Ukkonen, EJ, Taipale, J 2006, 'Genome-wide prediction of mammalian enhancers based on analysis of transcription-factor binding affinity', Cell, vol 124, no. 1, pp. 47-59.

Hallikas, OK, Aaltonen, JM, von Koskull, H, Lindberg, LA, Valmu, L, Kalkkinen, N, Wahlström, T, Kataoka, H, Andersson, L, Lindholm, D, Schröder, J 2006, 'Identification of antibodies against HAI-1 and integrin alpha 6 beta 4 as immunohistochemical markers of human villous cytotrophoblast', Journal of Histochemistry and Cytochemistry, vol 54, pp. 745-752.

Harjunpää, A, Taskinen, MM, Nykter, M, Karjalainen-Lindsberg, M, Nyman, H, Monni, O, Hemmer, S, Yli-Harja, O, Hautaniemi, S, Meri, S, Leppä, S 2006, 'Differential gene expression in non-malignant tumour microenvironment is associated with outcome in follicular lymphoma patients treated with rituximab and CHOP', British Journal of Haematology, vol 135, no. 1, pp. 33-42.

Hemmoranta, H, Hautaniemi, S, Niemi, J, Nicorici, D, Laine, J, Yli-Harja, O, Partanen, J, Jaatinen, T 2006, Transcriptional profiling reflects shared and unique characters for CD34(+) and CD133(+) cells', Stem Cells and Development, vol 15, no. 6, pp. 839-851.

Hienonen, T, Salovaara, R, Mecklin, J, Järvinen, HJ, Karhu, A, Aaltonen, LA 2006, 'Preferential amplification of AURKA 91A (IIe31) in familial colorectal cancers', International Journal of Cancer, vol 118, no. 2, pp. 505-508.

Hua, F, Hautaniemi, S, Yokoo, R, Lauffenburger, DA 2006, 'Integrated mechanistic and data-driven modelling for multivariate analysis of signalling pathways', Journal of the Royal Society Interface, vol 3, no. 9, pp. 515-526.

Karppinen, S, Barkardottir, RB, Backenhorn, K, Sydenham, T, Syrjäkoski, K, Schleutker, J, Ikonen, T, Pylkäs, K, Rapakko, K, Erkko, H, Johannesdottir, G, Gerdes, A, Thomassen, M, Agnarsson, BA, Grip, M, Kallioniemi, A, Kere, J, Aaltonen, LA, Arason, A, Møller, P, Kruse, TA, Borg, Å, Winqvist, R **2006**, Nordic collaborative study of the BARD1 Cys557Ser allele in 3956 patients with cancer: enrichment in familial BRCA1/BRCA2 mutation-negative breast cancer but not in other malignancies', **Journal of Medical Genetics**, vol 43, no. 11, pp. 856-862.

Kilpivaara, O, Alhopuro, P, Vahteristo, P, Aaltonen, LA, Nevanlinna, H 2006, 'CHEK2 1157T associates with familial and sporadic colorectal cancer: [electronic letter]', Journal of Medical Genetics, vol 43, pp. e34.

Kokko, A, Laiho, P, Lehtonen, R, Korja, S, Carvajal-Carmona, L, Järvinen, HJ, Mecklin, J, Eng, C, Schleutker, J, Tomlinson, IPM, Vahteristo, P, Aaltonen, LA 2006, 'EPHB2 germline variants in patients with colorectal cancer or hyperplastic polyposis', BMC Cancer, vol 6, no. 145.

Kokko, A, Ylisaukko-oja, S, Kiuru, M, Takatalo, M, Salmikangas, P, Tuimala, J, Arango, D, Karhu, A, Aaltonen, LA, Jäntti, J **2006**, 'Modeling tumor predisposing FH mutations in yeast: effects on fumarase activity, growth phenotype and gene expression profile', **International Journal of Cancer**, vol 118, no. 6, pp. 1340-1345.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Lehtonen, H, Kiuru, M, Ylisaukko-oja, S, Salovaara, R, Herva, R, Koivisto, P, Vierimaa, O, Aittomäki, K, Pukkala, E, Launonen, V, Aaltonen, LA, Lehtonen, H **2006**, 'Increased risk of cancer in patients with fumarate hydratase germline mutation', **Journal of Medical Genetics**, vol 43, no. 6, pp. 523-526.

Palin, K, Taipale, J, Ukkonen, E 2006, 'Locating potential enhancer elements by comparative genomics using the EEL software', Nature Protocols, vol 1, no. 1, pp. 368-374.

Ropero, S, Fraga, MF, Ballestar, E, Hamelin, R, Yamamoto, H, Boix-Chornet, M, Caballero, R, Alaminos, M, Setien, F, Paz, MF, Herranz, M, Palacios, J, Arango, D, Ørntoft, TF, Aaltonen, LA, Schwartz, S, Esteller, M **2006**, 'A truncating mutation of HDAC2 in human cancers confers resistance to histone deacetylase inhibition', **Nature Genetics**, vol 38, no. 5, pp. 566-569.

Vahteristo, P, Syrjäkoski, K, Heikkinen, T, Eerola, H, Aittomäki, K, von Smitten, K, Holli, K, Blomqvist, C, Kallioniemi, O, Nevanlinna, H 2006, 'BARD1 variants Cys557Ser and Val507Met in breast cancer predisposition', European Journal of Human Genetics, vol 14, no. 2, pp. 167-172.

Vanharanta, S, Pollard, PJ, Lehtonen, HJ, Laiho, P, Sjöberg, J, Leminen, A, Aittomäki, K, Arola, J, Kruhøffer, M, Ørntoft, TF, Tomlinson, IP, Kiuru, M, Arango, D, Aaltonen, LA **2006**, 'Distinct expression profile in fumarate-hydratase-deficient uterine fibroids', **Human Molecular Genetics**, vol 15, no. 1, pp. 97-103.

Varjosalo, MT, Li, S, Taipale, J 2006, 'Divergence of hedgehog signal transduction mechanism between Drosophila and mammals', Developmental Cell, vol 10, pp. 177-186.

Vierimaa, O, Georgitsi, M, Lehtonen, R, Vahteristo, P, Kokko, A, Raitila, A, Tuppurainen, K, Ebeling, T, Salmela, PI, Paschke, R, Gundogdu, S, De Menis, E, Mäkinen, MJ, Launonen, V, Karhu, A, Aaltonen, LA **2006**, 'Pituitary adenoma predisposition caused by germline mutations in the AIP gene', **Science**, vol 312, no. 5777, pp. 1228-1230.

Volikos, E, Robinson, J, Aittomäki, K, Mecklin, J, Järvinen, H, Westerman, AM, de Rooij, FWM, Vogel, T, Moeslein, G, Launonen, V, Tomlinson, IPM, Silver, ARJ, Aaltonen, LA 2006, 'LKB1 exonic and whole gene deletions are a common cause of Peutz-Jeghers syndrome: [electronic letter]', Journal of Medical Genetics, vol 43, no. 5, pp. e18.

Wolf-Yadlin, A, Kumar, N, Zhang, Y, Hautaniemi, S, Zaman, M, Kim, H, Grantcharova, V, Lauffenburger, D, White, F 2006, 'Effects of HER2 overexpression on cell signaling networks governing proliferation and migration', Molecular Systems Biology, vol 2, no. 54.

Ylisaukko-oja, SK, Cybulski, C, Lehtonen, R, Kiuru, M, Matyjasik, J, Szymanska, A, Szymanska-Pasternak, J, Dyrskjot, L, Butzow, R, Orntoft, TF, Launonen, V, Lubinski, J, Aaltonen, LA 2006, 'Germline fumarate hydratase mutations in patients with ovarian mucinous cystadenoma', European Journal of Human Genetics, vol 14, no. 7, pp. 880-883.

Ylisaukko-oja, S, Kiuru, M, Lehtonen, HJ, Lehtonen, R, Pukkala, E, Arola, J, Launonen, V, Aaltonen, LA **2006**, 'Analysis of fumarate hydratase mutations in a population-based series of early onset uterine leiomyosarcoma patients', **International Journal of Cancer**, vol 119, no. 2, pp. 283-287.

2007

Aaltonen, LA, Johns, L, Järvinen, HJ, Mecklin, J, Houlston, RS 2007, 'Explaining the familial colorectal cancer risk associated with mismatch repair (MMR)-deficient and MMR-stable tumors', Clinical Cancer Research, vol 13, no. 1, pp. 356-361.

Aktan-Collan, K, Haukkala, A, Pylvänäinen, K, Järvinen, HJ, Aaltonen, LA, Peltomäki, P, Rantanen, E, Kääriäinen, H, Mecklin, J 2007, 'Direct contact in inviting high-risk members of hereditary colon cancer families to genetic counselling and DNA testing', **Journal of** Medical Genetics, vol 44, no. 11, pp. 732-738.

Alazzouzi, H, Suriano, G, Guerra, A, Plaja, A, Espin, E, Armengol, M, Alhopuro, P, Velho, S, Shinomura, Y, Gonzales-Aguilera, JJ, Yamamoto, H, Aaltonen, LA, Moreno, V. Capella, G, Peinado, MA, Seruca, R, Arango, D, Schwartz, S 2007, 'Tumour selection advantage of non-dominant negative P53 mutations in homozygotic MDM2-SNP309 colorectal cancer cells', Journal of Medical Genetics, vol 44, no. 1, pp. 75-80.

Andersen, CL, Schepeler, T, Thornsen, K, Birkenkamp-Demtröder, K, Mansilla, F, Aaltonen, LA, Laurberg, S, Ørntoft, TF 2007, 'Clusterin expression in normal mucosa and colorectal cancer', Molecular and Cellular Proteomics, vol 6, no. 6, pp. 1039-1048.

Birkenkamp-Demtroder, K, Mansilla, F, Sorensen, FB, Kruhoffer, M, Cabezon, T, Christensen, LL, Aaltonen, LA, Verspaget, HW, Orntoft, TF 2007, 'Phosphoprotein Keratin 23 accumulates in MSS but not MSI colon cancers in vivo and impacts viability and proliferation in vitro', **Molecular oncology.**, vol 1, pp. 181-195.

Erkko, H, Xia, B, Nikkilä, J, Schleutker, J, Syrjäkoski, K, Mannermaa, A, Kallioniemi, A, Pylkäs, K, Karppinen, S, Rapakko, K, Miron, A, Sheng, Q, Li, G, Mattila, H, Bell, DW, Haber, DA, Grip, M, Reiman, M, Jukkola-Vuorinen, A, Mustonen, A, Kere, J, Aaltonen, LA, Kosma, V, Kataja, V, Soini, Y, Drapkin, RI, Livingston, DM, Winqvist, R **2007**, 'A recurrent mutation in PALB2 in Finnish cancer families', **Nature**, vol 446, no. 7133, pp. 316-319.

Georgitsi, M, Raitila, A, Karhu, A, van der Luijt, RB, Aalfs, CM, Sane, T, Vierimaa, O, Mäkinen, MJ, Tuppurainen, K, Paschke, R, Gimm, O, Koch, CA, Gundogdu, S, Lucassen, A, Tischkowitz, M, Izatt, L, Aylwin, S, Bano, G, Hodgson, S, De Menis, E, Launonen, V, Vahteristo, P, Aaltonen, LA 2007, 'Germline CDKN1B/p27Kip1 mutation in multiple endocrine neoplasia', Journal of Clinical Endocrinology and Metabolism, vol 92, no. 8, pp. 3321-3325.

Georgitsi, M, Raitila, A, Karhu, A, Tuppurainen, K, Mäkinen, MJ, Vierimaa, O, Paschke, R, Saeger, W, van der Luijt, RB, Sane, T, Robledo, M, de Menis, E, Weil, RJ, Wasik, A, Zielinski, G, Lucewicz, O, Lubinski, J, Launonen, V, Vahteristo, P, Aaltonen, LA 2007, 'Molecular diagnosis of pituitary adenoma predisposition caused by aryl hydrocarbon receptor-interacting protein gene mutations', Proceedings of the National Academy of Sciences of the United States of America, vol 104, pp. 4101-4105.



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CSB/Aaltonen

Georgitsi, M, Karhu, A, Winqvist, R, Visakorpi, T, Waltering, K, Vahteristo, P, Launonen, V, Aaltonen, LA 2007, 'Mutation analysis of aryl hydrocarbon receptor interacting protein (AIP) gene in colorectal, breast, and prostate cancers', British Journal of Cancer, vol 96, no. 2, pp. 352-356.

Kharait, S, Hautaniemi, S, Wu, S, Iwabu, A, Lauffenburger, D, Wells, A 2007, 'Decision tree modeling predicts effects of inhibiting contractility signaling on cell motility', BMC Systems Biology, vol 1, no. 9, 13 s.

Laakso, M, Tuupanen, S, Karhu, A, Lehtonen, R, Aaltonen, LA, Hautaniemi, S **2007**, 'Computational identification of candidate loci for recessively inherited mutation using high-throughput SNP arrays: genetics and population analysis', **Bioinformatics**, vol 23, no. 15, pp. 1952-1961.

Laiho, P, Kokko, A, Vanharanta, S, Salovaara, RK, Sammalkorpi, H, Järvinen, HJ, Mecklin, J, Karttunen, TJ, Tuppurainen, K, Davalos, V, Schwartz, S, Arango, D, Mäkinen, MJ, Aaltonen, LA 2007, 'Serrated carcinomas form a subclass of colorectal cancer with distinct molecular basis', Oncogene, vol 26, no. 2, pp. 312-320.

Lehtonen, HJ, Ylisaukko-oja, SK, Kiuru, M, Karhu, A, Lehtonen, R, Vanharanta, S, Jalanko, A, Aaltonen, LA, Launonen, V 2007, 'Stressinduced expression of a novel variant of human fumarate hydratase (FH)', Gene Expression, vol 14, pp. 59-69.

Lehtonen, HJ, Mäkinen, MJ, Kiuru, M, Laiho, P, Herva, R, van Minderhout, I, Hogendoorn, PCW, Cornelisse, C, Devilee, P, Launonen, V, Aaltonen, LA 2007, 'Increased HIF1 alpha in SDH and FH deficient tumors does not cause microsatellite instability', International Journal of Cancer, vol 121, no. 6, pp. 1386-1389.

Lehtonen, HJ, Blanco, I, Piulats, JM, Herva, R, Launonen, V, Aaltonen, LA 2007, 'Conventional renal cancer in a patient with fumarate hydratase mutation', Human Pathology, vol 38, no. 5, pp. 793-796.

Mansilla, F, Birkenkamp-Demtröder, K, Kruhøffer, M, Sørensen, FB, Andersen, CL, Laiho, P, Aaltonen, LA, Verspaget, HW, Ørntoft, TF 2007, 'Differential expression of DHHC9 in microsatellite stable and instable human colorectal cancer subgroups', British Journal of Cancer, vol 96, no. 12, pp. 1896-1903.

Mecklin, J, Aarnio, M, Läärä, E, Kairaluoma, M, Pylvänäinen, K, Peltomäki, P, Aaltonen, LA, Järvinen, HJ 2007, 'Development of colorectal tumors in colonoscopic surveillance in Lynch syndrome', Gastroenterology, vol 133, no. 4, pp. 1093-1098.

Niemela, AM, Hynninen, P, Mecklin, J, Kuopio, T, Kokko, A, Aaltonen, LA, Parkkila, A, Pastorekova, S, Pastorek, J, Waheed, A, Sly, WS, Ørntoft, TF, Kruhøffer, M, Haapasalo, H, Parkkila, S, Kivelä, A 2007, 'Carbonic anhydrase IX is highly expressed in hereditary nonpolyposis colorectal cancer', Cancer Epidemiology, Biomarkers & Prevention, vol 16, no. 9, pp. 1760-1766.

Raitila, A, Georgitsi, M, Karhu, A, Tuppurainen, K, Mäkinen, MJ, Birkenkamp-Demtröder, K, Salmenkivi, K, Ørntoft, TF, Arola, J, Launonen, V, Vahteristo, P, Aaltonen, LA 2007, 'No evidence of somatic aryl hydrocarbon receptor interacting protein mutations in sporadic endocrine neoplasia', Endocrine - Related Cancer, vol 14, no. 3, pp. 901-906.

Sammalkorpi, H, Alhopuro, P, Lehtonen, R, Tuimala, J, Mecklin, J, Järvinen, HJ, Jiricny, J, Karhu, A, Aaltonen, LA 2007, 'Background mutation frequency in microsatellite-unstable colorectal cancer', Cancer Research, vol 67, no. 12, pp. 5691-5698.

Tuupanen, S, Karhu, A, Järvinen, HJ, Mecklin, J, Launonen, V, Aaltonen, LA 2007, 'No evidence for dual role of loss of heterozygosity in hereditary non-polyposis colorectal cancer', Oncogene, vol 26, no. 17, pp. 2513-2517.

Tuupanen, S, Alhopuro, P, Mecklin, J, Järvinen, HJ, Aaltonen, LA 2007, 'No evidence for association of NOD2 R702W and G908R with colorectal cancer', International Journal of Cancer, vol 121, no. 1, pp. 76-79.

Vahteristo, P, Kokko, A, Saksela, O, Aittomäki, K, Aaltonen, LA 2007, 'Blood-derived gene-expression profiling in unravelling susceptibility to recessive disease', Journal of Medical Genetics, vol 44, no. 11, pp. 718-720.

Vanharanta, S, Wortham, NC, Langford, C, El-Bahrawy, M, van der Spuy, Z, Sjöberg, J, Lehtonen, R, Karhu, A, Tomlinson, IPM, Aaltonen, LA 2007, 'Definition of a minimal region of deletion of chromosome 7 in uterine leiomyomas by tiling-path microarray CGH and mutation analysis of known genes in this region', Genes, Chromosomes & Cancer, vol 46, no. 5, pp. 451-458.

Wolf-Yadlin, A, Hautaniemi, S, Lauffenburger, D, White, F 2007, 'Multiple reaction monitoring for robust quantitative proteomic analysis of cellular signaling networks', Proceedings of the National Academy of Sciences of the United States of America, vol 104, no. 14, pp. 5860-5865.

Yang, Q, Zhang, R, Horikawa, I, Fujita, K, Afshar, Y, Kokko, A, Laiho, P, Aaltonen, LA, Harris, CC 2007, 'Functional diversity of human protection of telomeres 1 isoforms in telomere protection and cellular senescence', Cancer Research, vol 67, no. 24, pp. 11677-11686.

2008

Alberici, P, Gaspar, C, Franken, P, Gorski, MM, de Vries, I, Scott, RJ, Ristimäki, A, Aaltonen, LA, Fodde, R 2008, 'Smad4 haploinsufficiency: a matter of dosage', PathoGenetics, vol 1, no. 2.

Alhopuro, P, Phichith, D, Tuupanen, S, Sammalkorpi, H, Nybondas, M, Saharinen, J, Robinson, JP, Yang, Z, Chen, L, Ørntoft, TF, Mecklin, J, Järvinen, H, Eng, C, Moeslein, G, Shibata, D, Houlston, RS, Lucassen, A, Tomlinson, IPM, Launonen, V, Ristimäki, A, Arango, D, Karhu, A, Sweeney, HL, Aaltonen, LA **2008**, 'Unregulated smooth-muscle myosin in human intestinal neoplasia', **Proceedings of the National Academy of Sciences of the United States of America**, vol 105, no. 14, pp. 5513-5518.

Alhopuro, P, Karhu, A, Winqvist, R, Waltering, K, Visakorpi, T, Aaltonen, L 2008, 'Somatic mutation analysis of MYH11 in breast and prostate cancer', BMC Cancer, vol 8, pp. 263.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Bayley, J, Launonen, V, Tomlinson, IPM 2008, 'The FH mutation database: an online database of fumarate hydratase mutations involved in the MCUL (HLRCC) tumor syndrome and congenital fumarase deficiency', BMC Medical Genetics, vol 9, no. 20, pp. -.

Georgitsi, M, De Menis, E, Cannavo, S, Mäkinen, MJ, Tuppurainen, K, Pauletto, P, Curto, L, Weil, RJ, Paschke, R, Zielinski, G, Wasik, A, Lubinski, J, Vahteristo, P, Karhu, A, Aaltonen, LA **2008**, 'Aryl hydrocarbon receptor interacting protein (AIP) gene mutation analysis in children and adolescents with sporadic pituitary adenomas', **Clinical Endocrinology**, vol 69, no. 4, pp. 621-627.

Georgitsi, M, Heliövaara, E, Paschke, R, Kumar, AVK, Tischkowitz, M, Vierimaa, O, Salmela, P, Sane, T, De Menis, E, Cannavo, S, Gundogdu, S, Lucassen, A, Izatt, L, Aylwin, S, Bano, G, Hodgson, S, Koch, CA, Karhu, A, Aaltonen, LA **2008**, 'Large genomic deletions in AIP in pituitary adenoma predisposition', **Journal of Clinical Endocrinology and Metabolism**, vol 93, no. 10, pp. 4146-4151.

Haiko, P, Mäkinen, T, Keskitalo, S, Taipale, J, Kärkkäinen, M, Baldwin, M, Stacker, S, Achen, M, Alitalo, K 2008, 'Deletion of vascular endothelial growth factor C (VEGF-C) and VEGF-D is not equivalent to VEGF receptor 3 deletion in mouse embryos', **Molecular and Cellular Biology**, vol 28 8), no. 15, pp. 4843-4850.

Heinonen, H, Nieminen, A, Saarela, M, Kallioniemi, A, Klefström, J, Hautaniemi, S, Monni, O 2008, 'Deciphering downstream gene targets of PI3K/mTOR/p70S6K pathway in breast cancer', BMC Genomics, vol 9, no. 348, pp. 1-12.

Houlston, RS, Niittymäki, I, Tuupanen, S, Karhu, A, Aaltonen, LA, CORGI Consortium, Colorectal Cancer Association Study Consortium and International Colorectal Cancer Genetic Association Consortium **2008**, 'Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer', **Nature Genetics**, vol 40, no. 12, pp. 1426-1435.

Jorissen, RN, Lipton, L, Gibbs, P, Chapman, M, Desai, J, Jones, IT, Yeatman, TJ, East, P, Tomlinson, IPM, erspaget, HW, Aaltonen, LA, Kruhøffer, M, Ørntoft, TF, Andersen, CL, Sieber, OM **2008**, 'DNA copy-number alterations underlie gene expression differences between microsatellite stable and unstable colorectal cancers', **Clinical Cancer Research**, vol 14, no. 24, pp. 8061-8069.

Kilpinen, S, Autio, R, Ojala, K, Iljin, K, Bucher, E, Sara, H, Pisto, T, Saarela, M, Skotheim, R, Björkman, M, Mpindi, J, Haapa-Paananen, S, Vainio, P, Edgren, H, Wolf, M, Astola, J, Nees, M, Hautaniemi, S, Kallioniemi, O **2008**, 'Systematic bioinformatic analysis of expression levels of 17,330 human genes across 9,783 samples from 175 types of healthy and pathological tissues', **Genome Biology**, vol 9, no. 9, pp. 1-14 (R139).

Koski (Ahvenainen), TA, Lehtonen, HJ, Lehtonen, R, Vahteristo, P, Aittomäki, K, Baynam, G, Dommering, C, Eng, C, Gruber, SB, Grönberg, H, Harvima, R, Herva, R, Hietala, M, Kujala, M, Kääriänen, H, Sunde, L, Vierimaa, O, Pollard, PJ, Tomlinson, IP, Björck, E, Aaltonen, LA, Launonen, V **2008**, 'Mutation screening of fumarate hydratase by multiplex ligation-dependent probe amplification: detection of exonic deletion in a patient with leiomyomatosis and renal cell cancer', **Cancer Genetics and Cytogenetics**, vol 183, no. 2, pp. 83-88.

Ovaska, K, Laakso, M, Hautaniemi, S 2008, 'Fast Gene Ontology based clustering for microarray experiments', BioData mining, vol 1, no. 11, pp. 1-8.

Parizot, B, Laplaze, L, Ricaud, L, Boucheron-Dubuisson, E, Bayle, V, Bonke, M, Smet, ID, Poethig, SR, Helariutta, Y, Haseloff, J, Chriqui, D, Beeckman, T, Nussaume, L **2008**, 'Diarch symmetry of the vascular bundle in Arabidopsis Root encompasses the pericycle and is reflected in distich lateral root initiation', **Plant Physiology**, vol 146, no. 1, pp. 140-148.

Petrova, T, Nykänen, A, Norrmen, C, Ivanov, KI, Andersson, LC, Haglund, C, Puolakkainen, P, Wempe, F, von Melchner, H, Gradwohl, G, Vanharanta, S, Aaltonen, LA, Saharinen, JMN, Gentile, M, Clarke, A, Taipale, J, Oliver, G, Alitalo, K **2008**, 'Transcription factor PROX1 induces colon cancer progression by promoting the transition from benign to highly dysplastic phenotype', **Cancer Cell**, vol 13, no. 5, pp. 407-419.

Pittman, AM, Niittymäki, I, Tuupanen, S, Karhu, A, Aaltonen, LA, CORGI Consortium and EPICOLON Consortium **2008**, 'Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer', **Human Molecular Genetics**, vol 17, no. 23, pp. 3720-3727.

Sammalkorpi, H, Alhopuro, P, Niittymäki, I, Ørntoft, TF, Hokland, P, Karhu, A, Aaltonen, LA 2008, 'Mutation analysis of MYH11 in acute myeloid leukemia', Leukemia and Lymphoma, vol 49, no. 9, pp. 1829-1831.

Siltanen, S, Syrjäkoski, K, Fagerholm, R, Ikonen, T, Lipman, P, Mallot, J, Holli, K, Tammela, TLJ, Järvinen, HJ, Mecklin, J, Aittomäki, K, Blomqvist, C, Bailey-Wilson, JE, Nevanlinna, H, Aaltonen, LA, Schleutker, J, Vahteristo, P 2008, 'ARLTS1 germline variants and the risk for breast, prostate, and colorectal cancer', European Journal of Human Genetics, vol 16, no. 8, pp. 983-991.

Tomlinson, IPM, Niittymäki, I, Tuupanen, S, Karhu, A, Aaltonen, LA, CORGI Consortium and EPICOLON Consortium **2008**, 'A genomewide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3', **Nature Genetics**, vol 40, no. 5, pp. 623-630.

Tuupanen, S, Niittymäki, I, Nousiainen, KJ, Vanharanta, S, Mecklin, J, Nuorva, K, Järvinen, H, Hautaniemi, S, Karhu, A, Aaltonen, LA 2008, 'Allelic imbalance at rs6983267 suggests selection of the risk allele in somatic colorectal tumor evolution', Cancer Research, vol 68, no. 1, pp. 14-17.

Varjosalo, M, Björklund, M, Cheng, F, Syvänen, H, Kivioja, T, Kilpinen, S, Sun, Z, Kallioniemi, O, Stunnenberg, HG, He, W, Ojala, P, Taipale, J **2008**, 'Application of active and kinase-deficient kinome collection for identification of kinases regulating hedgehog signaling', **Cell**, vol 133, no. 3, pp. 537-548.

2009



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Andersen, CL, Christensen, LL, Thorsen, K, Schepeler, T, Sorensen, FB, Verspaget, HW, Simon, R, Kruhoffer, M, Aaltonen, LA, Laurberg, S, Ørntoft, TF 2009, 'Dysregulation of the transcription factors SOX4, CBFB and SMARCC1 correlates with outcome of colorectal cancer', British Journal of Cancer, vol 100, no. 3, pp. 511-523.

Autio, R, Kilpinen, S, Saarela, M, Kallioniemi, O, Hautaniemi, S, Astola, J 2009, 'Comparison of Affymetrix data normalization methods using 6,926 experiments across five array generations', BMC Bioinformatics, vol 10, no. suppl. 1, pp. S24.

Autio, R, Saarela, M, Jarvinen, A, Hautaniemi, S, Astola, J 2009, 'Advanced analysis and visualization of gene copy number and expression data', BMC Bioinformatics, vol 10, no. Suppl. 1, pp. -.

Backlund, M, Paukku, K, Daviet, L, de Boer, RA, Valo, EA, Hautaniemi, S, Kalkkinen, N, Ehsan, A, Kontula, K, Lehtonen, JYA 2009, 'Posttranscriptional regulation of angiotensin II type 1 receptor expression by glyceraldehyde 3-phosphate dehydrogenase', Nucleic Acids Research, vol 37, no. 7, pp. 2346-2358.

Blomster, H, Hietakangas, V, Wu, J, Kouvonen, P, Hautaniemi, S, Sistonen, L 2009, 'Novel proteomics strategy brings insight into the prevalence of SUMO-2 target sites', Molecular and Cellular Proteomics, vol 8, pp. 1382-1390.

Cheng, F, Weidner-Glunde, M, Varjosalo, M, Rainio, E, Lehtonen, A, Schulz, TF, Koskinen, PJ, Taipale, J, Ojala, PM 2009, 'KSHV reactivation from latency requires Pim-1 and Pim-3 kinases to inactivate the latency-associated nuclear antigen LANA', PLoS Pathogens, vol 5, no. 3.

Habermann, JK, Doering, J, Hautaniemi, S, Roblick, UJ, Buendgen, NK, Nicorici, D, Kronenwett, U, Rathnagiriswaran, S, Mettu, RKR, Ma, Y, Krueger, S, Bruch, H, Auer, G, Guo, NL, Ried, T 2009, 'The gene expression signature of genomic instability in breast cancer is an independent predictor of clinical outcome', International Journal of Cancer, vol 124, no. 7, pp. 1552-1564.

Heliövaara, E, Raitila, A, Launonen, V, Paetau, A, Arola, J, Lehtonen, H, Sane, T, Weil, RJ, Vierimaa, O, Salmela, P, Tuppurainen, K, Mäkinen, M, Aaltonen, LA, Karhu, A 2009, 'The expression of AIP-related molecules in elucidation of cellular pathways in pituitary adenomas', American Journal of Pathology, vol 175, no. 6, pp. 2501-2507.

Jennings, JE, Georgitsi, M, Holdaway, I, Daly, AF, Tichomirowa, M, Beckers, A, Aaltonen, LA, Karhu, A, Cameron, FJ **2009**, 'Aggressive pituitary adenomas occurring in young patients in a large Polynesian kindred with a germline R271W mutation in the AIP gene', **European Journal of Endocrinology**, vol 161, no. 5, pp. 799-804.

Jorissen, RN, Gibbs, P, Christie, M, Prakash, S, Lipton, L, Desai, J, Kerr, D, Aaltonen, LA, Arango, D, Kruhoffer, M, Orntoft, TF, Andersen, CL, Gruidl, M, Kamath, VP, Eschrich, S, Yeatman, TJ, Sieber, OM **2009**, 'Metastasis-Associated Gene Expression Changes Predict Poor Outcomes in Patients with Dukes Stage B and C Colorectal Cancer', **Clinical Cancer Research**, vol 15, pp. 7642-7651.

Järvinen, HJ, Renkonen-Sinisalo, L, Aktan-Collan, K, Peltomäki, P, Aaltonen, LA, Mecklin, J 2009, 'Ten years after mutation testing for Lynch syndrome: cancer incidence and outcome in mutation-positive and mutation-negative family members', Journal of Clinical Oncology, vol 27, no. 28, pp. 4793-4797.

Koski, TA, Lehtonen, HJ, Jee, KJ, Ninomiya, S, Joosse, SA, Vahteristo, P, Kiuru, M, Karhu, A, Sammalkorpi, H, Vanharanta, S, Lehtonen, R, Edgren, SHT, Nederlof, PM, Hietala, M, Aittomäki, K, Herva, R, Knuutila, S, Aaltonen, LA, Launonen, V **2009**, 'Array comparative genomic hybridization identifies a distinct DNA copy number profile in renal cell cancer associated with hereditary leiomyomatosis and renal cell cancer', **Genes, Chromosomes & Cancer**, vol 48, no. 7, pp. 544-551.

Melo, SA, Ropero, S, Moutinho, C, Aaltonen, LA, Yamamoto, H, Calin, GA, Rossi, S, Fernandez, AF, Carneiro, F, Oliveira, C, Ferreira, B, Liu, C, Villanueva, A, Capella, G, Schwartz, SJ, Shiekhattar, R, Esteller, M **2009**, 'A TARBP2 mutation in human cancer impairs microRNA processing and DICER1 function', **Nature Genetics**, vol 41, no. 3, pp. 365-370.

Nymark, P, Kettunen, E, Aavikko, M, Ruosaari, S, Kuosma, E, Vanhala, E, Salmenkivi, K, Pirinen, R, Karjalainen, A, Knuutila, S, Wikman, H, Anttila, S 2009, 'Molecular alterations at 9q33.1 and polyploidy in asbestos-related lung cancer', Clinical Cancer Research, vol 15, no. 2, pp. 468-475.

Raimundo, N, Vanharanta, S, Aaltonen, LA, Hovatta, I, Suomalainen, A 2009, 'Downregulation of SRF-FOS-JUNB pathway in fumarate hydratase deficiency and in uterine leiomyomas', Oncogene, vol 28, no. 9, pp. 1261-1273.

Raitila, A, Georgitsi, M, Bonora, E, Vargiolu, M, Tuppurainen, K, Mäkinen, MJ, Vierimaa, O, Salmela, PI, Launonen, V, Vahteristo, P, Aaltonen, LA, Romeo, G, Karhu, A 2009, 'Aryl hydrocarbon receptor interacting protein mutations seem not to associate with familial non-medullary thyroid cancer', Journal of Endocrinological Investigation, vol 32, no. 5, pp. 426-429.

Rajecki, M, af Hällström, T, Hakkarainen, T, Nokisalmi, P, Hautaniemi, S, Nieminen, AI, Tenhunen, M, Rantanen, V, Desmond, RA, Chen, D, Guse, K, Stenman, U, Gargini, R, Kapanen, M, Klefström, J, Kanerva, A, Pesonen, S, Ahtiainen, L, Hemminki, A **2009**, 'Mre11 inhibition by oncolytic adenovirus associates with autophagy and underlies synergy with ionizing radiation', **International Journal of Cancer**, vol 125, no. 10, pp. 2441-2449.

Tuupanen, S, Turunen, M, Lehtonen, R, Hallikas, O, Vanharanta, S, Kivioja, T, Björklund, M, Wei, G, Yan, J, Niittymäki, I, Mecklin, J, Järvinen, H, Ristimäki, A, Di-Bernardo, M, East, P, Carvajal-Carmona, L, Houlston, RS, Tomlinson, I, Palin, K, Ukkonen, E, Karhu, A, Taipale, J, Aaltonen, LA **2009**, 'The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling', **Nature Genetics**, vol 41, no. 8, pp. 885-890.

Vierimaa, O, Villablanca, A, Alimov, A, Georgitsi, M, Raitila, A, Vahteristo, P, Larsson, C, Ruokonen, A, Eloranta, E, Ebeling, TML, Ignatius, J, Aaltonen, LA, Leisti, J, Salmela, PI **2009**, 'Mutation analysis of MEN1, HRPT2, CASR, CDKN1B, and AIP genes in primary hyperparathyroidism patients with features of genetic predisposition', **Journal of Endocrinological Investigation**, vol 32, no. 6, pp. 512-518.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Wu, J, Vallenius, T, Ovaska, K, Westermarck, J, Makela, TP, Hautaniemi, S 2009, 'Integrated network analysis platform for proteinprotein interactions', Nature methods., vol 6, no. 1, pp. 75-77.

Yuan, Z, Shin, J, Wilson, A, Goel, S, Ling, Y, Ahmed, N, Dopeso, H, Jhawer, M, Nasser, S, Montagna, C, Fordyce, K, Augenlicht, LH, Aaltonen, LA, Arango, D, Weber, TK, Mariadason, JM 2009, 'An A13 repeat within the 3'-untranslated region of epidermal growth factor receptor (EGFR) is frequently mutated in microsatellite instability colon cancers and is associated with increased EGFR expression', Cancer Research, vol 69, no. 29, pp. 7811-7818.

2010

Alhopuro, P, Björklund, M, Sammalkorpi, HP, Turunen, M, Tuupanen, SJ, Bistrom, M, Niittymäki, I, Lehtonen, H, Kivioja, T, Launonen, V, Saharinen, JMN, Nousiainen, KJ, Hautaniemi, S, Nuorva, K, Mecklin, J, Järvinen, H, Orntoft, T, Arango, D, Lehtonen, RJ, Karhu, AI, Taipale, J, Aaltonen, LA **2010**, 'Mutations in the Circadian Gene CLOCK in Colorectal Cancer', **Molecular Cancer Research**, vol 8, no. 7, pp. 952-960.

Ashrafian, H, O'Flaherty, L, Adam, J, Steeples, V, Chung, Y, East, P, Vanharanta, S, Lehtonen, H, Nye, E, Hatipoglu, E, Miranda, M, Howarth, K, Shukla, D, Troy, H, Griffiths, J, Spencer-Dene, B, Yusuf, M, Volpi, E, Maxwell, PH, Stamp, G, Poulsom, R, Pugh, CW, Costa, B, Bardella, C, Di Renzo, MF, Kotlikoff, MI, Launonen, V, Aaltonen, L, El-Bahrawy, M, Tomlinson, I, Pollard, PJ **2010**, 'Expression Profiling in Progressive Stages of Fumarate-Hydratase Deficiency: The Contribution of Metabolic Changes to Tumorigenesis', **Cancer Research**, vol 70, no. 22, pp. 9153-9165.

Daly, AF, Tichomirowa, MA, Petrossians, P, Heliövaara, E, Jaffrain-Rea, M, Barlier, A, Naves, LA, Ebeling, T, Karhu, A, Raappana, A, Cazabat, L, De Menis, E, Fajardo Montanana, C, Raverot, G, Weil, RJ, Sane, T, Maiter, D, Neggers, S, Yaneva, M, Tabarin, A, Verrua, E, Eloranta, E, Murat, A, Vierimaa, O, Salmela, PI, Emy, P, Toledo, RA, Isabel Sabate, M, Villa, C, Popelier, M, Salvatori, R, Jennings, J, Ferrandez Longas, A, Labarta Aizpun, JI, Georgitsi, M, Paschke, R, Ronchi, C, Välimäki, M, Saloranta, C, De Herder, W, Cozzi, R, Guitelman, M, Magri, F, Lagonigro, MS, Halaby, G, Corman, V, Hagelstein, M, Vanbellinghen, J, Barra, GB, Gimenez-Roqueplo, A, Cameron, FJ, Borson-Chazot, F, Holdaway, I, Toledo, SPA, Stalla, GK, Spada, A, Zacharieva, S, Bertherat, J, Brue, T, Bours, V, Chanson, P, Aaltonen, LA, Beckers, A **2010**, 'Clinical Characteristics and Therapeutic Responses in Patients with Germ-Line AIP Mutations and Pituitary Adenomas: An International Collaborative Study', **Journal of Clinical Endocrinology and Metabolism**, vol 95, no. 11, pp. E373-E383.

Dias, JD, Liikanen, I, Guse, K, Foloppe, J, Sloniecka, M, Diaconu, I, Rantanen, V, Eriksson, KM, Hakkarainen, T, Lusky, M, Erbs, P, Escutenaire, S, Kanerva, A, Pesonen, S, Cerullo, V, Hemminki, A 2010, 'Targeted Chemotherapy for Head and Neck Cancer with a Chimeric Oncolytic Adenovirus Coding for Bifunctional Suicide Protein FCU1', Clinical Cancer Research, vol 16, no. 9, pp. 2540-2549.

Enzerink, A, Rantanen, V, Vaheri, A 2010, 'Fibroblast nemosis induces angiogenic responses of endothelial cells', Experimental Cell Research, vol 316, no. 5, pp. 826-835.

Gylfe, AE, Sirkiä, J, Ahlsten, M, Järvinen, H, Mecklin, J, Karhu, A, Aaltonen, LA 2010, 'Somatic mutations and germline sequence variants in patients with familial colorectal cancer', International Journal of Cancer, vol 127, no. 12, pp. 2974-2980.

Houlston, RS, Cheadle, J, Dobbins, SE, Tenesa, A, Jones, AM, Howarth, K, Spain, SL, Broderick, P, Domingo, E, Farrington, S, Prendergast, JGD, Pittman, AM, Theodoratou, E, Smith, CG, Olver, B, Walther, A, Barnetson, RA, Churchman, M, Jaeger, EEM, Penegar, S, Barclay, E, Martin, L, Gorman, M, Mager, R, Johnstone, E, Midgley, R, Niittymäki, I, Tuupanen, S, Colley, J, Idziaszczyk, S, Thomas, HJW, Lucassen, AM, Evans, DGR, Maher, ER, Maughan, T, Dimas, A, Dermitzakis, E, Cazier, J, Aaltonen, LA, Pharoah, P, Kerr, DJ, Carvajal-Carmona, LG, Campbell, H, Dunlop, MG, Tomlinson, IPM, CORGI Consortium, CC0ENT Consortium, COINB Collaborative Grp, COIN Collaborative Grp **2010**, 'Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33', **Nature Genetics**, vol 42, no. 11, pp. 973-U89.

Jolma, A, Kivioja, T, Toivonen, J, Cheng, L, Wei, G, Enge, M, Taipale, M, Vaquerizas, JM, Yan, J, Sillanpää, MJ, Bonke, AWM, Palin, K, Talukder, S, Hughes, TR, Luscombe, NM, Ukkonen, E, Taipale, J **2010**, 'Multiplexed massively parallel SELEX for characterization of human transcription factor binding specificities', **Genome Research**, vol 20, no. 6, pp. 861-873.

Jäämaa, SS, af Hällström, TM, Sankila, A, Rantanen, V, Koistinen, H, Stenman, UH, Zhang, Z, Yang, Z, De Marzo, AM, Taari, K, Ruutu, M, Andersson, LC, Laiho, M **2010**, 'DNA Damage Recognition via Activated ATM and p53 Pathway in Nonproliferating Human Prostate Tissue', **Cancer Research**, vol 70, no. 21, pp. 8630-8641.

Kaasinen, E, Aittomaki, K, Eronen, M, Vahteristo, P, Karhu, A, Mecklin, J, Kajantie, E, Aaltonen, LA, Lehtonen, R **2010**, 'Recessively inherited right atrial isomerism caused by mutations in growth/differentiation factor 1 (GDF1)', **Human Molecular Genetics**, vol 19, pp. 2747-2753.

Laakso, M, Hautaniemi, S 2010, 'Integrative platform to translate gene sets to networks', Bioinformatics, vol 26, no. 14, pp. 1802-1803.

Lahesmaa-Korpinen, A, Carlson, SM, White, FM, Hautaniemi, S 2010, 'Integrated data management and validation platform for phosphorylated tandem mass spectrometry data', Proteomics (Weinheim), vol 10, no. 19, pp. 3515-3524.

Niittymaki, I, Kaasinen, E, Tuupanen, S, Karhu, A, Jarvinen, H, Mecklin, J, Tomlinson, IPM, Di Bernardo, MC, Houlston, RS, Aaltonen, LA **2010**, 'Low-Penetrance Susceptibility Variants in Familial Colorectal Cancer', **Cancer Epidemiology, Biomarkers & Prevention**, vol 19, no. 6, pp. 1478-1483.

Ovaska, K, Laakso, MK, Haapa-Paananen, S, Louhimo, R, Chen, P, Aittomäki, JVJ, Valo, EA, Nunez-Fontarnau, J, Rantanen, V, Karinen, SH, Nousiainen, KJ, Lahesmaa-Korpinen, AK, Miettinen, ME, Saarinen, LA, Kohonen, P, Wu, J, Westermarck, J, Hautaniemi, S **2010**, 'Large-scale data integration framework provides a comprehensive view on glioblastoma multiforme', **Genome medicine**, vol 2, no. 9, pp. 65.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

Pittman, AM, Naranjo, S, Jalava, SE, Twiss, P, Ma, Y, Olver, B, Lloyd, A, Vijayakrishnan, J, Qureshi, M, Broderick, P, van Wezel, T, Morreau, H, Tuupanen, S, Aaltonen, LA, Eva Alonso, M, Manzanares, M, Gavilan, A, Visakorpi, T, Luis Gomez-Skarmeta, J, Houlston, RS **2010**, 'Allelic Variation at the 8q23.3 Colorectal Cancer Risk Locus Functions as a Cis-Acting Regulator of EIF3H', **PLoS Genetics**, vol 6, no. 9, pp. e1001126.

Raitila, A, Lehtonen, HJ, Arola, J, Heliovaara, E, Ahlsten, M, Georgitsi, M, Jalanko, A, Paetau, A, Aaltonen, LA, Karhu, A 2010, 'Mice with Inactivation of Aryl Hydrocarbon Receptor-Interacting Protein (Aip) Display Complete Penetrance of Pituitary Adenomas with Aberrant ARNT Expression', American Journal of Pathology, vol 177, no. 4, pp. 1969-1976.

Savilahti, EM, Rantanen, V, Lin, JS, Karinen, S, Saarinen, KM, Goldis, M, Mäkelä, M, Hautaniemi, S, Savilahti, E, Sampson, HA 2010, 'Early recovery from cow's milk allergy is associated with decreasing IgE and increasing IgG4 binding to cow's milk epitopes', Journal of Allergy and Clinical Immunology, vol 125, no. 6, pp. 1315-1321.

Savilahti, EM, Karinen, S, Salo, HM, Klemetti, P, Saarinen, KM, Klemola, T, Kuitunen, M, Hautaniemi, S, Savilahti, E, Vaarala, O 2010, 'Combined T regulatory cell and Th2 expression profile identifies children with cow's milk allergy', Clinical Immunology, vol 136, no. 1, pp. 16-20.

Sugiyama, N, Varjosalo, M, Meller, P, Lohi, J, Hyytiainen, M, Kilpinen, S, Kallioniemi, O, Ingvarsen, S, Engelholm, LH, Taipale, J, Alitalo, K, Keski-Oja, J, Lehti, K **2010**, 'Fibroblast Growth Factor Receptor 4 Regulates Tumor Invasion by Coupling Fibroblast Growth Factor Signaling to Extracellular Matrix Degradation', **Cancer Research**, vol 70, pp. 7851-7861.

Sugiyama, N, Varjosalo, M, Meller, P, Lohi, J, Chan, KM, Zhou, Z, Alitalo, K, Taipale, J, Keski-Oja, J, Lehti, K **2010**, 'FGF receptor-4 (FGFR4) polymorphism acts as an activity switch of a membrane type 1 matrix metalloproteinase-FGFR4 complex', **Proceedings of the National Academy of Sciences of the United States of America**, vol 107, no. 36, pp. 15786-15791.

Taskinen, MM, Valo, EA, Karjalainen-Lindsberg, M, Hautaniemi, S, Meri, S, Leppä, S **2010**, 'Signal Transducers and Activators of Transcription 5a-Dependent Cross-talk between Follicular Lymphoma Cells and Tumor Microenvironment Characterizes a Group of Patients with Improved Outcome after R-CHOP', **Clinical Cancer Research**, vol 16, pp. 2615-2623.

Tomlinson, IPM, Dunlop, M, Campbell, H, Zanke, B, Gallinger, S, Hudson, T, Koessler, T, Pharoah, PD, Niittymäki, I, Tuupanen, S, Aatonen, LA, Hemminki, K, Lindblom, A, Forsti, A, Sieber, O, Lipton, L, van Wezel, T, Morreau, H, Wijnen, JT, Devilee, P, Matsuda, K, Nakamura, Y, Castellvi-Bel, S, Ruiz-Ponte, C, Castells, A, Carracedo, A, Ho, JWC, Sham, P, Hofstra, RMW, Vodicka, P, Brenner, H, Hampe, J, Schafmayer, C, Tepel, J, Schreiber, S, Volzke, H, Lerch, MM, Schmidt, CA, Buch, S, Moreno, V, Villanueva, CM, Peterlongo, P, Radice, P, Echeverry, MM, Velez, A, Carvajal-Carmona, L, Scott, R, Penegar, S, Broderick, P, Tenesa, A, Houlston, RS **2010**, 'COGENT (Colorectal cancer GENeTics): an international consortium to study the role of polymorphic variation on the risk of colorectal cancer', **British Journal of Cancer**, vol 102, no. 2, pp. 447-454.

Vahteristo, P, Koski, TA, Naatsaari, L, Kiuru, M, Karhu, A, Herva, R, Sallinen, S, Vierimaa, O, Bjorck, E, Richard, S, Gardie, B, Bessis, D, Van Glabeke, E, Blanco, I, Houlston, R, Senter, L, Hietala, M, Aittomaki, K, Aaltonen, LA, Launonen, V, Lehtonen, R **2010**, 'No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome', **Familial Cancer**, vol 9, pp. 245-251.

Velho, S, Oliveira, C, Paredes, J, Sousa, S, Leite, M, Matos, P, Milanezi, F, Ribeiro, AS, Mendes, N, Licastro, D, Karhu, A, Oliveira, MJ, Ligtenberg, M, Hamelin, R, Carneiro, F, Lindblom, A, Peltomaki, P, Castedo, S, Schwartz, S, Jordan, P, Aaltonen, LA, Hofstra, RMW, Suriano, G, Stupka, E, Fialho, AM, Seruca, R 2010, 'Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours', Human Molecular Genetics, vol 19, no. 4, pp. 697-706.

Wei, G, Badis, G, Berger, MF, Kivioja, T, Palin, K, Enge, M, Bonke, M, Jolma, A, Varjosalo, M, Gehrke, AR, Yan, J, Talukder, S, Turunen, M, Taipale, M, Stunnenberg, HG, Ukkonen, E, Hughes, TR, Bulyk, ML, Taipale, J **2010**, 'Genome-wide analysis of ETS-family DNA-binding in vitro and in vivo', **EMBO Journal**, vol 29, no. 13, pp. 2147-2160.

A2 Review in scientific journal

2006

Hautaniemi, S, Vallenius, T, Mäkelä, T 2006, 'Systeemibiologia syöpätutkimuksessa: [katsaus]', Duodecim, vol 122, pp. 2484-2490.

2007

Karhu, A, Aaltonen, LA 2007, 'Susceptibility to pituitary neoplasia related to MEN-1, CDKN1B and AIP mutations: an update', Human Molecular Genetics, vol 16, no. Rev.is, pp. 1, s. R73-R79.

Varjosalo, M, Taipale, J 2007, 'Hedgehog signaling', Journal of Cell Science, vol 120, pp. 36.

2008

Junttila, MR, Li, S, Westermarck, J 2008, 'Phosphatase-mediated crosstalk between MAPK signalling pathways in the regulation of cell survival', FASEB Journal, vol 22, pp. 954-965.

Varjosalo, M, Taipale, J 2008, 'Hedgehog: functions and mechanisms', Genes & Development, vol 22, pp. 2454-2472.

2010

Laakso, MK, Karinen, SH, Lehtonen, RJ, Hautaniemi, S 2010, 'Computational Identification of Cancer Susceptibility Loci', Methods in molecular biology, vol 653, no. Part 1, pp. 87-103.



RC-SPECIFIC TUHAT COMPILATIONS OF PUBLICATIONS DATA 2005-2010

CSB/Aaltonen

A3 Contribution to book/other compilations (refereed)

2007

Aaltonen, LA, Järvinen, HJ 2007, 'Paksusuolisyövälle altistavat geenivirheet ja niiden diagnostiikka', Gastroenterologia ja hepatologia, Duodecim, Helsinki, pp. 544-549.

2009

Monni, O, Hautaniemi, S 2009, 'Bioinformatics of gene expression and copy number data integration', Statistics and Informatics in Molecular Cancer Research, Oxford Scholarship Online Monographs, pp. 78-102.

B1 Unrefereed journal article

2006

Aaltonen, LA 2006, 'Periytyvän kasvainalttiuden molekyylitausta: Äyräpään luento', Duodecim, vol 122, no. 18, pp. 2221-2230.

Arango, D, Alhopuro, P, Aaltonen, LA 2006, 'In response: SMAD4 levels and allelic imbalance in 18q21 in colorectal cancer', Clinical Cancer Research, vol 12, no. 5, pp. 1654-1655.

Echeverri, C, Beachy, P, Baum, B, Boutros, M, Buchholz, F, Chanda, S, Downward, J, Ellenberg, J, Fraser, A, Hacohen, N, Hahn, W, Jackson, A, Kiger, A, Linsley, P, Lum, L, Ma, Y, Mathey-Prevot, B, Root, D, Sabatini, D, Taipale, J, Perrimon, N, Bernards, R **2006**, 'Minimizing the risk of reporting false positives in large-scale RNAi screens', **Nature methods.**, vol 3, no. 10, pp. 777-779.

Vierimaa, O, Georgitsi, M, Lehtonen, R, Vahteristo, P, Kokko, A, Raitila, A, Tuppurainen, K, Ebeling, T, Salmela, PI, Paschke, R, Gundogdu, S, de Menis, E, Mäkinen, MJ, Launonen, V, Aaltonen, LA, Karhu, A **2006**, 'AIP-mutaatiot altistavat aivolisäkkeen adenomalle: [referaatti Science lehden artikkelista]', **Duodecim**, vol 122, no. 12, pp. 1419-1420.

B3 Unrefereed article in conference proceedings

2010

Laiho, M, Jäämaa, SS, af Hällström, TM, Sankila, A, Rantanen, V, Zhang, Z, Yang, Z, De Marzo, AM, Ruutu, M, Andersson, LC **2010**, DNA Damage Recognition via Activated ATM Pathway in Non-proliferating Human Prostate Tissue: Proceedings of the 52nd Annual ASTRO Meeting,, International Journal of Radiation: Oncology - Biology - Physics 78 3; suppl. 1 ELSEVIER INC..

E1 Popular article, newspaper article

2008

Aaltonen, LA 2008, 'Dna-tieto liian tulkinnanvaraista jokamiehen huvitukseksi', Helsingin Sanomat.



RC-SPECIFIC TUHAT COMPILATIONS OF OTHER SCIENTIFIC ACTIVITIES 2005-2010

CSB/Aaltonen

1 Analysis of activities 2005-2010

- Associated person is one of Lauri Antti Aaltonen,		Mervi Aavikko ,	Pia Pauliina Alhopuro,		
	Alexandra Gylfe,		Elina Heliövaara,	Eevi Kaasinen ,	Auli
Inkeri Karhu,	i Karhu , Johanna Kondelin ,		,	Virpi Launonen ,	Heli
Lehtonen,	onen, Miika Mehine,		lina Niittymäki,	Silva Saarinen ,	
Sari Tuupanen,	ari Tuupanen, Pia Marita Vahteristo,		Sampsa Hauta	aniemi ,	Ping Chen,
Sirkku Helena Karinen ,		Marko Kalevi Laakso ,	Anna-Maria Kristiina La	ahesmaa-Korpinen ,	
Riku Louhimo ,		Kristian Ovaska,	Ville Rantanen,		
Vladimir Rogojin,		Jianmin Wu ,	Jussi Taipale ,	Albertus Wilhelm Martir	nus Bonke ,
	Outi Hallikas ,	,	Arttu Jolma,	Songping Li,	
Maria Sokolova,		Mikko Turunen ,	Anna Valpuri Vähä	ärautio, G	onghong Wei,
	Jian Yan				

Activity type

Supervisor or co-supervisor of doctoral thesis	51
Prizes and awards	17
Editor of research journal	4
Peer review of manuscripts	72
Assessment of candidates for academic posts	5
Membership or other role in review committee	2
Membership or other role in research network	6
Membership or other role in national/international committee, council, board	13
Membership or other role in public Finnish or international organization	10
Participation in interview for written media	5
Participation in TV programme	1
Participation in interview for web based media	2



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2 Listing of activities 2005-2010

Supervisor or co-supervisor of doctoral thesis

Lauri Antti Aaltonen,

Doctoral Thesis supervision / Lehtonen R, Lauri Antti Aaltonen, 1999 → 2006, Finland Doctoral Thesis supervision / Ylisaukko-oja, Lauri Antti Aaltonen, 1999 ightarrow 2007, Finland Doctoral Thesis supervision / Hienonen, Lauri Antti Aaltonen, 2000 \rightarrow 2005, Finland Doctoral Thesis supervision / Laiho, Lauri Antti Aaltonen, 2000 → 2005, Finland Doctoral Thesis supervision / Vanharanta, Lauri Antti Aaltonen, 2002 → 2006, Finland Doctoral Thesis supervision / Alhopuro, Lauri Antti Aaltonen, 2003 \rightarrow 2007, Finland Doctoral Thesis supervision / Kokko, Lauri Antti Aaltonen, 2003 \rightarrow 2006, Finland Doctoral Thesis supervision / Lehtonen H, Lauri Antti Aaltonen, 2003 \rightarrow 2008, Finland Doctoral Thesis supervision / Sammalkorpi, Lauri Antti Aaltonen, 2003 \rightarrow 2008, Finland Doctoral Thesis supervision / Vierimaa, Lauri Antti Aaltonen, 2004 \rightarrow 2008, Finland Doctoral Thesis Supervision / Saarinen, Lauri Antti Aaltonen, 2005 $\rightarrow ...,$ Finland Doctoral Thesis supervision / Ahvenainen, Lauri Antti Aaltonen, 2005 \rightarrow 2010, Finland Doctoral Thesis supervision / Georgitsi, Lauri Antti Aaltonen, 2005 \rightarrow 2008, Finland Doctoral Thesis supervision / Koski, Lauri Antti Aaltonen, $2005 \rightarrow 18.06.2010$ Doctoral Thesis supervision / Tuupanen, Lauri Antti Aaltonen, 2005 \rightarrow 2009, Finland Doctoral Thesis supervision / Raitila, Lauri Antti Aaltonen, 2006 → 2009, Finland Doctoral Thesis supervision / Heliövaara, Lauri Antti Aaltonen, 2007 → Finland Doctoral Thesis supervision / Gvlfe, Lauri Antti Aaltonen, 2008 → Finland Doctoral Thesis supervision / Aavikko, Lauri Antti Aaltonen, 2009 → Finland Doctoral Thesis supervision / Kaasinen, Lauri Antti Aaltonen, 2009 \rightarrow ..., Finland Doctoral Thesis supervision / Sirkiä, Lauri Antti Aaltonen, 2010 $\rightarrow \dots$, Finland Auli Inkeri Karhu,

Supervision of doctoral thesis, Auli Inkeri Karhu, 2001 \rightarrow 2005, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2001 \rightarrow 08.10.2005, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 2003 \rightarrow 2007, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 2003 \rightarrow 2008, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2003 \rightarrow 08.06.2007, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2003 \rightarrow 02.11.2008, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2003 \rightarrow 02.11.2008, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 2005 \rightarrow 2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 2005 \rightarrow 2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2005 \rightarrow 12.06.2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2005 \rightarrow 24.10.2008, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2006 \rightarrow 21.12.2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2006 \rightarrow 21.12.2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2006 \rightarrow 21.12.2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2006 \rightarrow 21.12.2009, Finland Supervision of doctoral thesis, Auli Inkeri Karhu, 08.10.2006 \rightarrow 21.12.2009, Finland

Doctoral thesis supervision / Ylisaukko-oja, Virpi Launonen, 1999 \rightarrow 2007, Finland Doctoral thesis supervision / Laiho, Virpi Launonen, 2000 \rightarrow 2005, Finland



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Doctoral thesis supervision / Lehtonen H., Virpi Launonen, 2003 → 2008, Finland Doctoral thesis supervision / Ahvenainen, Virpi Launonen, 2005 → 2010, Finland Doctoral thesis supervision / Lehtonen R., Virpi Launonen, 2006, Finland Sampsa Hautaniemi,

Doctoral thesis supervision / Valiathan, Sampsa Hautaniemi, 2006, United States Doctoral thesis supervision / Laakso, Sampsa Hautaniemi, 2007 $\rightarrow \dots$ Doctoral thesis supervision / Lahesmaa-Korpinen, Sampsa Hautaniemi, 2007 $ightarrow \dots$ Doctoral thesis supervision / Autio, Sampsa Hautaniemi, 24.09.2008, Finland Doctoral thesis supervision / Karinen, Sampsa Hautaniemi, 2008 $\rightarrow \dots$ Doctoral thesis supervision / Chen, Sampsa Hautaniemi, 2009 $\rightarrow \ldots$ Doctoral thesis supervision / Ovaska, Sampsa Hautaniemi, 2009 $\rightarrow \dots$ Doctoral thesis supervision / Rantanen, Sampsa Hautaniemi, 2009 $\rightarrow \dots$ Jussi Taipale .

Doctoral Thesis supervision / Turunen, Jussi Taipale, 01.11.2006 \rightarrow ..., Finland Doctoral Thesis supervision / Jolma, Jussi Taipale, 01.2007 → ..., Finland Doctoral Thesis supervision / Variosalo, Jussi Taipale, 2008, Finland Doctoral Thesis supervision / Yan, Jussi Taipale, 2008 → ..., Finland Doctoral Thesis supervision / Hallikas, Jussi Taipale, 2009, Finland

Prizes and awards

Lauri Antti Aaltonen,

Academy Professor, Lauri Antti Aaltonen, 08.2002 \rightarrow 07.2007, Finland Nominee for Descartes Prize, Lauri Antti Aaltonen, 2005 University of Helsinki's Occupational safety and health award, Lauri Antti Aaltonen, 2005, Finland Matti Äyräpää Prize, Lauri Antti Aaltonen, 2006, Finland Professor of Tumor Genomics, Lauri Antti Aaltonen, 08.2007 → ..., Finland Academy Professor, Lauri Antti Aaltonen, 2009 → ..., Finland ERC Advanced Grant, Lauri Antti Aaltonen, 2010 Sampsa Hautaniemi . The Finnish Medical Foundation 50-years jubileum award, Sampsa Hautaniemi, 2010 Marko Kalevi Laakso, MBI Master's thesis award, Marko Kalevi Laakso, 01.01.2007, Finland Jussi Taipale . Sigrid Juselius Young Investigator Award, Jussi Taipale, 2005, Finland EMBO Young Investigator, Jussi Taipale, 2006 Medix Prize, Jussi Taipale, 2006 Academy Professor, Jussi Taipale, 2008 \rightarrow 2012 Anders Jahre Young Researcher award, Jussi Taipale, 2008 ERC Advanced Grant, Jussi Taipale, 2009 Professor of Medical Systems Biology, Jussi Taipale, 2009 $\rightarrow \ldots,$ Sweden Eric K. Fernström's Prize, Jussi Taipale, 03.11.2010, Sweden



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Editor of research journal

Lauri Antti Aaltonen,

Journal of Medical Genetics, Lauri Antti Aaltonen, 1999 \rightarrow 2005, United Kingdom International Journal of Cancer, Lauri Antti Aaltonen, 2003 \rightarrow 2005, United States Acta Oncologica, Lauri Antti Aaltonen, 2004 \rightarrow 2008, United Kingdom Cancer Genomics & amp; Proteomics, Lauri Antti Aaltonen, 2004 \rightarrow ..., Greece

Peer review of manuscripts

Lauri Antti Aaltonen,

British Journal of Cancer, Lauri Antti Aaltonen, 2005 → 2010 Cancer Research, Lauri Antti Aaltonen, 2005 → 2009 Chromosomes & amp; Cancer, Lauri Antti Aaltonen, 01.01.2005 → 31.12.2005 Clinical Gastroenterology and Hepatology, Lauri Antti Aaltonen, 2005 Gastroenterology, Lauri Antti Aaltonen, 2005 \rightarrow 2009 Genes, Lauri Antti Aaltonen, 01.01.2005 → 31.12.2005 Genes, Chromosomes & amp; Cancer, Lauri Antti Aaltonen, 2005 \rightarrow 2009 Human Molecular Genetics, Lauri Antti Aaltonen, 01.01.2005 \rightarrow 31.12.2006 International Journal of Cancer, Lauri Antti Aaltonen, $2005 \rightarrow 2011$ Journal of Clinical Investigation, Lauri Antti Aaltonen, 01.01.2005 \rightarrow 31.12.2005 Journal of Medical Genetics, Lauri Antti Aaltonen, $2005 \rightarrow 2010$ Journal of Molecular Diagnostics, Lauri Antti Aaltonen, 01.01.2005 \rightarrow 31.12.2005 Journal of the national Cancer Institute, Lauri Antti Aaltonen, 01.01.2005 \rightarrow 31.12.2005 Oncogene, Lauri Antti Aaltonen, $2005 \rightarrow 2020$ Science, Lauri Antti Aaltonen, 01.01.2005 → 31.12.2005 Acta-Dermato Venereologica, Lauri Antti Aaltonen, 2006 Acta-Oncologica, Lauri Antti Aaltonen, 2006 British Journal of Dermatology, Lauri Antti Aaltonen, 2006 Clinical Cancer Research, Lauri Antti Aaltonen, 2006 → 2010 Human Mutation, Lauri Antti Aaltonen, 2006 → 2009 Journal of Clinical Endocrinology and Metabolism, Lauri Antti Aaltonen, 2006 → 2008 Molecular and Cellular Oncology, Lauri Antti Aaltonen, 03.2006 \rightarrow 05.2006 Nature Clinical Practise Oncology, Lauri Antti Aaltonen, 2006 Proceedings of the National Academy of Sciences, Lauri Antti Aaltonen, 2006 \rightarrow 2010 Trends in Molecular Medicine, Lauri Antti Aaltonen, 2006 Cancer Detection and Prevention, Lauri Antti Aaltonen, 2007 Journal of Pathology, Lauri Antti Aaltonen, 2007 Lancet Oncology, Lauri Antti Aaltonen, 2007 Nature, Lauri Antti Aaltonen, 2007 \rightarrow 2010 New England Journal of Medicine, Lauri Antti Aaltonen, 2007 \rightarrow 2010 Molecular Genetics and Metabolism, Lauri Antti Aaltonen, 2008 EMBO Journal, Lauri Antti Aaltonen, 2009 European Journal of Endocrinology, Lauri Antti Aaltonen, 2009



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Familial Cancer, Lauri Antti Aaltonen, 2009 → 2010 Hormone Research, Lauri Antti Aaltonen, 2009 Journal of the American Medical Association, Lauri Antti Aaltonen, 2009 Lancet, Lauri Antti Aaltonen, 2009 Nature Genetics, Lauri Antti Aaltonen, 2009 → 2010 PLoS Biology, Lauri Antti Aaltonen, 2009 \rightarrow 2010 Clinical Genetics, Lauri Antti Aaltonen, $01.01.2010 \rightarrow 31.12.2010$ Auli Inkeri Karhu, Expert Review of Endocrinology & amp; Metabolism, Auli Inkeri Karhu, 08.10.2008 European Journal of Endocrinology, Auli Inkeri Karhu, 08.10.2009 International Journal of Cancer, Auli Inkeri Karhu, 08.10.2010 Virpi Launonen . British Journal of Cancer, Virpi Launonen, 01.01.2004 \rightarrow 31.12.2006, United Kingdom Oncogene, Virpi Launonen, 19.09.2005 → 31.12.2005 Genetic Disease Online Reviews, Virpi Launonen, 12.04.2006 → 31.12.2006, United States Journal of Dermatological Science, Virpi Launonen, 04.09.2006 → 31.12.2006, Japan Nature Clinical Practice Urology, Virpi Launonen, 17.10.2006 \rightarrow 31.12.2006, United States Sampsa Hautaniemi, BMC Bioinformatics, Sampsa Hautaniemi, 2003 $\rightarrow \dots$ BMC Genomics, Sampsa Hautaniemi, 2003 $\rightarrow \dots$ BioTechniques, Sampsa Hautaniemi, 2003 $\rightarrow \dots$ Bioinformatics, Sampsa Hautaniemi, 2003 $\rightarrow \dots$ Clinical Chemistry, Sampsa Hautaniemi, 2005 $\rightarrow \dots$ Data Mining and Knowledge Discovery, Sampsa Hautaniemi, 2005 $\rightarrow \dots$ FEBS Letters, Sampsa Hautaniemi, $2005 \rightarrow \dots$ IEEE Transactions on Signal Processing, Sampsa Hautaniemi, 2005 $\rightarrow \dots$ Neurocomputing, Sampsa Hautaniemi, 2005 $\rightarrow \dots$ Jussi Taipale , Development, Jussi Taipale, 2005

Molecular Cancer Research, Jussi Taipale, 2005 \rightarrow 2006 Current Biology, Jussi Taipale, 2006 Developmental Cell, Jussi Taipale, 2006 \rightarrow 2010 Cell Stem Cell, Jussi Taipale, 2007 Nature Cell Biology, Jussi Taipale, 2007 Nature Methods, Jussi Taipale, $2007 \rightarrow 2010$ Cancer Cell, Jussi Taipale, 2008 Nature, Jussi Taipale, 2008 PLOS Biology, Jussi Taipale, 2008 Genes & amp; Development, Jussi Taipale, 2009 → 2010 Molecular Systems Biology, Jussi Taipale, 2009 Cell, Jussi Taipale, 2010



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CSB/Aaltonen

Genome Research, Jussi Taipale, 2010

Proceedings of the National Academy of Sciences, Jussi Taipale, 2010

Assessment of candidates for academic posts

Lauri Antti Aaltonen,

Examiner for Professorship, Lauri Antti Aaltonen, 2005, United States Examiner for Docentship / Mottagui-Tabarin, Lauri Antti Aaltonen, 2006, Finland Examiner for Professorship / Sorsa, Lauri Antti Aaltonen, 2009, Finland

Jussi Taipale ,

Reviewer for Docentship, Jussi Taipale, 2007, Finland Reviewer for Professorship, Jussi Taipale, 2009, Canada

Membership or other role in review committee

Lauri Antti Aaltonen,

Member of the Finnish Cancer Society's Grant Review Board, Lauri Antti Aaltonen, 2004 \rightarrow 2007, Finland

Sampsa Hautaniemi ,

Member of the scientific evaluation board for HPC-Europa2, Sampsa Hautaniemi, 2009 ightarrow ...

Membership or other role in research network

Lauri Antti Aaltonen,

Membership in the Nordic Center of Excellence in Disease Genetics, Lauri Antti Aaltonen, 2000 \rightarrow ..., Finland

Membership in the Center of Excellence in Translational Genome-Scale Biology, Lauri Antti Aaltonen, 2006 \rightarrow 2011, Finland Sampsa Hautaniemi,

Board membership in Quantitative biology infrastructure network, Sampsa Hautaniemi, 2007 \rightarrow 2008, Finland

Chairman of the Bioinformatics infrastucture network, Sampsa Hautaniemi, 2009 \rightarrow 2012, Finland

Jussi Taipale,

Membership in the Nordic Center of Excellence in Disease Genetics, Jussi Taipale, $2005 \rightarrow ...$ Membership in the Center of Excellence in Translational Genome-Scale Biology, Jussi Taipale, $2006 \rightarrow 2011$, Finland

Membership or other role in national/international committee, council, board

Lauri Antti Aaltonen,

Membership in the EACR Committee for Central and Eastern Europe, Lauri Antti Aaltonen, 1999 $ightarrow \dots$

Membership in the Danish Cancer Society's Scientific and Medical Advisory Board, Lauri Antti Aaltonen, 2003 → 2006, Denmark

Membership in the EMBO Science & amp; Society Committee, Lauri Antti Aaltonen, 2003 \rightarrow 2006

Academy of Finland, Lauri Antti Aaltonen, 01.01.2005 \rightarrow 31.12.2005, Finland

Board membership in Viikki Doctoral Programme in Molecular Biosciences, Lauri Antti Aaltonen, 01.01.2005 → 31.12.2005, Finland Finnish Cancer Organisations, Lauri Antti Aaltonen, 01.01.2005 → 31.12.2005, Finland

Board membership in Helsinki Biomedical Graduate School (HBGS), Lauri Antti Aaltonen, 2008 ightarrow..., Finland

Sampsa Hautaniemi,

Board membership in Bioinformatics Communities Committee at ESFRI project ELIXIR, Sampsa Hautaniemi, 2008 \rightarrow ...

Deputy membership in the Institute of Biotechnology steering board, Sampsa Hautaniemi, 2010 \rightarrow 2014, Finland

Jussi Taipale,

Invited member of the scientific advisory board of the National Hellenic Research Foundation, Jussi Taipale, 2007 → ..., Greece



RC-SPECIFIC TUHAT COMPILATIONS OF OTHER SCIENTIFIC ACTIVITIES 2005-2010

CSB/Aaltonen

Board membership in Graduate School in Computational Biology, Bioinformatics and Biometry (ComBi), Jussi Taipale, 2008 \rightarrow ..., Finland

Board membership in Helsinki Graduate School in Biotechnology and Molecular Biology (GSBM), Jussi Taipale, 2008 → 2009, Finland Board membership in Finnish Graduate School in Computational Sciences (FICS), Jussi Taipale, 01.01.2010 → ..., Finland

Membership or other role in public Finnish or international organization Lauri Antti Aaltonen ,

Membership in Duodecim, Lauri Antti Aaltonen, 1986 $\rightarrow \dots$, Finland

Membership in the Europan Association for Cancer Research, Lauri Antti Aaltonen, 1997 \rightarrow ...

Membership in the European Molecular Biology Organization (EMBO), Lauri Antti Aaltonen, 2000 → ...

Membership in Biocentrum Helsinki, Lauri Antti Aaltonen, 2001 → ..., Finland

Membership in the Finnish Academy of Science and Letters, Lauri Antti Aaltonen, 2002 \rightarrow ..., Finland

Director of Genome Scale Biology Research Program, Lauri Antti Aaltonen, 2009 → ..., Finland

Membership in the European Academy of Cancer Sciences, Lauri Antti Aaltonen, 2010 \rightarrow ...

Sampsa Hautaniemi,

Deputy member of the Faculty of Medicine Senate, Sampsa Hautaniemi, 2009 \rightarrow 2012, Finland Vice director (infrastructures and research training) of the Research Programs Unit, Sampsa Hautaniemi, 2010 \rightarrow 2012, Finland Jussi Taipale ,

Membership in Biocentrum Helsinki, Jussi Taipale, 2007 $\rightarrow ...$, Finland

Participation in interview for written media

Lauri Antti Aaltonen,

Interviews in newspaper / Helsingin Sanomat, Lauri Antti Aaltonen, 06.01.2005 → ..., Finland

Sampsa Hautaniemi,

Magazine and journal interviews, Sampsa Hautaniemi, 2007 $\rightarrow \dots$

Interviews for newspaper / Helsingin Sanomat, Sampsa Hautaniemi, 2008 $\rightarrow \ldots$, Finland

Jussi Taipale ,

Interview in a newspaper / Helsingin Sanomat, Jussi Taipale, 23.02.2006, Finland Interview in the Journal of Cell Biology, Jussi Taipale, 28.07.2008

Participation in TV programme

Lauri Antti Aaltonen,

Interview in YleTeema, Lauri Antti Aaltonen, 30.11.2005, Finland

Participation in interview for web based media

Lauri Antti Aaltonen,

Participation in Radio Helsinki podcast, Lauri Antti Aaltonen, 14.11.2005, Finland

Sampsa Hautaniemi,

Invited blog posting, Sampsa Hautaniemi, 2010



Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010 by CWTS, Leiden University, the Netherlands

Research Group: Aaltonen L

Basic statistics

Number of publications (P)	143
Number of citations (TCS)	2,649
Number of citations per publication (MCS)	18.72
Percentage of uncited publications	18%
Field-normalized number of citations per publication (MNCS)	1.83
Field-normalized average journal impact (MNJS)	2.00
Field-normalized proportion highly cited publications (top 10%)	2.42
Internal coverage	.95

Trend analyses



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2005-2008



2007-2010

2006-2009



0.00

MNJS

2005-2008

2006-2009

2007-2010

Performance (MNCS) by collaboration type



Web of Science(WoS)-based bibliometrics of the RC's publications data 1.1.2005-31.12.2010 by CWTS, Leiden University, the Netherlands

Research profile



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