



Europe Biobank Week

Abstracts book

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Topic 3: Environment, Biodiversity & Human Health

Title: Promises and pitfalls in the use of biobank resources

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This presentation will give a few real-live examples of cross-border use of biobank samples and data from the BBMRI-LPC project, discuss the tension between theory and practice of data sharing and highlight some of the results and promises.

Title: Big data and biobanking: from the Moli-sani project to the Neuromed-integrated health life platform

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Introduction: The Moli-sani project is a prospective, population-based cohort study which has randomly enrolled, between 2005 and 2010, 24,345 adults, from Molise municipalities to investigate genetic and environmental determinants of cardio-cerebro-vascular/neurodegenerative/cancer disorders. Follow up: incident events of hospital admission/death by regional data base; since 2017, recall of the cohort.

Material and methods: Special attention was given to socio-economic conditions and life habits with focus on Mediterranean diet and other nutrition variables. Anthropometric variables, blood pressure, electronic ECG and spirometry recordings were measured. All parameters have been taken in digital format SCP and processed by systems of computerized analysis.

Results: The parameters measured per each subject yielded a huge number of variables: Clinical history 2100; Family history 841; ECG 617; Spirometry 153; Biomarkers 592; Dietary habits 1,600; follow-up 680 for a total number per subject 6,583 and Grand Total 160,155,139. A biobank (Molibank) of biological samples (over 700,000 specimens, 28 per each subject) has been established with highly sophisticated systems for control, security, storage and redistribution of the samples. Such biobank has already allowed collaborations with International Consortia (NCDs, BioMarCare, EPICOR and others) for the discovery and measurement of new biochemical, genetic and epigenetic biomarkers of chronic disorders.

Conclusion: Moli-Sani offers an excellent scaffold for a big data approach towards personalised health and medicine. Based on the Moli-sani experience, we have recently developed a project called Neuromed-integrated platform/biobank to collect clinical/diagnostic/environmental data from all patients admitted to the hospitals of the Neuromed Group (over 40,000 admissions in 2016).

Title: How can clinical trials build capacity: the first year of PREVAC (Partnership for Research on Ebola VACcination) biobank in Guinea

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Introduction: The last Ebola epidemics showed the need for coordination of specimens storage during outbreaks. Guinea had engaged for establishing a national biobank.

PREVAC is a multi-centric phase II clinical trial evaluating two recombinant vaccines against Ebola run in Guinea, Liberia, Mali and Sierra Leone. Each site required local biobanks.

Material and methods: In the two PREVAC study sites in Guinea, clinical trial biobanks were set-up. In order to produce a sustainable biobank model for the country, training sessions, collaborations with the ministry of health for governance and for the continuation of the biobank after the trial were put in place.

Results: During the first year of activities 51613 samples were collected, no major temperature excursion were recorded, 10 Guinean laboratory technicians were trained on biobank management. Internal quality controls were performed on samples storage : the site of Conakry (urban) showed 100% compliance while the site of Maferinyah (rural) showed a 99,9% compliance. After the first year of collection a first batch was shipped from Guinea to Liberia where the primary endpoint will be determined with an Ebola-specific ELISA. We operated a ground shipping, showing the feasibility of samples sharing at high quality and acceptable costs between African countries.

Conclusion: The PREVAC biobank in Guinea is showing that a high quality biobank in a low resource setting can be achieved. Besides the initial installation costs, the PREVAC biobank is showing economic and logistic sustainability. Finally, it is building local capacity which will support the set-up of the national biobank.

Title: Use of large pedigrees from the FarGen project to gain information on consanguinity and inbreeding within the Faroese population.

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Introduction: The Multi-Generation register of the Genetic Biobank comprises hereditary records of all individuals registered in the Faroe Islands since 1800; the majority of the individuals (85%) have records dating back to 1650. The FarGen project is using the genealogy records to show consanguinity and inbreeding within the Faroese population.

Material and methods: Pedigrees were made from all 1530 individuals who had registered as FarGen participants. All the participants are in the Multi-Generation register and have answered a questionnaire about health status. Three pedigrees were constructed: 1) all 1530 participants, 2) participants with "good" health status, and 3) participants with "bad" health status.

Results: We present three large family pedigrees spanning seven generations, with data from group pedigrees with "good" and "bad" self-reported health status. From the 21 different ICD-10 groups of diseases reported in the FarGen project, we present 15 family pedigrees with specific disease status, which may contribute to our understanding of the pathogenesis of these diseases. We show whether consanguinity reflects the outcome, and trace its origins to the different islands on the Faroe Islands.

Conclusion: The pedigrees will be used in combination with genotype data to gain information on population stratification and the demographical history of the Faroe Islands.



Topic 4A: Impact of Policies and General Data Protection Regulation (GDPR) on Biosharing

Title: An approach to determine biobank GDPR compliancy aspects for a federative Biobank organization as the Dutch Parelsnoer Institute

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Introduction: The legal status of a federative biobank is determinative for the role of data processor/controller in the context of the General Data Protection Regulation (GDPR). Because the Parelsnoer Institute (PSI) involves eight university medical centers, a specific approach to determine its GDPR role as an umbrella biobank organization was assessed.

Material and methods: A GDPR compliancy web application was used, that was especially developed by BBMRI-NL to register processing activities related to the responsibilities of controller and processor for biobank organizations. The questionnaire was assessed by the IT and security coordinator, the legal advisor (privacy officer) and the operational manager of PSI.

Results: The performed analysis gave PSI as an organization insights into all the relevant GDPR articles and it helped to determine the responsibilities between PSI and the collaborating UMCs. Judgement on who is controller or processor was difficult and is very dependent of the legal status of the biobank institution and the way it interacts with health care institutions and principal investigators. The results were transferred as guidance to the Privacy Officers of the Dutch UMCs, which together are the controllers for the biobank of PSI.

Conclusion: Performing a GDPR analysis from a federative biobank perspective was not only informative for PSI but also for the partner institutes and the involved researchers. It addresses important issues such as register requirement and responsibilities. The GDPR DPIA & compliance web application was a valuable tool for this.

Title: Data in Question: Survey Results on ELSI Challenges in Biobank-Based Research

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Introduction: Researchers and stakeholders in the field of biobanking are facing different social, ethical and legal challenges that impact established practices related to biological samples and health-related data handling and sharing. Rethinking and improving informed consent is one of the key challenge in this regard.

Material and methods: BBMRI-ERIC Common Service ELSI, in collaboration with the COST action CHIPME, FP7 project RDconnect, H2020 project ADOPT BBMRI-ERIC, the IMI project DO-IT, and Biobank Norway conducted a survey that aimed to identify challenges arising from legal, ethical or social developments from the perspective of European biobankers and biomedical science researchers.

Results: The key focus of enquiry was the effect of the growing demand for engaging with third parties from patients, citizens and industry. The major topics covered by the survey are: (1) secondary use of data (2) informing and/or re-contacting participants (3) sharing of data with third parties from industry, (4) participant engagement, and (5) collaboration with industrial partners. Closing end of March 2018, the sample collected represents the experiences and opinions of 200 biobankers from 25 European countries, collected via an

online questionnaire fielded through the BBMRI ERIC research infrastructure network of biobankers and other associated networks.

Conclusion: The presentation will outline and discuss key results in the context of new legislation such as the EU General Data Protection Regulation, and the growing demand for engaging systematically with third parties.

Title: Legal – but also acceptable? Empirical study on healthcare-embedded biobanking without consent in light of the General Data Protection Regulation

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Introduction: Although aiming at European harmonization, the General Data Protection Regulation leaves some flexibility for adapting its framework for scientific research on national level. Further debate on how to enable effective international cooperation in data-rich medicine will thus be necessary. This should include attention to the acceptability of various regulatory options.

Material and methods: In an empirical questionnaire study 700 adult patients were approached at the Comprehensive Center for Inflammation Medicine (CCIM) in Kiel, Germany, in spring 2018. They were asked about their views on the use of left-over data and biomaterial from their clinical stay or visit for research purposes without explicit consent.

Results: While Germany has adapted the GDPR so that its national law now allows, under certain conditions, secondary data use for scientific or historical research and statistical purposes under certain conditions without consent, other countries are developing stricter regulations. Different international adaptations of the GDPR, different practices of information and consent necessitate discussing recommendations for researchers how to implement national regulations locally and enable collaboration internationally. Such debate should also be based on empirical evidence of the attitudes of populations involved in future research.

Our study examines the option of secondary use of biomaterial and data within healthcare-embedded biobanking without consent.

Conclusion: To the best of our knowledge this is the first study in Germany which adds a patient-oriented view to the discussion about the acceptability of secondary use of data for medical research without consent. In the paper, we present main findings and discuss their implications.

Title: Polish code of conduct for processing personal data on biobanking

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Introduction: In Poland, there is no specific legal regulation of the operation of biobanks and processing of personal data by biobanks. The commencement of the GDPR application resulted in the necessity to prepare a code of conduct regarding the processing of personal data by biobanks in Poland.

Material and methods: The presentation will be about the analysis of code of conduct for Polish biobanks provisions. The Code was created by BBMRI.pl based on dialogue with its participants and the analysis of polish regulations. It is under public consultation with representatives of biobanks and patient organizations and Data Protection Polish Authority.

Results: The code specifies e.g.: rights of persons whose data are processed, data controller's (biobank) duties, principles of acceptability of primary and secondary use of data for scientific purposes or rules for processing data of



deceased persons. The code is written in an accessible form-for each principle is an explanation and an example of the biobanking activities. It's includes the recommended consent forms for processing data and data processing registers.

The entire code will be subject to approval by the President of the Data Protection Office and will be binding for entities that will join it.

Conclusion: The Polish Code of Conduct constitutes the first common standard for the protection of personal data processed by biobanks. Regulations will take into account the BBMRI-ERIC ELSI group's, OECD's guideline's and WMA's declaration's standards. The Code can be universal and enable the exchange of data between polish and European's biobanks.



Topic 4B: Animal and Agricultural Biobanking and Preservation

Title: Adventure Biobanking – Citizen Science ‘Rangers’ and their Role in the Future Survival of Wildlife Populations

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South Africa has the world’s third richest biodiversity and hence has a natural advantage that presents a unique opportunity to generate a strong mass of scientific expertise. A Wildlife Biobank forms part of a Biodiversity Biobank, representing a geographically unique biomaterial resource that should be managed to the benefit of conservation and society. The acquisition, processing and utilization of wildlife genetic resources and the banking of such resources is a complex series of events, requiring expert input representing multiple disciplines and stakeholders. The citizen science ‘rangers’ role is critical to the success of acquiring quality wildlife biomaterials derived under difficult field conditions. The presentation will look at the adventures of collecting wildlife biomaterials in the field and lobby for expanding the role of citizen science rangers in wildlife biobanking.

Title: Banking on it! A dedicated biobank by and for the european zoos

Zjef Pereboom, Danny de Man, Helen Senn, Tania Gilbert, Philippe Helsen, Joerns Fickel, Andrew Kitchener, Imke Lüders, Baptiste Mulot and Christina Hvilson

The membership of the European Association of Zoos and Aquaria (EAZA) has recently established dedicated biobanking facilities for the European zoo community. This EAZA Biobank aims to be a primary resource for supporting population management and conservation relevant research, and will be an invaluable resource for zoo and wildlife veterinarians and breeding programme managers. The EAZA Biobank aims to hold blood / DNA / tissue and serum from as many individual animals in European zoos, and is designed such that samples are registered in the global zoos’ Zoological Information Management System (ZIMS) and are primarily available to benefit efforts ensuring the genetic and physical health of intensively managed zoo populations. These biological samples enable detailed genetic and genomic analyses, which is rapidly becoming a key tool to increase the chance of success of the European Ex-situ Programmes (EEPs), for example by improving the knowledge of relatedness and paternity issues, resolving taxonomic uncertainty, identifying the origin of individuals to help set up the correct breeding groups, and ensure that, as far as possible, captive populations represent the genetic diversity of their wild counterparts. Additionally, the unique coupling of whole blood and serum samples will enable medical / epidemiological research of interest for veterinarians, and enable e.g. disease surveillance of infectious diseases at small- as well as meta-population level, enable retrospective surveys, veterinary molecular diagnostics and identifying genetically inherited diseases. The EAZA Biobank is jointly hosted and funded by Copenhagen Zoo, RZSS Edinburgh Zoo & National Museums of Scotland, the Leibniz Institute for Zoo and Wildlife Research in Berlin, and the Antwerp Zoo Society, and works in close collaboration with similar initiatives.

Title: State of the art and prospects of cryobanking crop genetic resources

Bart Panis (1)

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As the expanding world population depends on a dwindling number of cultivars and of crop species, the concomitant loss of crop diversity increasingly threatens global food security. Therefore crop diversity and associated wild relatives urgently need to be conserved for future generations.

Seed conservation at -20°C is the most used ex situ method to conserve crop germplasm. However it is not an option for those crops that are sterile (do not produce viable seeds, like banana), or produce only recalcitrant (non-storable) seeds (like cocoa and coconut). Nor is it an option for species where specific gene combinations need to be maintained during propagation (potato and many fruit species such as apple). In such cases, vegetative material needs to be maintained in the field or in in vitro collections (micro plants grown in test tubes). Cryopreservation, or storage of biological material at ultra-low temperatures, is, however, the preferred method for the long-term conservation of plant genetic resources of vegetatively propagated crops.

Roughly, three main cryopreservation protocols can be distinguished for hydrated tissues: i) the “classical” slow freezing protocol (currently mainly applied to non-organized tissues such as cell suspensions and calluses); ii) the encapsulation/dehydration method that relies on synthetic seeds and iii) the methods relying on the application of highly concentrated vitrification solutions such as PVS2 (Plant Vitrification Solution 2). The droplet vitrification protocol was established because it combines the application of highly concentrated cryoprotective vitrification solutions with ultra-fast freezing and thawing. This method can now be considered as the first “generic” cryopreservation method for plant tissues, as it has now been successfully applied to different tissues (shoot cultures/embryos) and a wide variety of plant species from different climatic environments. Moreover, the technique is less cumbersome in its application compared with other cryopreservation methods and no sophisticated equipment is needed.

Currently, over 10,000 accessions starting from in vitro cultures are safely preserved for the long term through cryopreservation. More than 80 % of these belong to 5 crops; potato, cassava, bananas, mulberry and garlic. Other important cryopreservation collections representing thousands of accessions are those of dormant apple buds.

A recent study recommends to establish safety back-up facility for 5,000-10,000 cryopreserved crop accessions arising the different cryopreservation activities. It would act as a complementary facility to the Svalbard Global Seed Vault, in the Arctic tundra, which conserves crops that reproduce through ‘storable’ seeds. With these two facilities, the majority of existing crop diversity – tens of thousands of species and varieties of all food crops and wild relatives – will be preserved for present and future generations.

Title: NASC: Finding the balance between funding, cost-recovery and customer service

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Introduction: The Nottingham Arabidopsis Stock Centre (NASC) collects, preserves, reproduces and distributes diverse seed and other stocks of the model plant *Arabidopsis thaliana* and related species for research and education. NASC’s seed collection approaches one million stocks including insertion lines covering 28,937 genes and over 1,500 distinct natural accessions.

Material and methods: The number of seed and DNA stocks sent annually is now over 100,000, a rate that substantially exceeds anything imagined in the beginning. Throughout the years, NASC has used different funding/cost-recovery models based on increasing distribution numbers as well as customer feedback.

Results: Sustained funding is the greatest challenge faced by modern biobanks. Budgetary shortfalls as a result of the current economic situation have forced biobanks to rely on their cost-recovery/commercialization models to complement their grant/government funding. At the same time, biobanks have to improve and maintain high customer service standards in order to stay ahead of shifting customer expectations. Finding the balance between these three key elements (funding, cost-recovery and customer service) can be difficult and demanding. NASC has traditionally been funded by the Biotechnology and Biological Sciences Research Council (BBSRC) with a proportion of operational costs also covered through user fees.

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Conclusion: This combination of government funding and cost-recovery represents a reasonable balance between customer services provided and financial support required. We believe NASC's current funding strategy represents a useful model that can be considered when planning for similar resources to support research with other organisms.



Topic 4C: Quality Assessment and Management of Samples and Data

Title: Standardized and Improved Pre-analytical Workflows: A Key for Reliable Diagnostics, Research and Biobanking

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Molecular in vitro diagnostics and research have allowed great progress in medicine. Further progress is expected by new biomarker tests analyzing specimens' biomolecule profiles such as nucleic acids, proteins, and metabolites. However, profiles of these molecules can change significantly during specimen collection, transport, storage, and processing, caused by post collection cellular changes such as gene inductions, gene down regulations, biomolecules modifications or degradation. This can make the outcome from diagnostics or research unreliable or even impossible because the analytical test will not determine the situation in the patient body but an artificial specimen analyte profile generated during the pre-analytical workflow. High quality specimens with preserved analyte profiles are therefore crucial.

The EU SPIDIA Consortium (2008-2013) developed new pre-analytical technologies and generated broad evidence that guidance to laboratories on pre-analytical workflows improves analytical test results. Based on these results, the CEN/TC 140 for "in-vitro diagnostic medical devices" has released first 9 European Technical Specifications for pre-analytical workflows addressing different blood, other body fluids and tissue based molecular applications. They are currently progressing to International Standards at the ISO/TC 212 for "clinical laboratory testing and in vitro diagnostic test systems". The new SPIDIA4P consortium project (2017-2020) will broaden to a final portfolio of 22 pre-analytical CEN and ISO Standards, as well as corresponding External Quality Assurance (EQA).

The SPIDIA project received funding from the EU's FP7 under grant agreement no. 222916. The SPIDIA4P project receives funding from the EU's Horizon 2020 research and innovation program under grant agreement no. 733112.

Title: Quality assessment and management of clinical data within the Dutch Parelnoer Institute

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The Parelnoer Institute (PSI) is a collaborative biobanking project of all eight University Medical Centers in the Netherlands which was launched in 2007. Currently PSI covers 18 large disease specific cohorts (the so-called 'Parels' or 'Pearls') and several new 'Pearls' are being developed. PSI currently (July 2018) has stored more than 600,000 biospecimens with annotated clinical data of more than 35,000 patients.

PSI provides an infrastructure for storage and delivery of biomaterials and associated clinical data for the clinical entities. Additionally, PSI offers opportunities such as iterations/flexibility in the disease-specific information models, linkage with other clinical registries and large cohorts, and enrichment of the data collection with results from for instance GWAS studies. Furthermore, PSI works according to the FAIR principle in order to be future-proof.

Within the PSI infrastructure, clinical data are hosted in a validated web based application (ProMISE) that fulfils international standards for data management, quality assurance, and privacy protection using a Trusted Third Party. Essential elements for adequate data quality that PSI advocates are: limited number of elements in the information model to what is necessary for future research, integrated data collection in the routine of the EHR, usage of international standards (Detailed Clinical Models), simple and logical instructions for data collection and registration, mandated and authorized dedicated research employees, and quality control measurements, reports and dashboards within the data management system for the participating groups in order to optimize data quality.

Title: Five years of QM-harmonization in BBMRI.at – Review and Outlook

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Introduction: Irreproducible research represents a huge financial burden, wastes the time of patients and scientists and can be potentially harmful for study subjects. Low-quality biomaterial is a major source of irreproducible research data. Hence, BBMRI.at has elaborated measures and taken decisive actions to increase sample and data quality in Austrian Biobanks.

Material and methods: BBMRI.at work-package "quality management" defined the following milestones: a) to facilitate establishment/improvement of ISO9001-based quality management systems (QMS), b) to harmonize processes based on international, evidence-based documents (e.g. CEN Technical Specifications (CEN/TS) for preexamination processes), and c) to establish intra-consortial cross-audits. Data from seven BBMRI.at partner biobanks/biobank sections is reported.

Results: At the end of BBMRI.at project phase I, six of the seven BBMRI.at partner biobanks/biobank sections were included in an ISO9001-based QMS, and the remaining facility was in the process of implementation. Likewise, realization processes in 6/7 partner biobanks were compliant with a common process description based on the preanalytical technical specifications issued by CEN/TC140. Furthermore, the first round of intra-consortial ISO9001:2008 cross-audits was successfully completed at all BBMRI.at biobanks.

Conclusion: Harmonization of quality-related efforts offers considerable advantages, especially when it comes to comparability of biomaterial/-data from different biobanks. BBMRI.at provides a good platform for common improvement. In a next step, the work-package aims to guide its partner biobanks towards implementation of the biobank-specific standard ISO20387 in a changing regulatory environment.

Title: German Biobank Alliance – Ring trial concept "Liquid" as part of the Quality Management system for Biobanks

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Introduction: The development of a quality management (QM) system for the German biobank community, compliant with international standards, is a central task for the German Biobank Alliance (GBA). To this end, a ring-trial and



a quality assurance concept shall be established by evaluating reliable quality control (QC) biomarkers for body fluids.

Material and methods: To validate and refine the processes across the participating eleven GBA biobanks a ring-trial concept was designed to control the quality of liquid biospecimen and DNA preparations from whole blood. This includes the analysis of reference samples and samples from disease groups using defined pre-analytical conditions.

Results: The quality of DNA preparations from whole blood across all biobanks was assessed in a ringtrial using reference materials supplied by the Integrated Biobank of Luxembourg (IBBL).

In order to evaluate the quality of human plasma and serum samples, an extensive literature review was performed. Out of 548 metabolites identified, a QC-biomarker panel of 27 metabolites was selected. This panel will be tested in a ring-trial, which was approved by the ethics committees of all eleven participating sites, with healthy volunteers and patients with different diseases such as diabetes, multiple sclerosis, heart failure and cardiovascular diseases.

Conclusion: GBA aims at establishing a well-organized workflow for sample logistics and a comprehensive data management system to collect and manage data from collaborating biobanks as well as to provide consistently high-quality samples by means of a reliable and economic QM-system for biobanking of human blood to enable high-quality research.

Title: Processing, Storage and Management Systems of Cell Samples in TMM Biobank

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Introduction: The biobank of Tohoku Medical Megabank Project (TMM biobank) is the first large scale population-based biobank in Japan with a collection of approximately 3 million tubes of biospecimens and associated data obtained from over 150,000 participants.

Material and methods: In addition to liquid samples including serum, plasma, urine and milk, we also have isolated and stored PBMCs (peripheral blood mononuclear cells) manually from most of the participants. We are also establishing Epstein-Barr virus (EBV)-transformed lymphoblastoid cell lines (LCLs) and activated T cells with CD3/CD28 and IL-2 from cryopreserved PBMCs.

Results: So far EBV-LCLs from more than 2,400 participants have been established and activated T cells from more than 2,800 participants with whole-genome information have been stored. The overall efficiency of EB-LCLs establishment is 85.1%. We have been using a customized LIMS system for the management of PBMC isolation and establishment of EBV-LCLs. In order to confirm the samples' integrity, we performe MassARRAY analysis of our DNA specimens from EBV-LCLs and activated T cells and conduct collation of the genomic data.

We set up the system to deliver these cellular materials with reliable genome sequence data and/or associated individual's health data.

Conclusion: We consider that they are not only important resources of individual genomes but also valuable research tool for functional analysis such as toxicological assay or analyzing the individual epigenetic signature, RNAs, metabolites and proteins, which would help deciphering causes of diseases or finding better cures based on individuals' genetic backgrounds.

Title: New Data Quality BIRT-Reports for Biobanks

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Introduction: Human Biobanks are collections of high-quality human biological material, donor-specific data, and sociodemographic information about the donor. Sample-specific data such as harvesting time, processing conditions or sample type should all be documented in a Biobank Management System.

Material and methods: The hospital-integrated biobank ICB-L uses the commercial software CentraXX from KAIROS ® to enable sample and data management. For guaranteeing high data quality, we developed data quality BIRT (Business Intelligence and Reporting Tools) -reports based on SOPs or study-specific requirements and made it available in CentraXX.

Results: The developed BIRT-Reports are based on more than 80 items and reach far beyond a simple completeness check. Discrepancies regarding dates, organizational management information as well as administrative sample information according to the predetermined SOPs (Standard Operating Procedure) are pointed out. The quality reports are automatically executed by the system at defined time points or can be initiated manually if required. A regular and intensive exchange between biobank- and IT-management staff ensures a constant adaption and further developments of reports.

Conclusion: Applying our BIRT-report, a high degree of security for complete and accurate data is achieved and communicated transparently. Data quality is further improved by the consistent use of barcodes and scanners, as well as by increased use of workflows being modelled with the help of bpm (business process modelling) processes.



Topic 5A: Achieving Long-Term Sustainability in Biobanking

Title: Value and sustainability in biobanking: insights from Karolinska Institutet Biobank

Mark Divers (1)

(1) Karolinska Institute Biobank, Sweden

The most important role of biobanks of human samples is to support improved healthcare and public health. Demonstrating this is becoming more important and recognized as a vital element in achieving biobank sustainability. This talk will highlight a case study from KI Biobank of long-term impact of research based on active use of biobanking, and will offer some reflections on one biobank facility's long journey in sustainability.

Title: Can a Sustainable Biobank Infrastructure Help to Scale Sample Collections for Precision Medicine ? A Companion Diagnostics Use Case

Oliver Karch (1)

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The goal of achieving long-term sustainability in Biobanking poses different challenges when comparing institutional / academic biobanks to Biobanks run by Pharmaceutical enterprises. While large infrastructure investments of institutional Biobanks are often lacking sustainable funding, clinical Biobanks at Pharma companies are expected to match demands of quickly changing research focus which may render utilization and scalability of retrospective sample collections difficult. On the other hand, clinical research in precision medicine is often hampered by availability of high quality samples which have been characterized at molecular level, e.g. to capture rare mutations to be used as stratification markers. Consequently, screening for cancer subtypes at individual hospital level is unlikely to provide sufficient sample material for development of companion diagnostics. Hence, strategies to combine prospective and retrospective sample collections, alternative sampling approaches (e.g. liquid biopsy) and pooling sample resources provided by a network of institutional and pharma biobanks would be beneficial. The impact on clinical studies will be discussed.

Title: BBMRI.it working group on sustainability of biobanks

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Introduction: In a context of complexity, biobanks are called to develop a solid quality system, new methodological processes and adequate tools and technologies. They can guarantee service continuity only in the presence of secure and long-term financing. BBMRI.it has set up a National working group on biobank sustainability.

Material and methods: The working group includes thirty-four researchers, officially delegated by twenty-three National institutions, including Istituto Superiore di Sanità, Research Centers, Universities, Hospitals and companies. Video recorded teleconferences were managed through a web platform, and relevant documents were shared through a cloud computing service, and are now available to the BBMRI.it community.

Results: The activity started in April 2017 and the results were presented to biobankers and stakeholders at the National BBMRI.it day in November 2017. All the participants collaborated on the drafting of the summary document, which includes: 1. Biobank sustainability; 2. Business plan in biobanking: how to implement a tool for sustainability; 3. Institutional role of the biobank / Biological Resource Center, biobanks and clinical investigation; 4. Measures to promote sustainability; 5. Cost recovery; 6. Proposal for a National tariff for biobanking services (preservation and distribution of samples and data; additional services); 7. Relevant documents, links and bibliography.

Conclusion: The three dimensions of sustainability were discussed: operational, social and financial sustainability. The group agreed on the need for biobanks to be officially recognized by their institutions, and proposed concrete measures to support biobanks at the institutional, Regional and National level. Ethical and organizational issues of cost recovery were discussed.

Title: Professionalization of biobanks around the world: Results from an in-depth 2017 Sustainability Survey

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Introduction: Background: A survey on sustainability conducted in 2017 confirms our hypothesis that biobanks that have or are in the process of preparing a business plan are trending toward more professional structures. This abstract presents additional data from this survey.

Material and methods: Methods: In April 2017 an electronic 26 questions survey was distributed worldwide and in multiple languages. Business planning by research biobanks was the main focus. One question was whether the biobank was assessed against annual performance measures and the results against the measures are presented.

Results: Results: 276 biobanks responded (China 65, France 40, USA 34, Spain 27, Germany 24, rest 86). ~2/3 were established in the last 10 years. About half had 65,000 samples or less, with a range from a few to several million samples. Data on status of business planning, tracking of performance parameters, contribution to research and financial recovery will be detailed for the participating biobanks. Trends in tracking these parameters show that the size of the biobank is correlated. Biobanks that measured performance parameters used several of the metrics frequently, except data quality and cost recovery (key activity for financial sustainability).

Conclusion: Conclusion: Four-fifths of biobanks used collection and utilization parameters, "trust metrics," important for social sustainability. The biobanking community is progressively professionalizing through business planning for sustainability. Guidance like the recently published 4th edition ISBER Best Practices and the upcoming ISO biobanking standards will help reinforce this professionalization of global biobanking.

Title: Bimetra biobank: A project-based hospital-integrated biobank, growth through adaptation to customers needs

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Introduction: Bimetra Biobank, a hospital-integrated biobank, working on a project-based manner, has limited governmental funding. Foreseeing these harsher financial times, the Bimetra Biobank performed a survey in the beginning of 2017 regarding their services, which was presented at GBW 2017, in order to validate need, expansion or possible elimination of certain services.

Material and methods: The most appreciated services - high quality biobank storage facility with emergency procedures, quality management system with standard operation procedures, own small processing lab with services and sample- and related data management - were further sustained and partly expanded.

Results: A global yearly performance analysis showed that our adaptation to customers needs led to an increase in our processing services. Additionally, through the promotion of our biobank with our integrated sample- and data management system with ELN, an increase in characterised biobank samples was achieved, with known pre-analytical factors, registered with SPREC.

Conclusion: As Bimetra biobank works in a project-based manner, new collections are frequently started and samples from collections are actively requested for analysis. In global, Bimetra Biobank remains a strong partner for high quality biobanking.



Topic 5B: IT Tools: Solutions and Visions for Biosharing

Title: MPEG-G, the new ISO standard for genomic information representation

Claudio Alberti (1)

(1) CTO and co-founder of GenomSys

The development and rapid progress of high-throughput sequencing (HTS) technologies has dramatically reduced the cost of whole human genome sequencing. Such achievements in the reduction of sequencing costs opened the doors to personalized medicine, where the genomic information of patients can be sequenced and analyzed as frequently as done today for standard blood tests. However, the ever-growing volume of sequencing data is already a serious obstacle to the wider diffusion of genomic medicine in public health. The associated IT costs, related to storing, transmitting and processing the large volumes of data, will soon largely exceed the costs of sequencing. The lack of appropriate representations and efficient compression technologies is widely recognized as a critical element limiting the potential of genomic data usage for scientific and public health purposes. In its 30 years of activity, ISO/IEC JTC 1/SC 29/WG 11 – also known as Moving Picture Experts Group (MPEG) – has developed many generations of successful standards transforming the world of media from analog to digital. The efficient compression and transport of audio and video, together with application formats and APIs, have enabled the interoperability and the integration we all witness in the world of digital media. Working Group 5 of ISO Technical Committee 276 (Biotechnology) has recently joined MPEG to work on the production of MPEG-G, a new open standard to compress, store, transmit and process genome sequencing data. The standard will not only offer higher levels of compression than those available with currently used formats, but it will provide new functionalities such as native support for selective access in the compressed domain, data protection mechanisms, flexible storage and streaming capabilities. This will enable various new applications scenarios, such as seamless streaming of compressed genomic data among processing centers without the latency introduced by compression or processing. Interoperability and integration with existing genomic information processing pipelines is enabled by supporting conversion from/to the legacy FASTQ/SAM/BAM file formats. The MPEG-G standard is currently the largest coordinated and international effort addressing the problems and limitations of current technologies and products towards a truly efficient and economical handling of genomic information. MPEG-G utilizes the latest technology to compress and transport sequencing data for complex use cases that are currently not supported by existing formats. Notable use cases addressed by MPEG-G include: • Selective access to compressed data • Data streaming • Compressed file concatenation • Genomic studies aggregation • Enforcement of privacy rules • Selective encryption of sequencing data and metadata • Annotation and linkage of genomic segments • Interoperability with main existing technologies and legacy formats • Incremental update of sequencing data and metadata.

Title: Integration of Online Analytical Processing system in biobank

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Introduction: Besides the storage of samples, one of the main purposes of biobanks is access provision of specimens data for researchers' projects. For large biobanks this task can be quite complicated, it may take too long to get some export results in reason that it usually done manually by biobank employees.

Material and methods: We used Online Analytical Processing (OLAP) system to create automatic exports of specimens and phenotypes data from Genome Database of Latvian population for internal and external research projects. Solution was provided with data from dedicated data mart in data warehouse (DWH). For visualization we integrated Saiku Analytics (community edition) plugin.

Results: By using business intelligence (BI) system Pentaho Business Analytics and Mondrian we have built virtual cube with 4 fact tables - phenotypes, biochemistry, diagnoses and drugs therapies. Information security was implemented as fine-grained subsystem with two levels - data access policy in DWH and security user groups in BI. After the integration of OLAP system time of processing export request decreased from few days to hours. Security policies and flexible user-friendly interface has allowed researchers to access system directly. Usage of Data Vault standards for DWH made possible to add new data sources and deliver them to end-users in weeks.

Conclusion: Our example shows that integration of OLAP system in large biobank can provide controlled access to its data infrastructure for different levels of biobanks users - from standalone internal researchers to hospitals representatives. Furthermore, automatization of researchers' requests processing workflow can dramatically decrease response time.

Title: Making biobank data and samples findable and accessible

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Introduction: To improve the utilization of existing biobank collections and improve the access to material, BBMRI-NL has further developed the data and sample catalogue and the corresponding request workflow to make biobank samples and data findable and accessible.

Material and methods: BBMRI-NL continued the development of the BBMRI-NL catalogue based on the latest insights from related projects such as BBMRI-ERIC, European Society of Radiology, and the MIABIS-community. To facilitate the request process we evaluated existing workflows and best practices to derive a generic model for sample/data requests.

Results: The BBMRI-NL catalogue has a central role in making data and samples findable and accessible via a federated approach that connects local registries and catalogues with the central catalogue. It has been completely curated and extended with imaging data to provide researchers with accurate data about the biobanks in the Netherlands. The Hyve has developed the request portal that supports the generic request process (Podium). To link the request portal to the catalogue we built upon the software architecture piloted by BBMRI-ERIC in which they linked the BBMRI-ERIC Directory to the Negotiator in their Common Services for IT.

Conclusion: Podium has become the single access point for (linked) data and sample requests from one or multiple biobanks in a harmonized manner. Podium stimulates utilization and access to materials and improves the reliability and accountability of access to samples.

Title: Optimizing Biobank Data Retrieval Combining IT-Based Lexical and Semantic Approaches

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Introduction: Unsatisfying search results in biobank catalogues are often triggered by unprecise query formulations of lexical or semantic nature. Relevant results might not be returned due to typos, search terms or languages that cannot be handled by search engines. We want to outline different query situations and provide appropriate IT-based solutions.

Material and methods: We identified different query formulations that are likely to produce no or incompatible search results. Next, we performed a review of existing IT-based state-of-the-art techniques which can be used to overcome wrong results under these query scenarios. A prototype was implemented to assess the efficiency of the proposed algorithms.



Results: Lexical errors like typos or misspellings are handled by natural language processing (NLP) approaches like “fuzzy” string matching. Morphological variations of keywords were normalized using stemming or lemmatization algorithms, such as Porter or Snowball. To address varying user-specific query formulations including different naming, languages, abbreviations or coding standards, we propose to use comprehensive corpora of entities, related translations and synonyms provided by different domain-specific (bio-)medical standard terminologies. For evaluation purposes, we implemented a prototype combining a NLP-based, open-source search engine and a graph-database including multiple, cross-referenced (bio-)medical ontology concepts which were annotated to biobank-related data within a local search catalogue.

Conclusion: We identified different query formulations where search catalogues leave out or return undesired search results. Misspellings, variations of phrases and languages entered by users must be handled individually by case-specific algorithms. Biobank data retrieval can be optimized if lexical and semantic approaches are combined and applied in the correct sequence.

Title: Rationalised development of a campus-wide cell line dataset for implementation in the Bimetra biobank LIMS system

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Introduction: Bimetra Biobank, a hospital integrated biobank, operates on a project-based manner, storing different types of human body material for research purposes, including several commercially obtained and primary (or in-house) derived cell lines. Additionally, animal-derived cell lines are often stored in the biobank.

Material and methods: As cell lines are very important both in basic research and preclinical screening phases, good authentication, annotation and quality of these cell lines is pivotal in the translational biomedical science field. We set out to develop a rationalised cell line dataset.

Results: Through comparison of different datasets of online cell banks (human, animal and stem cell), we established an extended cell line dataset that was further analysed until a smaller cell line dataset - the survey dataset - was obtained. The survey dataset was spread throughout our campus to all cell line users to rationalise the fields of the dataset and their potential use. Responses for human cell lines, animal cell lines and stem cell lines were captured.

Conclusion: A rationalised cell line dataset was thus obtained. This dataset was subsequently implemented in the biobank LIMS system, using existing standards. By clearly defining the semantics of these fields, an added value for our biobank is created and the data quality of the stored cell lines is increased.



Topic 5C - Harmonisation & Standardisation: quo vadis?

Title: Shifting biobanking management paradigm from Quality by Testing to Quality by Design

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Biobanks strive to achieve a high-degree of quality assurance throughout the biological material preservation-life-cycle, in order to attain consistency both in the biological material preservation and supply. The approach biobanks are currently using for quality management is the classical Quality by Testing which efficiency and contribution to process consistency may be questioned.

Modern, proactive approaches to quality management include process validation based in the acquisition of enhanced knowledge to proper process design, qualification and continual development. It encloses new challenges: data that needs to be captured, managed, disseminated and used to promote the effective and consistent biological material preservation.

Title: The National Cancer Institute's Best Practices for Biospecimen Resources: Current Recommendations

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Introduction: Enhanced biospecimen handling practices are increasingly important for cancer research as molecular analysis become routine in clinical trials and frequently available as part of standard of care. Biospecimens and associated clinical data procured in an established manner can significantly improve cancer biomarker validation/development and validation of clinical diagnostic assays.

Material and methods: The National Cancer Institute's Biorepositories and Biospecimen Research Branch developed the NCI Best Practices for Biospecimen Resources in order to establish a set of guidelines to improve the quality of biospecimen-related research. The guidelines include technical recommendations on biospecimen handling as well as ethical and regulatory best practices.

Results: The latest edition of these Best Practices focused on updating technical and operational best practices with recommendations based on more recent research, guidance and standards for collecting, processing and storing biospecimens; revised informatics best practices; and updated ethical, legal and policy sections describing new developments on return of research results, informed consent for genomics research, data sharing, and community engagement. These Best Practices aim to help patients by improving the reproducibility of cancer research data.

Conclusion: The NCI Best Practices are also foundational to the NIH Precision Medicine Initiative, part of which aims to establish the world's largest research biobank that will support studies that utilize biospecimens from a cohort of one million individuals in the United States.

Title: Biobank quality management in the BBMRI.be network: raise your standar (on behalf of the BBMRI.be quality workgroup)

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Introduction: From as early as 2005, different guidelines and standards covering biobank activities and sample handling methods have been developed to address the low reproducibility of biomarker research. Ten years on, the BBMRI.be quality workgroup wanted to gauge the status of these aspects in the biobanks of the BBMRI.be network.

Material and methods: We launched an online survey to the quality managers in our network to assess their current practices with regards to the standards and guidelines used for their quality management systems (QMS). A second survey was performed to assess the needs concerning support by the quality workgroup.

Results: Eleven out of twelve participating biobanks had implemented a QMS. Overall 11 different norms/guidelines were used as a basis, ranging from ISO9001, ISO15189 over OECD, ISBER and local best practices. SPREC was already implemented in 50% of the biobanks while the other half intends future implementation. Over 70% had never participated in a biobank self-assessment-survey. Only 45% had ever taken part in an ISBER/IBBL proficiency testing scheme. A clear desire for a peer-review audit in the coming year was expressed by 75% of biobanks. Seven biobanks intend to implement ISO20387 when available, six of which in combination with ISO9001 certification.

Conclusion: Overall, the biobanks of the bbmri.be network are actively implementing a quality approach in their practices. Room for improvement was identified for existing "self-testing" opportunities of the QMS. Implementation of ISO20387 can bring further professionalisation of activities. The quality workgroup is setting up an audit programme for the BBMRI.be biobanks.

Title: Development of a standardized healthcare integrated regional infrastructure for biobanking in Stockholm, Sweden

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Introduction: A healthcare integrated infrastructure for biobanking has been developed by the Karolinska University Laboratory in collaboration with Stockholm Medical Biobank and the Karolinska Institutet between 2012 and 2016. This standardized infrastructure called "PreBio" is available for all researchers and healthcare providers within the Stockholm County.

Material and methods: The infrastructure provides full traceability of samples from sample order to retrieval through handling and storage in the laboratory information and management system. Automated sample handling facilities are available at the four largest hospitals in Stockholm and provides reproducible aliquotation. Standard operating procedures regulate the work at all handling sites.

Results: Biobank samples are ordered by electronic referrals from the patient information system. Phlebotomy can be performed at healthcare facilities or at more than 30 accredited phlebotomy centers run by the laboratory. Sample types and volumes, centrifugation conditions, aliquotation volumes and number of aliquots have been harmonized for all users. Time from phlebotomy to -80 °C freezer is standardized into four categories: ≤ 2 hrs., ≤ 10 hrs., ≤ 24 hrs. and >24 hrs. To date approximately 800.000 samples from more than 30 clinical studies have been produced and stored in these facilities.

Conclusion: A standardized infrastructure for biobanking guaranties access to high quality samples for researchers and healthcare providers within the Stockholm County. This infrastructure utilizes the full capacity of the clinical laboratories facilities and can be made available twenty-four seven.



Topic 6A - Biosharing for Scientific Progress: Ethics and Stakeholder Engagement

Title: RD-Connect: working with rare diseases in international Biobanks and genomic databases. Ethical considerations.

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(1) RD-Connect Patient Advisory Council

The European Commission funded project RD-Connect (www.rd-connect.eu) provides a rare disease network of biobanks combining and integrating different types of omics: genomics, transcriptomics, proteomics, metabolomics, lipidomics and detailed phenotype data (phenomics) across research centres and across diseases.

The use of advanced computing techniques removes the need to consolidate the databases in one place. The use of security protocols allows patient information to be secured but does allow the exchange of information to approved users. This allows for more effective data sharing and building of a better understanding of the diseases under study leading to better diagnostics and treatments.

The use of a Patient Advisory Council with a Rare Disease Patient and Ethics Council are a key part of the project helped address the many regulatory, legal and ethics issues associated with the sharing of biological samples, genomics and other omic information as well as management of consent issues.

The RD-Connect platform provides an up and running system allowing new charities, hospitals or other research groups to participate in the sharing of information but as important the many diagnostic and analytical tools developed as part of RD-Connect. This allows cross disease study as well as detailed study within a specific rare disease.

The use of elaborate infrastructures for the handing of patient genetic information and biological samples is one of the key developments in the use of computing to enhance research. Though with this comes many questions and it is critical that the close and effective involvement of patients in this process. At the end of the day it is their data. Most patients would be the first to seek better understanding of their disease. Yet it with a clear contract that there is a level of scientific probity and ethics in the secure management of their samples or data.

Title: Sharing tissue and data for research: do citizens have responsibilities as well?

Evert-Ben van Veen (1)

(1) MedLaw

Many EU projects aim to unlock health data from their "silos". Datasharing has become the norm for scientists. See the FAIR movement. And if tissue is not shared, the data resulting from the analyses of the tissue should. But what about the citizens (patients or volunteers in cohorts), should they share? Ethically a case to be made for this responsibility but the legally the GDPR or at least its interpretation seems to go in a different direction. The presentation will discuss the arguments for this responsibility and the conditions for these arguments to be justified. It will then be discussed how these propositions can be aligned with a balanced interpretation of the GDPR and implementation where member states have leeway to regulate research.

Title: Motivations and Barriers to accessing Human Samples for UK Biomedical Research – how can we better facilitate access?

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Introduction: Human samples are widely used in UK biomedical research and will become increasingly important in precision medicine. However, little is known about how researchers access samples and what motivates them to use these sources. This research characterises the motivations and barriers to accessing and using human samples in the UK.

Material and methods: A survey was circulated to UK based biomedical researchers via relevant communication channels. The inclusion criteria were active biomedical researchers from academia or industry of post PhD level who were based in the UK. Willing participants were then invited to participate in follow up focus groups.

Results: The 229 eligible respondents were primarily from academic institutions (89%), spanning different career stages and research interests. Results showed that sample users utilised a range of different sources, with locality and clinical data being the most cited reasons for source choice. 64% of respondents said they would use a different sample source in the future, with sample range being the most popular motivator cited. Time spent on ethical approval was the biggest barrier to sample access, with 38.8% of respondents citing it as a significant barrier to access. Suggested solutions to barriers elicited from focus groups will also be presented.

Conclusion: These results have important implications for sample resources, by showing that researchers are willing to use new resources that are not local to them. We also show that time spent on ethical approval is seen as a bigger barrier to access than cost or the service from a sample resource.

Title: ELSI Servicedesk: Dealing responsibly with ethical, legal and societal issues in Personalised Medicine & Next Generation Sequencing in the Netherlands

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Introduction: Personalised Medicine promises to bring powerful new ways of improving treatment and health care tailored to the needs of individual patients. At the same time, this leads to a variety of different ethical, legal and societal issues (ELSI).

Material and methods: Within the Personalised Medicine Program of ZonMw (co-funded by Zilveren Kruis and the Dutch Cancer Society), we are developing an online ELSI Servicedesk to offer guidance and answers to ethical, legal and societal issues in personalized medicine & health research. It will be part of the Health-RI shared services portfolio.

Results: The Servicedesk aims to provide rapid and standardized feedback to researchers who face practical legal, ethical or societal challenges. It will triage the questions from researchers, health professionals and patients. Examples of questions are: where can I find a GDPR proof DTA template? How can I facilitate data sharing within my multicenter consortium?

The Servicedesk unlocks existing guidance, information and/or tools. When these do not suffice, a team of experts (e.g. legal specialists, ethicists and clinical geneticists) is available to answer more complex questions (helpdesk). Answers are generalized and made publicly available through the Servicedesk.



Conclusion: The Dutch biobanking community will greatly benefit from the accessible practical information and advice that will be offered by the national ELSI Servicedesk. The Servicedesk is being developed in collaboration with BBMRI-NL and Federa-COREON. The helpdesk will be operational from June 2018 onwards.

Title: Psychosocial determinants of attitudes toward biobanking and biosharing

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Introduction: The development of biobanks depends on cooperation with donors who are the source of human biological material. Effective cooperation depends both on the public relations efforts of biobanks representants, as well as on the willingness of donors to cooperate. The aim of the study is to analyze selected social and psychological variables that may be associated with attitudes towards biobanking, including the willingness of cooperation and scope of consent for international sharing of samples and data.

Material and methods: The data was collected in two rounds of research. First study was conducted on a representative group of over 600 adults in the Polish population and questionnaire included sociological variables such as perception of biobanks, public awareness concerning biobanks, openness to participate in biobanking projects, consent for international sharing and trust. The second study was carried out on the group over 250 participants and questionnaire included psychological tools for assessing attitudes towards biobanking (BANKS scale), personality, nature-nurture beliefs, temporal orientation, altruism and other psychological variables.

Results and conclusion: Specific social and psychological variables significantly correlate with positive attitude towards biobanking. Positive attitude and willingness to cooperate with biobanks is related to level of trust towards scientific institutions and scientists, donation experience, level of education as well as some aspects of present temporal orientation, specific personality traits (e.g. agreeableness) and convictions concerning the determinants of human nature (such as: nurture and interaction of nature and nurture. Biobankers who expect effective cooperation with donors should consider societal and psychological features of participants. It can be important both in personal communication between biobankers and donors as well as in social media communication and social campaigns addressed to potential donors and society.



Topic 6B: Data sharing in biobanking – Discoveries, Novelties, Realities

Title: Biobank networks: pitfalls, opportunities, solutions and conditions for successful international sharing of data and materials

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Modern biomedical research is in need of large, high-quality and well-annotated stocks of samples. We have meanwhile realized that real-world cases are as highly diverse within each disease category and not even large individual biobanks will be able to supply a sufficient number of biomaterials with specific characteristics and rich clinical information – this can only be achieved if biobank cooperate within a network, each contributing their capabilities. To prevent misinterpretation, the quality of both samples and clinical data must be comparable across participating biobanks.

With that in mind, it comes as no surprise that infrastructures supporting international sharing of biomaterial, such as BBMRI-ERIC, are developing into combined bio-data-hubs. The German Biobank Node for BBMRI-ERIC has formed an alliance of eleven biobanks to supply biobank-level data to the European Directory and, in addition, provide rich clinical, sample-level data into local data warehouses, hosted within each biobank. To make all these warehouses to work together with minimal effort (interoperability), biobanks need to provide access data from other source systems (technical interoperability) using a language common understood across the network (semantic interoperability). This process is particularly challenging due the wide variety of biobanks and the resulting level of heterogeneity as well as high standards in data protection and governance (data ownership, patient consent management).

Ultimately, successful sharing of data and materials begins with each biobank. This presentation will give an overview on challenges and opportunities in biobank networking and, ultimately, successful sharing of data and materials.

Title: Imaging and Metabolomics in BBMRI

D. Bos* (1), M.Beekman (2), E.Slagboom (2), D.Vojinovic (1), M.Kavousi (1), C.Van Duijn (1), J.Van der Grond (2), M.Vernooij (1), A.Van der Lugt (1)

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Introduction: Rapid technological developments in the field of medicine facilitated the possibility to unify large amounts of data. Following the huge growth of the medical imaging field, linkage of imaging to –omics-data through structured biobanks has now become possible. We illustrate this using a scientific example linking metabolomics data to imaging-phenotypes.

Material and methods: Using the BBMRI-NL catalogue, we searched for studies that had cross-sectional information on metabolomics and computed tomography-based measurements of calcification in the coronary arteries, aortic arch, carotid artery bifurcation or the intracranial internal carotid artery. We investigated the association of 166 metabolites with calcification volume in the different vessel beds.

Results: By combining the data from two BBMRI-NL biobanks, we included 1,501 samples from the population-based Rotterdam Study and Leiden Longevity Study (mean age 68±5 years, 52% women). We were sufficiently powered to be able to find noticeable differences in the metabolic association pattern for arterial calcification across different vessel beds. Most distinct association was found between larger concentrations of 3-hydroxybutyrate and larger amounts of calcification in the intracranial carotid artery (p-value:1.8×10⁻⁴), whereas this metabolite did not associate with calcification in other vessels.

Conclusion: We demonstrated that linkage of data retrieved from established biobanks holds significant advantages in terms of answering scientifically relevant research projects.

6B - Data sharing in biobanking – Discoveries, Novelties, Realities

S. Soini*(1), A.Kyttälä(2), P.Laiho(2), M.Sauramo(2), K.Silander(2), N.Eklund(3)

Title: THL Biobank - from samples to innovative ways to combine information

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Introduction: National Institute for Health and Welfare, THL, maintains national health and welfare registers since 50s' and governs THL Biobank. THL Biobank has extensive cross-sectional legacy and new prospective population cohorts with rich material, including multi-omics and lifestyle data. THL offers researchers unique possibilities to conduct studies in the post-GWAS era

Material and methods: THL, in line with its data sharing policy, develops harmonized procedure to access both biobank and registry data. National registry data enables selection of study population and/or long follow-up times to be combined with samples and data from THL Biobank. Research participants can be stratified and recalled for new studies.

Results: Availability queries of selected conditions can be performed from national registries i.e. hospital discharge registry and cancer registry. The registry data query results can further be enriched with data from THL Biobank, a nationwide population biobank. The biobanked blood samples have been processed into genomic and metabolomics data which is complemented by substantial information on lifestyle and clinical measurements, collected from the sample donors at baseline. When selecting a suitable population based on the national registry data, the THL Biobank population data and samples can be utilized in new and innovative ways to study the causes of different conditions.

Conclusion: Finding suitable target groups from Finnish national registries and linking them with the refined data stored in THL Biobank offer researchers completely new perspective to understand the 'omics' background of different diseases and its relation to lifestyle. Linking data like this facilitates research far beyond what regular biobanks can offer.

Title: Integration of care and research, FAIR data principles and semantic interoperability through PRISMA, the PSI information model

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Introduction: The Parelsoer Institute (PSI) is an interinstitutional longitudinal biobank collaboration between the eight Dutch University Medical Centres (UMCs). From the onset, PSI has adhered to the principle of registration at the source, the one-off registration of patient data, and making these data both meaningful and reusable for scientific purposes.

Material and methods: Over its 10-year existence, PSI has developed a system-independent information model that far exceeds the typical data models commonly encountered in multi-centre projects, by incorporating extensive metadata for semantic interoperability, detailed clinical models to both facilitate implementation into, and extraction from EHRs, while maintaining flexibility to allow localised interpretation.

Results: The Parelsoer Repository for Information Specification, - Modelling, and - Architecture (PRISMA) not only specifies all information pertaining elements required for generic longitudinal research proposals, but also encompasses underlying meta-information for institute-independent reusability, as well as clinical context. PRISMA offers information architects and IT-specialists a framework for implementation into local EHRs, and the flexibility to be harmonised and expanded with local initiatives. Implementation in several UMCs has both demonstrated the system-independent nature of PRISMA, as well as the possibility to collect research data in a clinical setting without imposing an unnecessary registration burden on health care professionals or participants.



Conclusion: PRISMA has provided UMCs with an opportunity to outfit whatever data capture solution suits their local needs, while still providing the collaborative with meaningful data. Current efforts strive to further the formalised annotation of PRISMA, to make it a computable model that can be interpreted by computers without human intervention.

Title: Real challenges in Healthcare Embedded Biobanking

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Introduction: At present, the greatest potential for improvement in healthcare data and sample collection, which should be promoted with targeted research funding, lies not only in the improvement of equipment and techniques among practitioners and researchers, but rather in the more efficient use of existing bio-medical treatment and research data.

Material and methods: Today's challenges are the fractional collection and storage of data. Data isn't, as in the automotive industry for example, centrally available and is not monitored over the entire life cycle, but is available in numerous structures and semantics at various practitioners, researchers and health facilities or at the patient himself.

Results: The socio-economically important data remain partly unattainable in numerous different systems, partly structured and unstructured. Due to missing decisions or agreements (not missing standards), there is no integration of data to a large and almost complete extent. Many data processing systems in the healthcare sector do not allow targeted transfer/export of data to data integration platforms on the basis of pseudonymized consent. This prevents the central integration of data and the targeted use of data for the improvement of all processes in treatment and research. A simple targeted translation between treatment and research only comes about, if at all, rudimentarily.

Conclusion: Due to this lack of central data integration, it isn't possible to fully document a patient's course and compare it with a patient sub-collective. A possible prediction of the further course is therefore not possible today as "decision support". The change from curative to preventive medicine is thus severely restricted.



Topic 6C Biospecimen and Pre-Analytics Research for Reproducibility: from Sampling to Scientific Discovery

Title: How biobanks can contribute to increase reproducibility of results

Peter Riegman and the SPIDIA4P consortium (1)

(1) Erasmus MC Tissue Bank

An impressive amount of reports and assessments show that irreproducible results cause scientific waste, which severely hinder medical innovation and therewith the promise of precision medicine. Biomaterials and laboratory protocols are recognized as major contributors, putting biobanks directly in the center. High sample and data quality alone do not solve this. Although, high sample quality can raise the sensitivity of discovery, but can later, after publication, hinder the obliged validation during product development. Different collection methods in multi-center cooperation cause center specific variations that can completely overshadow the objected differences. ISO International standards on sample pre-analytics, together with evidence based biobanking enables understanding sample variations and bring collection methods closer together. Study design, analysis and reporting are also seen as causes for irreproducibility. Here biobanks could support study feasibility evaluation of proposed studies before distributing samples. Together with education on study design the here proposed measures could significantly reduce scientific waste.

Title: Overview about the actual research in the field of preanalytics of liquid biospecimen

Fay Betsou (1)

(1) Head of the Luxembourg delegation on ISOTC276

The presentation will highlight the concepts of qualification and quality stratification of biospecimens. It will present a new tool that enables application of relevant assays for qualification and/or quality stratification, with a special emphasis on clinical liquid biological materials.

Title: Influence of Dehydration Protocol on Residual Humidity and Quality of Nucleic Acids in Fixed Tissue

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Introduction: Tissue fixation, dehydration/processing, and paraffin-embedding have been empirically established for preservation of tissue morphology but are less suitable for molecular analyses of nucleic acids, specifically with cross-linking fixative (formalin). Previous observations indicated reduced nucleic acid quality after incomplete dehydration (water carryover in dehydration baths) and prolonged storage at room temperature.

Material and methods: We systematically varied fixation type (crosslinking, non-crosslinking), dehydration (100%, 95%, 90% final ethanol concentration), storage duration (1 year maximum) and conditions (4°C, room temperature in dry or humid air) of mouse liver tissue. Temperature-dependent water release was assayed by water-specific coulometry. RNA quality was evaluated by electrophoresis and fragment-length qRT-PCR.

Results: Van der Waals bound water (released below 100°C) correlated well with storage conditions and type of fixative, but only weakly with tightly bound water (hydrogen bonds, released >= 100°C). Tightly bound water content in the samples correlated strongly with dehydration protocol and type of fixation (crosslinking > non-crosslinking), and was also increased by prolonged storage at high humidity. Nucleic acids quality was compromised by tightly bound water. Storage at room temperature (100% humidity) led to accelerated

degradation compared to dry storage at room temperature or storage at 4°C, with slight advantage of the former.

Conclusion: Tightly bound residual water strongly influenced the quality of nucleic acids from fixed and paraffin-embedded tissue. For optimal quality, tissue should be treated with non-crosslinking fixatives, dehydrated in such a way that the last ethanol bath does not contain more than 1% water, and stored dry at room temperature.

Title: OPTIMARK project: new quality markers for FFPE non-tumoral tissue

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Introduction: Quality of tissue samples is a critical factor on biomedical research results reproducibility. Emerging biospecimen science is focused on clarifying the impact of pre-analytical factors on the sensitivity and specificity on the developing biomarkers.

Material and methods: OPTIMARK project, a multi-center initiative carried out by 13 biobanks within the R&D working group of Spanish National Biobank Network, aims to select and validate the pre-analytical factors relevant on tissue samples. We firstly focused on FFPE tissue samples, in which we analyzed antigenicity in relation with long term storage

Results: A total of 374 retrospective non-tumor tissue samples (colon, brain, lung, breast, stomach and endometrium) preserved from less than 1 year to more than 20 years were tested. Eight different cellular markers with ubiquitous distribution among tissues were selected, according to Human Protein Atlas database, to evaluate quality of antigenicity.

Among the markers tested, Ki-67 protein was identified as a potential quality biomarker for proliferative tissues (colon, stomach), showing a significant correlation among the loss of antigenicity and sample age. Additionally, TTF-1, BCL-2 and beta-catenin showed a slight correlation in lung tissue samples between staining intensity and sample age.

Conclusion: In conclusion, a heterogeneous antigenicity response was found depending on the tested IHC markers, being Ki-67 the marker that showed a significant loss of signal in up to 20 year-old samples.

Title: Q-Map - Quality Markers to assess processing delay of blood and urine samples

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(1) Helmholtz Center Munich, Institute of Epidemiology, Oberschleissheim, Germany, (2) Hannover Medical School, Hannover, Germany, (3) Helmholtz Center Munich, Germany



Introduction: Biobanks store samples and data of participants for long time frames, often without knowing the desired downstream analysis to be conducted later on with these samples.

To judge the quality of the sample after preparation and long-term storage, markers that indicate deviations from the ideal handling procedures are highly desirable.

Material and methods: To establish markers for the pre-centrifugation phase of plasma and urine, EDTA blood and centrifuged and uncentrifuged urine samples collected from 20 healthy volunteers at the Hannover Unified Biobank, the biobank of Hannover Medical School, were exposed to pre-centrifugation/pre-freezing delays between 0 and 24 hours at 4°C or at RT.

Results: Plasma and urine samples were analyzed by a targeted metabolomics approach at the Metabolomics Platform of the Helmholtz Center Munich. For plasma 186 metabolites were measured, for urine 163 metabolites. We find significant concentration changes for different metabolite classes like amino acids and lipids (acylcarnitines, phosphatidylcholines, lysophosphatidylcholines). To establish reliable quality markers for quality prediction, we will use statistical methods like random forest analysis to find single metabolites, metabolite ratios and values of certain metabolite groups. With these we might be able to indicate adverse handling conditions in single samples.

Conclusion: The results of this study will help biobankers to judge the quality of samples and to detect suitable samples for certain downstream analysis. Effects of centrifugation on stability of urine samples will also be analyzed. In future we want to test the validity of these markers also in patient cohorts.



Topic 7A: Hospital-Based Biobanks, Clinical Trials & Precision Medicine

Title: Hospital biobanks - critical building blocks for personalised medicine

Olli Carpen (1)

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Personalised medicine will remain an empty concept, unless sufficiently large scale disease-related sample sets are collected and analysed across the world, and the information fluently translated to the health care system. Especially, longitudinal phenotype information obtained from electronic hospital records, when combined with biological specimens, will provide an essential toolkit for understanding individual variation within disease entities. These databases in combination with artificial intelligence tools and user interphases, allow comparison an individual's disease profile to a reference group, and provide evidence-based predictions of disease outcome and optimal treatments. To achieve this goal, hospital-integrated biobanks, connected with comprehensive electronic health records, provide an elementary platform. I will describe the Finnish nationwide hospital biobank network's "consent all comers approach" for bringing tools for early recognition, successful targeted treatments, and effective preventive strategies to a variety of diseases. Finally, I will provide examples of the possibilities, challenges and achievements along our road towards personalised medicine ecosystem.

Title: Highly automated, strictly quality-controlled Hospital-Based Biobanking as a step towards Highly automated, strictly quality-controlled Hospital-Based Biobanking as a step towards Precision Medicine – the two Pillars to implement: Broad Consent biosample-collection & Support of Clinical Trials

Roland Jahns (1)

(1) University Hospital of Wuerzburg, Interdisciplinary Bank of Biomaterials and Data, Greenland

In the last decade biobanks have been recognized as important resources for the progress in biomedical research, particularly for the recent progress in precision medicine. Hospital-based biobanks may in the future play a key-role for such personalized endeavours by implementing two essential pillars: (a) the collection of patient biomaterials (e.g., blood & tissues) together with the related clinical data under the conditions of an unrestricted broad informed consent and secure strict pre-analytical quality-control, and (b) the full support of (investigator-initiated and/or industry-sponsored) clinical trials.

Hospital-based biobanks must secure (quality-controlled) long-term storage of their hosted biosamples for current and future (non predictable) research issues that often require cross-border exchange of biosamples and related data. On this background the internationally prevailing informed consents are inadequate, requiring an expanded and also cross-border oriented scope, balancing individual donor-rights and research interests. For medical research this fact has also been acknowledged by the current EU-GDPR.

Title: Genomics to medicine - P5 FinHealth

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(1) National Institute for Health and Welfare, Finland, (2) Nightingalehealth, Finland, (3) Helsinki University, Finland

Introduction: Research on omics, including genomics and metabolomics, has identified thousands of genomic regions and several blood metabolites linked to common human diseases and features. This new knowledge combined with large amounts of samples collected to biobanks could provide new tools for predictive, preventive, personalized, and participatory (P4) healthcare.

Material and methods: P5 study, P4 + Population health, studies the value of returning genetic and metabolomic risk information on coronary heart disease, type 2 diabetes and venous thromboembolism. We have invited 6.000

FinHealth 2017-study participants in our study. Prospective clinical significance of the risk scores will be analyzed in 30.000 Finnish individuals.

Results: So far, clinical utilization of omics is fairly small and, above all, unvalidated. We will analyze the genetic and metabolomic profile of the P5 participants and develop and test a protocol for returning them health related risk information. The impact of our intervention will be studied by following up the participants by questionnaires and through national health registers for five years.

Conclusion: We hypothesize that 1) combining genetic and metabolic risk with traditional risk factors adds value to the personal risk assessment of these diseases, 2) such risk information can be provided online, and 3) receiving genetic and metabolomic risk information has an effect on the health of the study participants.

Title: Biobanking within the IBCSG Clinical Trials to Advance Precision Medicine

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Introduction: Biobanking is an integral component to the clinical trials of the International Breast Cancer Study Group (IBCSG) since nearly 40 years. Biological material samples are centrally collected from patients entered to the IBCSG clinical trials, from participating centers around the world.

Material and methods: The BIG 1-98 / IBCSG 18-98 clinical trial and translational research will be showcased. This study was a four-arm randomized double-blind phase III clinical trial that compared 5 years of tamoxifen or letrozole monotherapy, or sequences of 2 years and 3 years treatment with one drug and then the other.

Results: The trial enrolled 8010 patients from 240 centers in 27 countries. Tumor tissue was centrally banked for downstream translational research. These samples were collected to answer specific questions within the clinical trial, to accurately assess and confirm local results, and to have available for future research. Translational research included targeted sequencing for 328 cancer related genes, SNPs (CYP2D6, CYP19A1, ESR1-2), and further translational research answering trial questions of clinical relevance.

Conclusion: The IBCSG has a major aim to support excellence in translational research by facilitating researchers, either members of the IBCSG or collaborators, to access biological material that have been ethically collected and approved for research. These well-annotated BIG 1-98 clinical trial samples have enabled translational research that guide precision medicine.

Title: Naples human mutation gene biobank (nhmgb): a key-resource for patients and families without an established molecular diagnosis

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Introduction: The NHMGB, established in 1991, collects biological samples from patients with rare genetic myopathies/ cardiomyopathies, often with poor prognosis. It is part of Cardiology and Medical Genetics, Campania University, so the follow-up of patients consents to store samples for both basic research and to development of biomarkers and experimental therapies.



Material and methods: The Biobank stores about 11.000 biological samples (blood, DNA, RNA, muscle tissues, proteins, sera and plasma) from patients in various stages of the disease. Each sample is associated to clinical data collected in the same time, allowing the study of the disease evolution and the identification of new disease genes.

Results: NHMGB collection participated in several projects, i.e. the “Undiagnosed diseases project” that included 350 families clinically well characterized analyzed by the next generation sequencing. It also participated to the BBMRI-LPC Whole Exome Sequencing with a list of DNA samples from undiagnosed patients. Furthermore it receives requests of samples with specific diseases and specific mutations from researchers involved in the gene therapy or pharmacological program. It should be noted that having storage blood samples from patients who died before the molecular era in the bank, led to a correct diagnosis, a proper identification of family carriers and access to prenatal diagnosis.

Conclusion: NHMGB is an example of how a hospital-based biobank is useful not only for researchers to deepen their studies, but even more for patients who can have access to clinical trials and take advantage of a precision medicine, and their relatives for the assessment of the carrier status.



Topic 7B: Artificial Intelligence & Big Data in Biobanking

Title: Biophysics-Inspired AI for Digital Pathology

Sergiy Zhuk (1)

(1) IBM Master Inventor, Ireland

The talk will present examples of using a fusion of concepts from AI, machine learning, biophysics and optimisation in digital pathology for medical image analysis and knowledge extraction.

Specifically, the following topics will be covered:

1. Generation of 3D geometric models of tumour cells and probabilistic representations of cell's chromatin density patterns from 2D whole slide images (lung cancer)
2. Dynamic lesion classification (benign/malignant) and shape estimation from NIR images of the ICG perfusion angiography (colorectal cancer)
3. Knowledge discovery and mining of unstructured data, e.g. text mining by means of IBM Watson Knowledge Studio

Title: Connecting Biobanks and Molecular Public Archives: achieving FAIRness of Biosamples with ELIXIR and BioSchemas

Tony Burdett (1)

(1) Technical Coordinator for the Samples, Phenotypes and Ontologies Team

The ELIXIR Bioschemas project aims to improve data interoperability in life sciences, by making data more Findable, Accessible, Interoperable and Reusable ('FAIR'). In this talk, I will present the EMBL-EBI BioSamples Database, and how we are using BioSchemas to facilitate data exchange, data integration and data interoperability with biobanks.

A key challenge for both biobanks and public molecular archives lies in facilitating the exchange of sample metadata. Biobanks have a need to show the scientific impact of the tissue samples they have collected and made available to experimenters, and public archives want to ensure high quality, consistent metadata, in order to maximise the utility of the data they contain to computational biologists, as well as links back to sources where possible.

The BioSamples database provides a single point of access for all sample information stored at EMBL-EBI or NCBI, and as an ELIXIR deposition database is an important part of European life science infrastructure. It is ideally positioned to enable linkage between archives and communities that both contribute data - including biobanks - and consume data, by providing unique accession numbers to samples and corresponding external references to genomic archives.

In this talk, I will present three targeted efforts that expand the BioSamples infrastructure to improve samples metadata interoperability:

1. Improving its quality via new machine learning based tools,
 2. Validating the metadata stored and
 3. Providing lightweight exchange mechanisms between BioSamples and biobanks.
- Harmonisation of sample metadata from multiple sources in order to provide an integrated view over related data can be a significant overhead when those sources have different policies, interfaces and utilise incompatible standards. This often presents as a "long tail" problem in the choice the metadata attributes used. BioSamples contains over 5 million samples (45 million data points samples-attributes). I will present some of the solutions we have developed, including services to annotate sample descriptions to ontologies, and linking datasets from multiple assays back to their original samples. I'll also present a new machine learning approach to clustering sample metadata descriptions and proposals for semi-automated curation applications.

Once harmonized, to be useful the BioSamples metadata needs to encompass the minimum information described by molecular archives checklists, and be exchanged using standard formats for which tools are readily available. I will

describe a new ELIXIR metadata validation project that is using JSON schemas to define metadata standards and reference implementations for validation against these standards.

Finally, I will showcase how the clean validated metadata can be exchanged between BioSamples Database and biobanks. The ELIXIR Bioschemas project extends the Schema.org markup language - currently used by Google and other major search engines - to semantically annotate metadata in the life science domain. I lead the samples specification group and will demonstrate how it has been deployed at EMBL-EBI. Taken together, these three development axes improve the information content of BioSamples metadata, thereby making it more amenable to querying by biobanks to bridge from the original specimen provided to the raw data that was generated from it.

Title: Advanced data-driven phenotyping with electronic health records

M. Koskinen* (1), J.Salmi (1), O.Manninen (1), H.Heikkinen (1), A.Loukola (1), K.Pitkänen (1), O.Carpén (2)

(1) Helsinki Biobank, Helsinki, Finland, (2) University of Helsinki and Helsinki Biobank, Helsinki, Finland

Introduction: Hospital ICT system architecture has commonly been designed with only clinical work in mind; thus, analysis of hospital data demands extensive data extraction, curation and annotation. Advanced tools and pipelines need to be developed to allow efficient use of clinical data, including electronic health records (EHR), in research settings.

Material and methods: Helsinki Biobank is a clinical biobank with access to data-lake infrastructure containing various clinical data repositories within the hospital, allowing efficient utilization and integration of multimodal data for research. As part of a publicly and privately funded digital phenotyping project (DigPhen), we aim at advancing the utilization of EHR data.

Results: In collaborating with clinical experts, patient cohorts covering neurology, oncology and cardiology are formed, allowing the specific clinical characteristics of these indications to be defined and the data relevant to each disease entity obtained. Stratified datasets are analysed using various hypothesis free, AI-based approaches in order to identify treatment responses, assessing disease trajectories, or predicting clinical outcomes. This in turn allows us to develop methodology for advanced phenotyping – above and beyond simplistic disease classifications.

Conclusion: The project increases understanding of disease subtypes, trajectories and comorbidities, as well as predicting outcomes and treatment responses. Furthermore, it advances the capabilities of Helsinki Biobank to collect, curate and integrate multimodal disease-specific data. The project results in advanced machine learning tools applicable in clinical big data and state-of-the-art phenotyping.

Title: Turning Hospital Biobank Data into Real-World Evidence using Artificial Intelligence Tools

S. Kurki* (1), A.Karlsson (2), S.Oksanen (1), M.Tukiainen (1), L.Kallio (1)

(1) Auria Biobank, Turku University Hospital, Finland, (2) Department of Medicine, University of Turku, Finland

Introduction: Hospital biobanks connected to electronic health records are a source of real-world data accumulating outside clinical trials. Such data is valuable for hospitals as well as pharmaceutical industry to study the use and efficacy of treatments. However, artificial intelligence (AI) tools are required to extract information from unstructured patient reports.

Material and methods: Auria Biobank has developed an AI tool that can rapidly extract information, such as smoking status, cancer stage and disease symptoms, from millions of unstructured patient reports. The tool is used to accelerate scientific discovery both in pharmaceutical industry collaboration projects and academic research.

Results: The AI tool has been successfully used in several pharmaceutical industry collaboration projects to (semi)automatically extract information from



patient texts, such as smoking status in lung cancer and ejection fraction in heart failure. The accuracy of the tool tested against an expert clinical investigator has reached 85%-95%. Manual curation in these cases would have been practically impossible. The tool runs inside the hospital firewall system and operates only on patient reports that the researcher has permission to access, maintaining the protection of patient identity. The tool is built using open-source components that allow it to be easily developed further.

Conclusion: Hospital biobanks provide a unique source of real-world evidence for the pharmaceutical industry. However, analyzing unstructured patient reports require novel AI methods. Auria Biobank has developed an AI tool that is used to accelerate scientific discovery. Further work is ongoing to extend the applicability and accuracy of the tool.



Topic 7C: Education Tools and Programs for Biobankers

Title: Next generation biobankers : the French Example

Emmanuelle GORMALLY(1)*, Philip LAWRENCE, Isabelle HARDY

(1) Dean of the Life Sciences Faculty at the Catholic University of Lyon, France

During the last two decades, the increasing demand for high quality biological resources along with associated data has prompted the development of new roles for life and medical scientists. Indeed, (in order) to professionalize this activity and to obtain (and maintain) a reproducible quality of biological resources, professionals working in biological resources platforms have had to develop specific skills, in addition to their skills in life and/or medical sciences.

In France, a Master course was developed to train young scientists to work as the operational coordinators of a biobank/biological resources platform in collaboration with a scientific director and biobank technical staff. A set of life science/medical skills was identified, as well as specific skills including biological resources management, quality and data management, knowledge of laws and regulations specific to biological resources and ethics.

Different pedagogical methods are used to allow the acquisition of specific knowledge but also to acquire professional skills through projects and hands on activities.

Since its launch in 2010, 55 professionals graduated from the Master course in biobank management. 90% currently occupy managerial jobs in research biobanks or in biological resources platforms in industry.

This Master course is one of the elements that will raise awareness for the field of biobanking and biological resources, attract young scientists towards a new career path and participate in assuring the (future/continuing/ever increasing) quality of biological resources and their associated data. To further raise awareness about the field of biobanking, its specific needs and its community, a shared Master program at the European level associated with online courses could be developed along with an introduction to the field in undergraduate, PhD and other postgraduate programs.

Title: German Biobank Alliance: Implementation of an educational program for technical personnel in biobanks

M. Hartung* (1), E.Herpel (2), R.Baber (3), C.Rufenach (1), S.O'Donoghue (4), P.Watson (5), S.Nussbeck (6)

(1) German Biobank Node, Charité – Universitätsmedizin Berlin, Germany, (2)Institute of Pathology, Heidelberg University Hospital, Heidelberg, Germany, (3) LIFE – Leipzig Research Center for Civilization Diseases; University Leipzig, Germany, (4) Office of Biobank Research and Education (OBER); the University of British Columbia, United States, (5) Office of Biobank Research and Education (OBER), the University of British Columbia, United States, (6) UMG Biobank, University Medical Center Göttingen, Germany

Introduction: The quality of biomaterials and samples depends on correct implementation of standardized guidelines and procedures by the responsible technical personnel. However, there are limited education and training opportunities for these personnel. Therefore, the German Biobank Alliance (GBA) will develop an educational program for technical personnel in biobanks.

Material and methods: The educational needs of the technical personnel was assessed through an online survey and a workshop. In collaboration with the Office of Biobank Education and Research (OBER) of the University of British Columbia, GBA will develop online education modules tailored to the needs of the technical personnel in German biobanks.

Results: The survey was answered by 79 participants, of whom 43 were technical personnel. The results specifically highlight a need for education in quality aspects of sample and data handling. The online content of the Canadian education platform will be refined and adapted to address this need. Accompanying practical on-site trainings will refer to findings from ring trials being conducted within the GBA biobanks. Quality biomarkers, identified via a GBA wide study, will also be incorporated into the education program for the

technical personnel. An internal communication platform offers continuous exchange between the technical staff at the different sites.

Conclusion: In conclusion, the education program of GBA will contribute largely to harmonization of the quality management as well as sample and data quality assurance. Through continuous practical and theoretical training of the technical personnel, GBA enables consistent high quality of samples across multiple biobank locations.

Title: A Distance Learning Master Study in Biobanking: A retrospection of the first year

G. Hartl* (1), B.Huppertz (2), K.Sargsyan* (1)

(1) Biobank Graz, Medical University Garz, Graz, Austria, (2) Medical University Graz, Graz, Austria

Introduction: In autumn 2016 the Master course “MSc Biobanking” started at Medical University of Graz. International students from 7 different countries joined a highly topical education in biobanking. To guarantee high quality and to meet the requirements of such an education, an evaluation of each lecture has been performed.

Material and methods: This master distance learning course is composed of 13 modules and each module of 4-5 lectures. Each lecture was followed by an evaluation. On one hand paper pencil questionnaires after every face-to-face week were performed, on the other hand online questionnaires at the end of each module were filled in.

Results: Within the first year 24 lectures were evaluated. The questionnaire comprises 13 questions and two “further comments” questions for suggestions and what the students liked about this course. Overall the results for the first year were very positive. The question how satisfied the participants were with this course was evaluated with an arithmetic mean of 1.30 (1 = applies to a high extent until 5 = does not apply). The current program met the expected content to a high level with a mean of 1.37. During the face-to-face courses the students liked the high number of group works most.

Conclusion: It is known that the extensive growth of biobanks requires training of highly qualified personnel in the field of biobanking. Based on the results of the evaluation of the first year “MSc Biobanking” we conclude that this educational program meets the requirements of the developmental needs and challenges in biobanking.



Topic 8B: Museum biobanking & preservation

Title: Zoos, a museum and its biobank

Andrew C. Kitchener (1)

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At National Museums Scotland we have been collecting vertebrate tissue samples for extraction of DNA and screening for pathogens by external researchers for more than 25 years, although some samples date back to the 1950s and 1960s. More than 99% of samples are linked to traditional museum specimens in our collections, including skins, skeletons and spirit specimens, so that taxon identities can be checked independently. There are two main sources of these samples. Firstly, native species that are often collected as part of national surveys, providing large numbers of samples per species spread over wide geographical areas. The second major source is zoos. Specimens were collected initially for use in taxidermy in exhibitions, but have provided important research material for a wide range of studies, including anatomy, functional morphology, archaeology, palaeontology, ontogeny, taxonomy, sexual dimorphism, etc. However, our deep frozen collection of muscle tissue samples derived from these animals is in great demand by external researchers. Currently we hold several thousand samples of both zoo and wild vertebrate specimens. These are used mainly in phylogenetic, phylogeographical, species hybridisation, population genetics, taxonomic and wildlife-crime research, thus providing an important source of high-quality DNA for such studies. Examples of these studies and the importance of zoo-derived samples held within our biobank collection will be given. The advent of the BBSRC-funded CryoArks biobanking project will provide much needed infrastructure and resources to allow our collection to be integrated into a UK-wide biobanking network thereby facilitating future access to and enhancing the research potential of the collection.

Title: From Bottled Babies to Biobanks: A history of making knowledge from body parts

Karin Tybjerg

This paper considers practices of collecting parts of bodies for generating medical knowledge from the pathological collections from early 19th century to the biobanks of 21st century. It shows how investigative practices in the 19th century collections such as the Saxtorpean collection of babies and fetuses are similar to those of modern biobanks such as the collection of PKU samples in the Danish National Biobank. There is thus a continuum of practices between historic and modern collections and biomedicine may be viewed as not just a lab science but also a collection science. The paper also touches on the relations between historic and scientific collections – between preservation and investment – and lastly touches on the public presentation of human remains and biobanks and how the historic collections may serve to make the biobanks more comprehensible while the biobanks make the historic collections of human specimens more ethically acceptable.

Title: SCAN: Schistosomiasis Collections at the Natural History Museum

A. Emery* (1), F.Allan (1), M.Rabone (1), D.Rollinson (1)

(1) Natural History Museum, London, United Kingdom

Introduction: Schistosomiasis is a neglected tropical disease affecting 200 million people, mostly in sub-Saharan Africa. The disease is caused by schistosomes – flatworm parasites that invade the blood vessels of their host. SCAN supports fieldwork and acts as a repository providing access to schistosomes and their snail vectors for research.

Material and methods: SCAN provides access to a legacy of cryopreserved adult schistosome specimens collected over decades, while supporting a number of research projects sampling the accessible larval stages of the parasite and the snails which transmit these parasites. Specimens are returned to our molecular collections facility for cataloguing, storage and distribution

Results: Active since 2010, SCAN continues to grow. In 2017, we accessioned 58748 schistosomes (42558 miracidia, 16190 cercariae) and 17250 snail specimens through direct fieldwork and by acting as a repository for several independent projects (e.g. projects within SCORE, <https://score.uga.edu/>). Data are served through the museum data portal (<http://data.nhm.ac.uk/>), GBIF (<https://www.gbif.org/>) and the SCAN website (<http://scan.myspecies.info/>).

The specimens in our collection are being re-purposed for a number of genetic applications beyond those for which they were originally collected. Examples include evolutionary/speciation studies, comparative genomics, exome capture resequencing, marker development for diagnostics, controls for independent collections, phylogeographic studies.

Conclusion: With a combination of expertise and resources, SCAN acts as a link between field teams and researchers that individually may lack the means to deliver the research outcomes that can be achieved through partnership.

Title: A novel method of restoration & preservation of mounted anatomical pathology specimens

M. Kulkarni* (1), P.Vaideeshwar (2), P.Rambade (1), S.Dalvi (1), D.Kadam (1), P.Chavan (1), S.Gursale (1)

(1) Tata Memorial Hospital, Mumbai, India

Introduction: Tata Memorial Hospital, Mumbai, is premier specialist cancer centre in the Indian subcontinent, has sizable collection of mounted, old precious specimens, collected over a long period of time. While refurbishing, few of these specimens often fell from sewn strings. We developed novel method of remounting such specimens on Perspex sheets with Cyanoacrylate adhesive.

Material and methods: Specimens gently washed under tap water, drained & soaked in ethanol. Few drops of Cyanoacrylate adhesive placed on Perspex sheet. Drained from ethanol, specimen placed onto sheet, gently pressed & allowed to dry for 15 minutes. Specimen thus stuck with sheet placed in the glass jar, filled with mounting fluid & sealed.

Results: We tried this technique initially for our 10 old & precious specimens, including brain. Specimen could be viewed well from all sides. Specimens remained well adhered to the Perplex sheet for several months. Perplex sheet has a better light transmission & visibility than glass. Ethanol helped in the removal of excess water & restoration of the colour of the specimen. As observed, few drops of Cyanoacrylate adhesive did not interfere in the natural shape, size & colour of the specimen. Specimens could be easily oriented & mounted. Gross pathologic characteristics could be well illustrated in the specimen.

Conclusion: Cyanoacrylate adhesive technique was extremely useful in permanent preservation of old delicate anatomical pathology specimens which could not be sewed/tied to glass rods again & had fell out of position in mounting jars. It is easy, cost effective, serves as an educational tool & can be applied to all specimens.



Topic 9A: Academic – industrial partnerships to accelerate scientific discovery

Title: The FinnGen project: an example of a public-private partnership

Scientific Director: Professor Aarno Palotie
Academic coordinator: University of Helsinki

www.finnngen.fi

The FinnGen research project will construct a unique resource of 500 000 Finns that enables ambitious study designs to improve our understanding of the genetic background of diseases and subsequently for implementation of genome medicine in clinical practice and drug development. The FinnGen research project aims on one hand to provide useful and exciting new data but also construct a world-class resource that can be applied for future studies that combine genetic and health data in a way that is currently not possible on this scale.

The FinnGen study collection will consist of 200 000 already existing legacy samples, mainly from the National Institute for Health and Welfare's (THL) Biobank and 300 000 prospective samples collected by all of the six Finnish hospital biobanks and the Blood Service's biobank. Thus the collection will be a mix of population based cohorts (e.g. Finrisk, Health 2000) and disease specific samples. The prospective samples collected by hospital biobank will primarily represent diseases that are treated in tertiary referral clinics.

The study aims to produce close to complete genome variant data from all the 500 000 participants using GWAS genotyping and imputation that is based on a population specific whole genome sequencing (WGS) imputation backbone. This is possible due to the strong bottleneck effect, which the Finnish population has experienced. Using the population specific imputation strategy, variants can be reliably imputed down to very low frequency. This provides currently the most cost-efficient strategy for low frequency association studies. WGS would be more than 50 times more expensive than the GWAS strategy and would be unlikely to provide a corresponding leap in return of investment.

The study will utilize the extensive longitudinal health register data available on all Finns. To monitor the use of national health care services, Finland has established national registers that record every hospital and outpatient visit, every prescription drug purchase, all cancer cases, causes of death, usage of social services and many others. These registers cover the entire lifespan from birth to death and have been digitalized for decades, some from the 1960s. This data provides unique opportunities to study disease associations (GWAS and PheWAS), disease trajectories and comorbidities.

The FinnGen project is an academic-pharma partnership that involves nine Finnish biobanks, all Finnish University Hospitals and their respective Universities, the Institute of Health and Welfare (THL), the Finnish Red Cross Blood Service and seven large pharmaceutical companies (Abbvie, AstraZeneca, Biogen, Celgene, Genentech, Merck/MSD and Pfizer) to develop the potential of these resources to serve medicine.

Title: Standardizing Liquid Biopsy at the European Level - IMI's CANCER-ID

Thomas Schlange (1)

(1) Bayer AG, Germany

Within the Innovative Medicines Initiative (IMI) projects of general relevance to the European Health Care Sector are being addressed in public-private partnerships. The CANCER-ID program which was established in 2015 and has currently 40 partners from academic and clinical research groups as well as from diagnostic and pharmaceutical corporations. The consortium aims at standardization of Liquid Biopsy technologies in the fields of Circulating Tumor Cells (CTCs), circulating free tumor DNA (ctDNA) and microRNAs (miRNA) in Non-Small Cell Lung Cancer (NSCLC) and metastatic breast cancer. By developing best practice documents including biospecimen storage and shipment, enrichment and analysis and collection of technology benchmarking

data, CANCER-ID tries to facilitate regulatory approval and the use of Liquid Biopsies in multicentered clinical studies.

9A - Academic – industrial partnerships to accelerate scientific discovery

Title: The UK working in partnership to support medicines discovery

P. Quinlan* (1), J.Peach (2), J.Sims (3), E.Lawrence (3), M.Styles (1), B.Thomson(1), B.Fuller (3), A.Gander (3), J.Garibaldi (1), R.Emes (1), B.Davidson (1)

(1) University of Nottingham, Nottingham, United Kingdom, (2) Medicines Discovery Catapult, United Kingdom, (3) University College London, London, United Kingdom

Introduction: There are a large number of biobanks in the UK, yet, small and medium-sized enterprises (SMEs) will cite access to samples as a challenge. The UK organisations are coming together to help ensure that UK samples can be accessed by SMEs in order to drive future medicines discovery.

Material and methods: The UKs national node (UKCRC Tissue Directory and Coordination Centre) and the Medicines Discovery Catapult (MDC) have formed a collaboration. This was initiated after a survey undertaken by the MDC identified challenges experienced by SMEs when accessing samples from UK sources.

Results: The survey undertaken by the MDC found 80% of SMEs questioned had an issue accessing samples from UK sources and as a result 69% accessed samples from outside the UK. It is clearly in the interest of all parties that UK based SMEs can source samples from the UK rather than seeking samples from abroad. The collaboration will see the promotion of the tissue directory (<https://directory.biobankinguk.org>) and MDC developing a help-desk, via Tissue Solutions Ltd, so that SMEs can experience an easier process for securing samples from the UK.

Conclusion: The UK has developed an integrated avenue for SMEs to approach biobanks via a collaboration with the national node to enhance discovery of biobanks and via Tissue Solutions Ltd to aid the procurement of samples found. The anticipated result is a significant increase in SMEs accessing samples from UK resources.

Title: EPTRI–European Paediatric Translational Research Infrastructure. Strengthening the development and use of paediatric biomarkers to offer better medicines for children

Angelica Intini* (1), Donato Bonifazi (1), Florence Bietrix (2), Alberto Borobia (3), Antonio Carcas (3), M. Gavish (4), E. Jacqz-Aigrain (5), M. Lupo (6), L. Mangiarini (7), M. Migdal (8), G. Migliaccio (9), M. Phylactides (10), G. Pontrelli(11), F. Rocchi (11), L. Ruggieri (12), M. Vedunova (13), M. Kleanthous(14), M. Lavitrano (15)

Consorzio per Valutazioni Biologiche e Farmacologiche, (2) European Infrastructure for Translational Medicine, (3) Servicio Madrilenio de Salud, (4) Technion - Israel Institute of Technology, (5) Assistance Publique - Hopitaux de Paris, France, (6) TEDDY Network of Excellence for Paediatric Clinical Research, (7) Fondazione Penta-For The Treatment and Care of Children with HIV-Onlus, (8) Instytut Pomnik Centrum Zdrowia Dziecka, (9) Consorzio per Valutazioni Biologiche e Farmacologiche, (10) The Cyprus Foundation for Muscular Dystrophy Research, (11) Ospedale Pediatrico Bambino Gesù, (12) Fondazione per la Ricerca Farmacologica Gianni Benzi Onlus, (13) Nizhegorodskiy Gosudarstvennyy Universitet IM N.I. Lobachevskogo, (14) The Cyprus Foundation for Muscular Dystrophy Research, (15) Biobanks and Biomolecular Resources Research Infrastructure Consortium

Introduction: Despite the recognised importance of biomarkers in medical practice and drug development, there is a relative dearth of validated paediatric biomarkers. Frequently, biomarkers found to be efficacious in adults are extrapolated to children, although the pathogenesis of paediatric diseases is often different and ontogeny influences drug response and disease development.



Material and methods: Biomarkers use in paediatric is one of the field addressed in the European-funded ID-EPTRI project. EPTRI aims to act as a “paediatric common service”, as synergistic complement to the existing RIs, aimed to promote new medicines designed for children.

Results: Within EPTRI a Platform Supporting Biomarkers use in Paediatric Medicines Development will be set-up in collaboration with existing RIs, in particular BBMRI-ERIC. The platform aims to identify the critical issues that hamper the advancement of paediatric biomarkers and define tools and facilities to be provided by EPTRI to create optimal conditions for the development and use of paediatric biomarkers. The platform will enhance the use of -omics science, the biobanking activity and the process of biomarkers identification, characterization and validation in paediatrics. A feasibility study on biomarker development will be proposed to test the platform.

Conclusion: Biomarkers research contribute to many phases of drug development, in paediatric it has to face specific and additional challenges compared to adult research. Fostering the recognition and validation of biomarkers relevant in paediatric diseases, EPTRI will increase the offer of efficacious and safe medicines for children.

Title: Biobank Sweden – A New Organization of Biobank Infrastructure in Sweden

Lena Thunell (1)

(1) Linköping Biobank Facility / Biobank Sweden, Sweden

Introduction: The Swedish Biobank landscape has evolved during the past decades and we are now organized into a new national biobank infrastructure, Biobank Sweden. This is a joint initiative between Swedish Healthcare & Universities with medical faculty, and industrial partners. Biobank Sweden is a continuation of BBMRI.se and member of BBMRI-ERIC.

Material and methods: The general goals are; i) To build a coherent and long-term sustainable national biobank infrastructure for healthcare, academia and industry with the optimal conditions for both national and international cooperation; ii) A nationally accessible, cost effective biobank network which secures access to high quality samples; iii) Benefit for patients.

Results: The vision is to create a national single entry-point for sample access with local Service Centers, established in collaboration between participating universities and healthcare providers. At each location a dedicated Service Coordinator will act as contact point for sample access and participate in harmonization actions across Service Centres. In order to capitalize on the diversity of biobank expertise among different universities, National Service Centers will be established at each Node. These will cover areas such as biobank ethics and law, healthcare integrated biobanking, sample quality, cohort enhancement and exploitation, cytology biobanking, frozen tissue biobanking, IT-infrastructure and sample freezer logistics planning.

Conclusion: The vision of Biobank Sweden is to build a long-term sustainable organization to be able to offer co-ordinated access to high-quality biological samples and data for medical research and clinical trials.



Topic 9C: Pitch Your Scientific Idea -Innovative Techniques and Methods in Biobanking -(no product pres)

Title: Ensuring Quality from Above: Drone4Care, an innovative unmanned vehicle-powered solution to improve supply-chain for health matters and patient care quality

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Introduction: 20% of temperature-sensitive products are damaged during transport and 25% of vaccines are degraded once they reach their destination due to incorrect shipping or broken cold chain. Managing the cold chain is mandatory to minimize pre-analytical variability and maintain sample integrity and quality during inventory, transfer and transport to end-users.

Material and methods: While, globally, institutions are looking at upgrading the samples quality by standardization in improved pre-analytical workflows, locally people forget to look at other steps along the biobank's value chain: transportation! Today, everything moves: people, goods and data; so why not human biospecimen samples and healthcare products?

Results: First, we focus on a next-generation of connected high-performance passive temperature control packaging systems to minimize temperature excursions. Second, a drone-based solution is presented due to their carbon footprint reduction, timeliness and delivery costs. Using drones in the healthcare sector raises several challenges (autonomous flight, collision detection, obstacle avoidance, payload, autonomy, flying distance...). The implications of tackling all above issues are at the core of Drone4Care's task. Our operational activities would be effective for smart cities and remote locations.

Conclusion: Drones are meant to close that gap. We don't use drone because it's cool. We are using them because they do a job better than the alternatives. Drone4Care is reinventing the operational efficiency in transportation and distribution of health matters to improve patient care quality and scientific research.

Title: Synthesis, characterization, and biological evaluation of multimodal dendrimer-based probe for targeted imaging of receptor for advanced glycation end-products

M. Wozniak* (1), C.Konopka (2), J.Hedhli (2), A.Ploska (1), A.Siekierzycka (3), I.Dobrucki (4), L.Kalinowski (3), L.Dobrucki (5)

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Introduction: The endothelial expression of the receptor for advanced glycation end-products (RAGE) is enhanced in patients with a range of peripheral occlusive vascular diseases. We evaluate a novel ⁶⁴Cu-labeled RAGE-targeted probe as PET-optical imaging agent to non-invasively assess spatial and temporal changes of RAGE expression in murine model of hindlimb ischemia.

Material and methods: RAGE-probe was synthesized by functionalization of PAMAM dendrimer with Nε-carboxy-methyl-lysine, conjugated with both rhodamine and Copper-64 for optical and PET-imaging. Binding were examined at the cellular level using HUVEC cells incubated in glucose using fluorescence and gamma well-counting methods. Biodistribution and targeting properties were evaluated in murine model using microPET-CT-imaging.

Results: Biodistribution studies demonstrated similar retention of both targeted tracer and non-targeted control. Image analysis correlated well with the gamma well-counting (GWC) results (r2=0.63). Both GWC and microPET imaging demonstrated a focal uptake of the tracer in ischemic hindlimb and permitted quantitative analysis. ⁶⁴Cu-labeled RAGE targeted probe's uptake was ~343% higher than that for the non-targeted control at 1 week post hindlimb ischemia (HLI), which returned to basal levels at 2 wks post HLI (p< 0.05).

Conclusion: In the setting of hindlimb ischemia, augmented RAGE expression/activation can be monitored non-invasively with targeted RAGE imaging. This targeted multimodal imaging strategy may allow for both serial in vivo studies of RAGE/AGE pathophysiology and postmortem quantitative microscopic assessment of RAGE/AGE axis in biobank stored specimens.

Title: Tissue Microarray and Quantitative Digital pathology for Research and Diagnostic Application

P. De Blasio* (1), I. Biunno (2)

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Introduction: Tissue microarrays (TMAs) and Quantitative Digital Pathology (QDP), represent powerful tools for the identification and validation of molecular targets with clinical significance. Cell Line Macro Arrays (CLMA's) are also used in Stem Cell Research as low cost "high-throughput platform" to screen for bona fide iPSC clones and study 3D Organoids/spheroides

Material and methods: The Cell Line Microarrays were constructed with the Galileo TMA instrument. The iPSC cell lines were prepared by Dr. Ida Biunno (co-author) of IRGB-CNR.

and analyzed with the Visiopharm OncoOptics Quantitative Digital Imaging Software.

Results: Individual clones obtained from human fibroblast reprogrammed into pluripotent stable stem lines were screened, using CLMA/ TMA technology in addition to Quantitative Digital Imaging SW, in order to identify the clones with the highest probability of differentiating toward endoderm, mesoderm and ectoderm cell fate.

Conclusion: We have successfully used the CLMA/TMA platform in addition to Quantitative Imaging SW to select the iPSC clones which were then differentiated into pancreatic pseudo-islets and cardiomyocytes.

Title: Alternative methods for the creation of strategic collections of high interest in biobanking

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Introduction: Collections of some pathologies such as pediatric or neurological diseases (Alzheimer) are difficult to create, due to the characteristics of these patients. After clinical laboratory tests have been performed, it can be possible to obtain the blood clot as a source of DNA, which can be used in biomedical research.

Material and methods: In this study blood was extracted by venopunction from 5 healthy volunteers and DNA from the blood clot, was extracted using the AllPrep DNA/RNA Mini Kit (Qiagen, Hilden (Germany)) fresh and stored for a month. In parallel, blood clot samples from different pathologies were recruited to create strategic collections.



Results: The quality of DNA extracted from blood clot and whole blood was compared by spectrophotometry, fluorimetry and agarose electrophoresis. Functionality was assessed by Multiple Long PCR. Results obtained showed difference in the quality of the DNA obtained with ratios A260/280 somewhat less than 1.80 for blood cot. However, the efficiency and the functionality of the DNA were similar in both cases. Although, for blood clot better results were obtained from tubes without gel.

At the same time samples from the clinical laboratory were recruited, and four strategic collections were created: pediatric control, autism pediatric, Alzheimer patients and diabetic patients.

Conclusion: The data obtained showed that the blood clots are an alternative when it is very difficult to obtain a sample for research, so that the clinical laboratory samples could be used. This has allowed the creation of 4 strategic collections in biobanking that are of great interest for biomedical research.