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The European research infrastructure for biobanking and biomolecular resources in health and life sciences

ESBB
European, Middle Eastern & African Society for Biopreservation and Biobanking

EUROPE BIOBANK WEEK CONGRESS 14-17 MAY 2024

ORAL PRESENTATIONS

Abstracts

Produced by the Europe Biobank Week Programme Committee and BBMRI-ERIC's Department of Outreach, Education and Communications

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Oral Presentation Abstracts

KN1. Keynote One

O1: UK Biobank: scale, depth, duration ... but, most importantly, accessibility

Sir Rory Collins (1)
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Abstract

My talk will provide information about UK Biobank, highlight recent enhancements, and introduce our cloud-based Research Analysis Platform that is facilitating research access.

UK Biobank is a prospective cohort study of 500,000 men and women aged 40-69 years when recruited in 2006-10 that integrates large-scale genomic data (including sequencing) and deep phenotyping data (including lifestyle factors, physical measures and multi-modal imaging) with long-term longitudinal health records. The recent addition of large-scale metabolomic and proteomic data is creating an even more powerful resource, enabling better understanding of disease biology and discovery of novel drug targets. With its unique combination of scale, depth, duration and accessibility, UK Biobank is enabling tens of thousands of researchers worldwide to perform innovative discovery science that has already yielded more than 10,000 peer-reviewed publications (over 3,000 in 2023 alone).

Ready access to all of these data on the 500,000 UK Biobank participants allows researchers worldwide to advance discovery science and improve human health. To accommodate the rapid growth of the resource and enable even more researchers across the world to access these data without limitations of transferring, collating, storing, and accessing data at this scale, UK Biobank launched a cloud-based Research Analysis Platform. The availability of research credits for early-career researchers and those from low- and middle-income countries further democratises access to this unique research resource.

KN2. Keynote Two

O2: Biobanking with Underrepresented Populations: The Critical Role of Participant Engagement

Dr. Gillian Bartlett (1)
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EC. Ethics Café

O3: Ethics Café

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This interactive session aims at facilitating an exchange of thoughts and knowledge about pressing ethical issues. The ignition for this year's topic on ethics committees and biobanks is given by Prof. Josef Haas. We count on your insights and experiences to bring different viewpoints forward and engage in the exchange that this format provides. Session chairs: Dr. Michaela Th. Mayrhofer and Dr. Pieter Moons.

TRACK 1: One Health & Precision Medicine

T1. Role of biobanks in the future of healthcare research

O4: "DISRUPTOR in Wroclaw Medical University: Digital Medicine: an Innovative approach for SuppoRt and UPgrade of the diagnosis and Therapy based On Research

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Introduction

Medical Research Agency (MRA), the institution representing Poland in ECRIN is responsible for the research development in medical and health field together with implementation of the Government Plan for Development of Biomedical Sector 2022-2031 perspective.

Materials

MRA implemented first national public programs for non-commercial clinical trials with obligatory biobanking. In 2023 an initiative of Digital Medicine Centres Network formation–Regional Digital Medicine Centres(RDMC) was announced with biobanking as a strategic goal.18 RDMC were chosen, where DISRUPTOR project took 1st place.

Two prominent clinical areas: rare diseases, cardiovascular diseases concentrated on heart failure, characterized by significant diversity in terms of the amount, nature and type of generated data including omics are in the project scope, supported by data from the pathomorphology, radiology and biobanking. Also cardiological regional networks, national rare disease networks together with medical equipment data will fill the DISRUPTOR-RDMC for further algorithms usage where links for diagnostic process improvement, selection of appropriate therapy, important for improving the patients' quality of life will be proposed.

Results

DISRUPTOR constitutes the first step towards the broad implementation of digital medicine within Biobank, Hospital and University. The aim is to standardize the acquisition and processing of health data for scientific and analytical purposes, ensure their highest quality and enable secure exchange of structured data, finally implementing innovative digital solutions."

Conclusions

RDMCs associated in DMCs Network will be prepared to serve real-time data analysis, support clinical trials and hospital care in the area of digital solutions, and retrospective analysis as well. The biobanking will be performer according to BBMRI.pl "Quality Standards for Polish Biobanks"."

Keywords

ECRIN, HealthCare Research, Digital Medicine, Biobanks, Quality Standards for Polish Biobanks, Clinical Trials, Digital Medicine Centres Network, Algorithms, AI

O5: Detection of hereditary cancer variants in early cancer cohort of Latvian National Biobank

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Introduction

Rapid cost decrease in whole genome sequencing (WGS) prices has enabled the generation of large amounts of WGS data. One of the clinically significant applications of these data is the identification of hereditary cancer cases as NCCN guidelines can provide strategies to prevent, delay and effectively treat oncology conditions. We aimed to utilize already available resources of the Latvian National Biobank to preselect a cohort of cancer patients with a higher probability of carrying germline cancer predisposition variants, perform WGS, identify patients with pathogenic variants, carry out genetic counselling and use NCCN guidelines to improve patients' health.

Materials and Methods

Included participants were aged 18 to 50 with a first cancer diagnosis. Prioritizing those with a self-reported family history of cancer. We utilized WGS to detect pathogenic variants. In total, WGS data for 158 patients were obtained using the MGIEasy PCR-Free DNA Library Prep Set on the DNBSEQ-T10×4RS sequencing platform.

Results

We detected 228 variants across all samples and after review by professional geneticists, 33

patients were invited for genetic counseller visit. So far 12 patients attended the visit and an independent genetic test confirmation was carried out in diagnostic laboratory for all. We have confirmed 8 inherited cancer families and furthered their health care to NCCN guidelines.

Conclusion

We have demonstrated feasibility of approach to report data back to participant and discovered 8 families with inherited cancer variant that have been appointed for further high-risk screening. Further we plan to investigate new potentially pathogenic variants.

Keywords

biobank, whole genome sequencing, hereditary cancer

O51: Towards microbiome preservation: Importance, Knowledge gaps & first insights from soil microbiome preservation experiment

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Introduction

Microbiomes are complex communities of microorganisms that, together with their “theatre of activity”, are characteristic of a particular habitat. Microbiomes provide various crucial ecosystem services and are thus essential to the well-being of plants, animals and the environment. To enable science to fully harness the functions of microbiomes to address global challenges, methods and technologies are needed to capture, create or perturb microbiomes and to allow their long-term stable maintenance. MICROBE is an EU project dedicated to tackling these challenges.

Material & methods

In a comprehensive study, two well-defined and previously characterised soil samples were used to assess a range of different preservation methods (i.e., different storage temperatures, with or without cryoprotectants, with or without a controlled cooling rate). The samples were analysed at different time points to assess the viability and composition of the soil microbiome using both cultivation-based and cultivation-independent methods.

Results/findings

Preliminary results indicated that tested preservation methods could maintain the overall culturability of bacteria and fungi, as relatively constant colony-forming units were detected under tested experimental conditions across different treatments and time points. The assessment of the effects on the community composition is ongoing.

Discussion/conclusion

The best practices identified in this study will be shared with the scientific community for further validation. Furthermore, insights into the effect of the different preservation methods on different microbiome members will be used to design the most promising strategies for other matrices.

Keywords

Microorganisms, soil microbiome, preservation methods, cryopreservation

T5. Success stories – Biobanks fostering medical progress

O19: Transferring Risk Mutation Data from Biobank to Healthcare

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Description

Clinical healthcare data is commonly used in biobank research projects along with the data analysed from biobank samples. We have piloted the opposite direction by transferring biobank sample data to healthcare, as supported by the Finnish Biobank Act.

Biobanks have become a significant source of genetic information, when genomic data produced in research projects is returning to

biobanks. We have developed and piloted the implementation of a process in which biobank sample donors with a high genetic breast cancer risk (*BRCA1*, *BRCA2*, *PALB2* mutation carriers) are recalled for result disclosure and referred to healthcare. This opportunistic screening has been enabled by the massive (300 000 individuals) return of the FinnGen research project genotype data to Finnish hospital biobanks.

The results show that approximately one sample donor in 200 falls within the high-risk profile. It was observed that 75% of the mutation carriers had not been previously identified in healthcare. These sample donors were referred to diagnostic testing, genetic counselling and enhanced follow-up or risk reducing interventions. We have also collected and studied the feedback from the sample doors participating in this recall. The feedback is positive supporting the adoption of these type of novel preventive care paths in healthcare. In addition, we have studied the cost-effectiveness of the piloted care path. The results show that returning genomic data to sample donors resulted in noteworthy health gains and was feasible and cost-effective.

Keywords

risk mutation, biobank data return, preventive care path, screening

O20: The Story behind BBMRI-ERIC Federated Platform

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Description

The BBMRI-ERIC Federated Platform, launched in 2023, facilitates data and biospecimens sharing among researchers. It integrates diverse data from divers sources, a challenge since its inception.

The concept emerged within the BBMRI community in 2009 [1]. Efforts to create a service enabling real-time and more granular search of biospecimens and data intensified in 2015. However, the co-development of such service by BBMRI was discontinued in 2019 [2],[3]. Tender by BBMRI-ERIC headquarters in 2021, resulted in inclusion of open-source and commercial solutions in the Platform.

The Platform, grown from in-kind contributions, requires intensive coordination and strong community engagement. Key lessons learned from the pilot phase include clear stakeholder relationships, enhanced biobank motivation, well-organised communication, community feedback, documentation updates. The onboarding of new biobanks is supported by the EvolveBBMRI project.

The sustainability of maintaining multiple solutions within one platform and the feasibility of keeping the initiative as in-kind contribution are areas for further discussion. The platform's development demonstrates potential of collaborative efforts in datafication of the biobanks and ability to integrate data from multiple sources.

Acknowledgements

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[1] https://doi.org/10.1007/978-3-642-03722-1_7
[2] https://pubmed.ncbi.nlm.nih.gov/28883174/
[3] https://pubmed.ncbi.nlm.nih.gov/32663150/

Keywords

Keywords distributed infrastructure, sensitive data, federated search, biobanks identification, biospecimens and data

O21: Biobanks as facilitators of medical research: Success stories

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Introduction

Biobanks play a vital role in advancing biomedical research by granting researchers access to valuable biological samples for a diverse array of research studies. BBMRI-ERIC is one of the largest Research Infrastructures for health research in Europe providing a gateway for access to biobanks and biomolecular resources and consists of National Nodes

across 24 Member States and IARC/WHO as International Organisation.

Materials and methods

BBMRI-ERIC has achieved significant success in engaging its extensive biobanking community in cutting-edge EU-funded projects that drive forward medical research and ultimately benefit patients. This active involvement not only enhances the visibility of our biobanks and their collections but also contributes to their sustainability.

Results

Our biobanking community has been actively participating in two EU projects that focus on facilitating sample and data provision. Firstly, the ISIDORe project that counts on the participation of 40 biobanks that offer retrospective and on-demand access to high-quality biological samples, along with detailed clinical phenotype data to support research projects related to COVID-19, various SARS-CoV-2 variants, and other epidemic-prone pathogens. The second project is CanSERV where 32 biobanks from across our community come together to facilitate access to retrospective and on demand collections of high-quality biological samples and high-quality associated data (i.e., OMICS) from cancer patients and control probands.

Discussion and conclusion

In both projects, BBMRI-ERIC´s biobanking community participates in making cutting-edge and customised research services available to the wider research community enabling innovative R&D projects and fostering precision medicine for patients benefit across Europe and beyond.

Keywords

biobanks, canSERV, ISIDORe, cancer, COVID-19

O22: Semmelweis Federated Data Warehouse for integrating biobanks and genomic data by privacy-preserving sharing at institutional level

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Description

The Semmelweis Biobank Network has a history of more than 10 years, preserving biological samples and clinical data of approximately 100,000 individuals, generated in routine operations and/or scientific projects of the University's clinics. However, these data repositories are managed by diverse, fragmented and non-interoperable data collection systems. Secure sharing of sensitive health as well as genetic data is still undeveloped in our country.

The project aims to create a versatile ecosystem to unite fragmented datasets from research and diagnostic workflows using Privacy-preserving Federated Learning. This will enable summary statistics of harmonized datasets over available biobanked samples and data.

Data harmonization plan considers interoperability by applying BBMRI standards, including MIABIS data model, as well as using standard codes for phenotype and disease ontologies for rare and common conditions. Conformity to OMOP data model for clinical data and the development of a software for the prospective augmentation of datasets. Existing datasets were mapped to identify synergies and intersections. Functionalities of federated analysis target (1) genetic landscape for Hungarian population, providing queries for variant frequencies over a gVCF catalog, (2) association analysis and (3) complex probabilistic models.

The development of an interoperable data warehouse with federated artificial intelligence-based data sharing to ensure the protection of

personal data can be considered as a pilot project based on the FedX Federated AI platform of E-Group. The members of the BBMRI Hungarian Node can use the know-how and methodology to implement data harmonization and sharing within their institutions.

Keywords

Privacy-preserving Federated Learning, gVCF catalog, MIABIS, data sharing

T8. Public Health Emergencies – Growing importance in the One Health concept

O30: Taï Chimpanzee Project – A platform for an One Health approach

Ulrich, Markus (1), Düx, Ariane (1), Bläsing, Dominic (1), Patrono, Livia Victoria (1), Leendertz, Fabian (1)
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Description

As part of the Taï Chimpanzee Project, habituated chimpanzees have been monitored in the Taï National Park in Côte d'Ivoire since 1979. Increasing disease outbreaks in the endangered chimpanzee communities made continuous health monitoring and pathogen screening necessary. Starting in 2000, a regular, non-invasive sampling protocol for known individual chimpanzees was implemented to collect fecal and urine samples. In addition, mortality monitoring of wild animals was introduced to gain knowledge about circulating pathogens.

Necropsies of carcasses are performed by trained veterinarians wearing full personal protective equipment. Tissue and swap samples are stored on-site in liquid nitrogen. All relevant samples from suspected cases are tested for pathogens in the field laboratory and later for confirmation in our laboratories. Samples are shipped in regular intervals as exempt animal specimen. All samples were obtained with research permits from local authorities and are subject to Nagoya Protocol, and depending on species, animal samples may be classified as CITES.

More than 90,000 samples have been collected and stored for the Taï chimpanzee project so far. The longitudinal behavioral observations combined with health monitoring of chimpanzees and their ecological neighbors as well as humans in Tai National Park helped to identify several infectious agents (HMPV, RSV, HCoV-OC43, MPXV, *Bacillus cereus* biovar*anthracis*, *Mycobacterium leprae*, *Mycobacterium tuberculosis*) and major risks for ape mortality. This helped in the development of strategies to protect apes from human pathogens and to inform the public health authorities on potential risks for humans to reduce zoonotic infection events and stabilize great ape communities.

Keywords

onehealth, wildlife, chimpanzee, subsaharan africa, tropical rainforest

O31: One Health Surveillance – a holistic approach to One Health assessment in Sub-Saharan Africa and Mecklenburg-Western Pomerania

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Description

To understand the complexity of pathogen evolution and emerging diseases a multi-modal, transdisciplinary approach is necessary. Combining human centered surveillance of risk groups and clinical surveillance with wildlife and livestock monitoring and the collection of environmental background data in a longitudinal, multi-centric approach enables comprehensive analyses in a One Health framework.
Thus, the One Health Surveillance Core Unit of the Helmholtz Institute for One Health focuses on the establishment of four sampling pillars:

Human biobanking: Based on two sampling approaches (risk groups and clinical surveillance) NPO swabs, serum, blood, stool and biopsy data are collected.

Animal biobanking: Local wildlife is monitored using mostly non-invasive approaches based on urine and feces, but also systematic protocols

to sample swabs, blood, feces, urine, and tissue or perform full necropsies if indicated. To further get insight into the possible emergence of zoonotic diseases bushmeat, arthropods and domestic animals are sampled.

Environmental biobanking: With a strong focus on biodiversity monitoring different environmental sampling strategies are used. Based on standardized protocols soil and water are collected, flies are caught, and leaves are swabbed.

Further enrichment: Additionally micro climate is measured using different data loggers for temperature and soil humidity, drone images are taken, and (standardized) questionnaires and (semi) structured interviews are used. Further ancient/museum samples are collected and analyzed to get insight into pathogen evolution.

A sufficient data management plan and biobanking strategy is necessary to deploy fast analytic pipelines and enable machine/deep learning approaches to combine all sample/data types in a meaningful way.

Keywords

One Health Surveillance, Biodiversity, Pandemic Preparedness, Human Cohort Study

O32: Unlocking the Power of Veterinary Samples: A Promising Source for "One-Health" Research

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Description

The importance of biobanks collecting high quality human biospecimens for research is widely recognized and promoted. However, the potential benefits of animal samples often remain underestimated, although the integration of veterinary biobank collections could add valuable findings for both human and animal medicine.

The One-Health concept goes far beyond the research of zoonoses, which is probably the best-known area in this field. Comparative medicine has a high potential, as pets and owners live in the same environment and are often affected by similar diseases such as obesity, diabetes, and cancer. To ensure reliable comparison between data obtained from animal or human samples, it is essential to adhere to the same standards when collecting and storing biospecimens and associated data. Following ISO standards for pre-analytics is crucial to guarantee accurate and consistent results for molecular analyses.

In summary, investigating veterinary samples in an integrative, transdisciplinary approach will open new insights into the transmission, etiology, diagnosis, prevention, and treatment of diseases, leading to a deeper understanding of disorders. Ultimately, this concept can result in the development of effective therapies that are effective across different species. To achieve this goal, bridging the gap between researchers in human and veterinary medicine is necessary. This is already recognized by incorporating the Veterinary University of Vienna with its VetBiobank into the Austrian national node, BBMRI.at. Collaboration with biobanking initiatives such as BBMRI-ERIC will enhance the impact and success of this research.

Keywords

veterinary biospecimens, One-Health, Comparative Medicine, preanalytics, BBMRI-ERIC

O33: "HPV vaccination outcomes: HPV seroprevalence in Lisbon area after 10 years

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Introduction

In Portugal, since 2008, young girls (born after 1991) are vaccinated against Human Papillomavirus (HPV), a sexually transmitted virus. Data regarding the effect that vaccine implementation had on HPV infection incidence is limited. After more than a decade, infections with genotypes covered by the vaccine should show a reduced seroprevalence in this population, whereas other high-risk genotypes could have grown in seroprevalence.

Methods

This study aims to analyze the HPV exposure profile of Lisbon residents over a minimum of 5 years, using the Biobank-iMM Healthy Donor Collection. The assessment involves measuring HPV-16, 18, 56, and 59-specific IgG antibodies in the donors' serum through VLP-based ELISA. Throughout this epidemiologic study of the HPV genotypes prevalence, it will be possible to correlate the results with their vaccination status, time of vaccination, sex, and age group.

Results

From a preliminary testing of the population (n=117), 97% of the vaccinated individuals and 13% of the unvaccinated individuals (all females, born in the 1980s) showed to be positive for the presence of the Ab anti-HPV-16 ($p<0,0001$).

Discussion

From the pilot study, the vaccinated population still shows protection conferred by the vaccine. The unvaccinated group born in the 1990s (all males) seems to benefit from herd immunity, comparing with the individuals born the decade before (1980s, 34% males), $p<0,05$. Further testing of the remaining
samples and HPV genotypes is needed to clarify the need of an update on the vaccination plan."

Keywords

HPV, vaccine, antibody, seroprevalence, Lisbon

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T12. Bioinformatics and -omics and big data

O45: Systematic molecular analyses of the NAPKON cohorts - an overview

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Description

As part of the German National Pandemic Cohort Network (NAPKON), a total of 7097 patients were recruited in 3 different cohorts (2753 SÜP, 710 HAP, and 3634 POP patients) by the end of 2023. Biosamples from 2520 patients (1217 SÜP, 300 HAP, and 1003 POP patients) for whom a complete set of biosamples was available, were comprehensively characterised using molecular techniques on 7 different analysis platforms.

Genotype data were generated, and proteome, metabolome, transcriptome and cytokine profiles were determined at up to three time points (baseline, acute phase, 3-month follow-up) for each HAP and SÜP patient and at baseline for POP. In addition, epigenetic (methylation, protein-DNA interaction) and virological data, including SARS CoV-2 sequences were collected. The various analytical data are integrated into the NUKLEUS infrastructure (NUM clinical epidemiology and study platform) with direct links to the respective aliquots analysed. The systematic analysis data is now available for research projects via the NAPKON use and access process.

As part of a DFG application, exome sequence data is also being generated and integrated into NAPKON. In another follow-up project, SAPCRiN (Sample Analysis in Post COVID Research in NAPKON), the 12-month follow-up samples will be analysed using the above-mentioned methods and evaluated with a focus on post Covid.

Here we provide an initial overview of the various methods and types of data generated in the context of systematic molecular analyses in NAPKON, with a focus on technical implementation and quality assurance. Both are essential for the provision and use of high-quality data.

Keywords

Systematic molecular analyses, NAPKON, OMICs

O46: Developing Inflammatory Polygenic Scores in Copenhagen Hospital Biobank data as a research tool to uncover links between inflammation, sex, and disease

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Introduction

Measuring nearly 50 inflammatory plasma biomarkers in a large biobank's complete sample set is complex and expensive. For cohorts lacking biomarker measurements but where genomic data is available, a cost-effective alternative includes calculating polygenic scores (PGSs) representing an individual’s genetic predisposition for increased or reduced inflammation levels. We present initial findings of our newly developed inflammatory PGSs in the Copenhagen Hospital Biobank (CHB), derived from real biomarker measurements in a cohort subset of 10,000 healthy individuals.

Methods

We measured plasma levels of 47 inflammation and vascular stress biomarkers in 10,000 sex and age-balanced participants from the Danish

Blood Donor Study (DBDS). General and sex-stratified genome-wide association studies (GWASs) were conducted on all 47 biomarkers. The GWAS results enabled the calculation of PGSs in unrelated DBDS participants and CHB patients (N≈300,000). Four "multi-PGSs" are under development, combining the individual PGSs into multi-scores reflecting four inflammatory pathway groups.

Results

GWASs identified genome-wide-significant loci for 39 out of 47 biomarkers, including sex-specific genetic variants for 25 of the 39. Individual inflammatory PGSs and two multi-PGSs (proinflammatory and growth factor/vascular stress/endothelial cell-associated) have so far been calculated in CHB. Preliminary tests on clinical register data in CHB reveal clear associations between increased inflammatory multi-PGS and diseases, including sex-specific differences.

Conclusion

These PGSs will play a crucial role in future CHB projects, shedding light on the genetic predispositions within inflammatory pathways in each sex with respect to the diseases investigated. This research will contribute to a deeper understanding of the links between inflammation, sex, and disease.

Keywords

inflammation, polygenic scores, big data

O47: Use of genomic data returned to biobank for predicting pharmacodynamic efficacy

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Introduction

CYP2C19 is a liver cytochrome P450 enzyme catalyzing the bioactivation of clopidogrel, an antiplatelet prodrug used in prevention of cardiovascular morbidity and mortality. Variation in *CYP2C19* gene is known to impact effectiveness of clopidogrel.

We examined the association of *CYP2C19* genotypes with adverse outcome events (AE) and platelet reactivity after initiation of clopidogrel medication.

Material & methods

Clinical data of the Turku University Hospital was combined with genotypes returned to Auria Biobank from the FinnGen project. All patients with genotype data and clopidogrel prescription, were included in the study (N=2807).

Patients were grouped into normal/fast and poor metabolizers based on *CYP2C19* genotypes.

AE was defined as an emergency procedure related to cardiovascular or cerebrovascular thrombotic event occurring within a year after clopidogrel initiation. For a subgroup of patients (N=265), adenosine diphosphate stimulated platelet reactivity data was available. The number of AEs and platelet reactivity was compared between genotype groups.

Results

The number of patients with AE (N=83) was higher in the poor metabolizers as compared with normal/fast metabolizers, but the difference was not statistically significant. In the subgroup analysis, number of patients with increased platelet reactivity was higher in poor metabolizers ($p<0.05$).

Conclusions

In this real-world biobank cohort, we were not able to demonstrate difference between normal/fast and poor metabolizers in the rate of AEs, which may be due to difficulty in identifying relevant AEs from hospital records. However, significant difference in platelet reactivity was observed, suggesting that genotype data in biobank may be useful in predicting pharmacodynamic efficacy.

Keywords

genotype, pharmacogenetics, biobank, clopidogrel

O48: MaMaMeDx: Metabolomic digital twins to improve diagnostics and screening using breast cancer biobank samples as an example.

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Description

The combination of serum samples from breast cancer patients from the PATH Biobank, collected according to high qualitative standards, with lifespin's metabolomic sample analysis opens a major opportunity for improved early breast cancer detection and stratification.

In the MaMaMeDx project, 3000 biobank samples from diseased donors are being digitized using the lifespin system and the metabolomic constitution of the patients is analyzed. lifespin's holistic approach, i.e. the acquisition of more than a hundred quantitative parameters per sample without having to know specifically what one is expecting, enables samples from different breast cancer subtypes to be compared with each other and with samples from healthy donors. This search for biomarker ratios and patterns specific to the manifestation of different breast cancer subtypes enables the reconstruction of metabolomic correlations with the course, type and severity of the disease and the identification of characteristic metabolite patterns to specific tumor stages. lifespin has a comprehensive control collective (> 200,000) of digitized metabolomic profiles available for matching. Knowledge of these marker ratios should lead to improvements in early breast cancer detection, diagnosis and differentiation, as well as to the follow-up of therapy outcomes (monitoring), in order to detect possible diseases earlier and in a more differentiated manner than before.

These cross-sample, cross-study and cross-clinic approaches require comprehensive insights into the entire process chain, from sample collection and handling to biobanking. This is possible by the lifespin platform, providing the presented valuable, correlations and challenges for different building blocks and players along the process chain.

Keywords

Metabolomic Profiling, Metabolomics, Digital Twinning, Big Data Analyses, Quality Control Markers

T16. Epidemiological biobanking – cohorts as a shop window for populations

O60: Study protocols, biobank features and sample collection for a longitudinal population cohort study in Norway. The Tromsø study.

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O61: Addressing the Asian Genomic Gap – the National Precision Medicine Programme

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O62: From population-based towards healthcare integrated biobanking - the journey of the Leipzig Medical Biobank

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Introduction

The Leipzig Medical Biobank (LMB) at the University Leipzig is a state-of-the-art biobank for quality-assured collection, processing, storage and provision of biospecimens. Founded in 2010, mainly liquid samples such as urine, serum, plasma and saliva from epidemiological studies of the Leipzig Research Centre for Civilisation Diseases (LIFE) were processed. In 2017, healthcare-integrated biobanking of oncology patients with solid and haematological malignancies was launched with clinical partners and the Institutes of Pathology and Laboratory Medicine. In 2021, the collection of liquid, tissue and plaque samples from patients with cardiovascular diseases started.

Methods

Samples are processed in a highly standardised and traceable manner and stored at temperatures of -80°C or <-150°C. Storage is performed in strict compliance with an

uninterrupted cold chain and constant monitoring. All samples are barcoded and can be linked to quality data and donor clinical data.

Results

More than 1.4 million samples and data sets are available from around 60.000 visits (40.000 donors) for researchers. The biobank has been able to support researchers with around 160.000 samples in > 100 projects. In addition to research on stress, allergy, heart disease and dementia, improvements have also been made in diagnostics. Reference ranges of certain biomarkers covering the whole lifespan have been investigated and are now used at the University Hospital Leipzig.

Conclusion

The LMB is the central biobanking facility of the Medical Faculty of the University Leipzig. Samples are processed according to SOPs using state-of-the-art techniques and equipment. The LMB is open to scientific collaborations with internal and external partners and promotes high-level research.

Keywords

population based biobanking, healthcare integrated biobanking, biomarker research

O63: Swedish biobank cohorts and legal aspects on their potential for use within Precision Medicine

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Introduction

Swedish biobank cohorts are a valuable resource for Precision Medicine research. The Swedish personal identity number facilitates the combination of samples and data with data from national health registers and medical records. This makes it possible to study disease risk factors in samples obtained before a disease has become apparent. The large potential of these resources can, however, be inhibited by the complexity of the national and international legal landscape.

Methods

Biobank Sweden aims to improve the use of existing samples and data through 1) increased visibility of biobank cohorts; 2) highlighting their importance for medical research; 3) support to individual researchers; 4) initiating a national workshop series where legal counsels across Sweden discuss how different legal interpretations can be harmonized to enable more effective research.

Results

There are several well-documented biobank cohorts in Sweden. Some are moving towards becoming databanks through extensive sample characterization. Others have intact sample collections (blood, plasma, DNA, tissue) that can be used to study unique trends over time. The ongoing legal workshop series has so far addressed the need for a harmonized interpretation of roles and responsibilities regarding personal data processing, when biobank samples and data are shared between different research entities. In addition, national template-agreements for sample and/or data sharing could increase the possibilities for more effective research.

Discussion and conclusion

We hope that sharing our findings will increase the interest in Swedish biobank cohorts and spark a discussion that will enable knowledge transfer between countries battling similar but different legal issues.

Keywords

Swedish biobank cohorts; legal landscape; national legislation; GDPR; data sharing; MTA.

T.20 Artificial intelligence in Precision Medicine

O78: Legal and ethical issues of using AI in Biobank in countries with an average income level

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Introduction

Using artificial intelligence (AI) in biobanks in countries with average income levels raises several legal and ethical considerations.

Methods

For tool development, a survey was designed and sent to biobank managers and IT biobank specialists worldwide. We have created a list of about 36 biobank managers from East Europe.

Results

Privacy concerns, particularly in AI analyzing sensitive genetic and health data, necessitate the establishment of robust safeguards to protect donors' confidentiality.

Intellectual property rights become a legal intricacy as AI-generated findings raise questions about ownership.

Legal challenges may arise in cross-border collaborations, necessitating a global perspective to address ethical concerns on an international scale.

Conclusion

As a result of the project, we have identified comprehensive aspects of the legal and ethical related to integrating AI into biobanking in middle-income countries. By addressing these challenges, it seeks to contribute to the development of robust frameworks that balance scientific progress with ethical imperatives, fostering responsible AI use in the advancement of medical research.

Keywords

Biobank, artificial intelligence, legal and ethical governance

O79: Fast-track AI-assisted immunohistochemical stain quantification and statistical analysis of TMAs

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Background

Spatially resolved biomarker examination is integral to cancer research. Associated processes are both time and cost-intensive. In response, we introduced a streamlined workflow, encompassing Tissue Micro Array (TMA) construction, automated annotation of structured and ELSI confirmed clinico-pathological data, AI-based immunohistochemical (IHC) stain detection, and script-based statistical analysis.

Methods

The pipeline comprised: 1. ELSI-confirmed data centralization from structured histopathological and clinical reports, 2. prospectively collected FFPE tissue specimens, 3. TMA-construction using the TMA Grandmaster (3DHistech), 4. AI-assisted image analysis with QuPath (version 0.5.0) / HALO AI (version 3.6) and 5. statistical analysis using R within the R-Studio environment. The TMA design included replicates from representative tumor regions and layout randomization to minimize batch effects. Classifiers were trained to evaluate subcellular tumor compartments and surrounding stroma tissue. R-Scripts encompassed various stages, including data preparation (merging clinical and TMA data), pre-analyses (distribution, cut-off calculation, descriptive analyses), statistical testing (log rank test, uni- and multivariate analyses, Cox regression) and graph plotting.

Results

Streamlining semi-automated TMA construction with AI-assisted IHC-stain detection and script-based statistical analysis facilitates rapid, objective correlation of tumor marker expression with clinico-pathological variables. This leads to a significant reduction of workload, costs and consumables. TMA-sections minimize experimental variability and batch effects, enhancing inter-tissue comparability and result validity. Constructed TMAs are reusable and

remain accessible for subsequent experiments after several years, allowing for the application of advanced techniques such as MALDI-TOF, CyTOF, and spatial transcriptomics.

Conclusion

The presented semi-automated TMA construction, stain evaluation, and statistical analysis pipeline is time- and cost-effective, easily applicable and reproducible, extending beyond IHC-based analyses.

Keywords

Tissue microarray, Artificial Intelligence, Immunohistochemistry

O80: Unlocking the Potential of Big Data and AI in Medicine: Insights from Biobanking

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Introduction

AI is increasingly expected to improve accuracy and efficiency in diagnosis and treatment, particularly in identifying biomedically relevant patterns, facilitating progress towards individually tailored preventative and therapeutic interventions. These applications belong to current research practice that is data-intensive. While the combination of imaging, pathological, genomic, and clinical data is needed to train algorithms to realize the full potential of these technologies, biobanks often serve as crucial infrastructures for data-sharing and data flows.

Methods

The paper is built on outcomes and observations from our involvement in several projects about AI and biobanking.

Results

In this paper, we argue that the 'data turn' in the life sciences has increasingly re-structured major infrastructures, which often were created for samples and data, as predominantly data infrastructures. An increased use of AI-based technologies marks the current developments at the forefront of the big data research in life science and medicine that engender new issues and concerns along with opportunities.

Discussion

At a time when secure health data environments, such as EHDS, are in the making, we argue that such meta-infrastructures can benefit both from the experience and evolution of biobanking, but also the current state of affairs in AI in medicine, regarding good governance, the social aspects and practices, as well as critical thinking about data practices, which can contribute to trustworthiness of such meta-infrastructures.

Keywords

biobanks, precision medicine, artificial intelligence, big data, infrastructures

O81: AI, Digital Pathology and TMA to explore cancer biology

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Description

Over the years, the major bottleneck in translational research has been the difficulty in analysing large numbers of clinically well-defined tissue specimens. AI based on deep learning algorithms, in conjunction with Digital Pathology, will play a crucial role in the evaluation of multiple biomarkers (multiplexing) and quantitative analysis for precision medicine.

In addition, Tissue microarray technology (TMA) has played a crucial role in overcoming this challenge by enabling the simultaneous analysis of thousands of tissue samples in a single experiment, offering a high degree of standardization, as all tissue samples in the same experimental conditions. TMAs in conjunction with AI and Digital Pathology can be created from various types of diseased and non-diseased tissues, including formalin-fixed and frozen tissues, xenograft tissues, cell lines, and organoids/spheroids. The versatility of TMA technology extends beyond *in situ* analysis (immunohistochemistry-fluorescence) to include genomic and transcriptomic investigations, Spatial Biology facilitating proteomic comparisons.

Keywords

#AI, #DigitalPathology, #TMA, #NGS, #SpatiaBiology, #Multiplexing, #ørganoids, #ValidatedAntibodies

TRACK 2: Quality & Innovative methods

T2. Pre-analytic impact on sample quality – means & measures

O7: NMR fingerprinting for sample quality assessment

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O8: Enhancing Reproducibility in Biobanking: A Molecular-Based Model for Quality Control of Serum and Plasma Samples in Multi-Omics Assays

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Introduction

The development of biobanks entailed improving the technological hinterland for efficient management in large amounts of samples, clinical data, and big data from multi-omics assays. This study addresses the critical issue of sample quality and reproducibility in biobanking, emphasizing the necessity for high-quality biospecimens to meet the demands of advancing technologies.

Material & Methods

The research introduces the molecular-based quality control model designed to assess and enhance the quality of serum and plasma samples. The model comprises three essential steps, including visual hemolysis assessment, spectrophotometric analysis for hemolysis monitoring, and examination of RNA purification efficiency using qPCR. The OMICS-QC model was validated using 347 serum and 694 plasma samples collected and preserved under standardized conditions in the Biobank at the Medical University of Bialystok, Poland.

Results

Within the OMICS-QC model, we established a process composed of successive steps with hemolysis monitoring and control of RNA extraction. Visual hemolysis assessment and spectrophotometric analysis enabled quick and cost-effective identification of compromised samples, while the RNA purification process was scrutinized using synthetic RNA spike-ins and endogenous microRNA assays. The OMICS-QC model successfully qualified samples with RNA Integrity Number (RIN) ≥ 7 for omics analysis, ensuring the reliability of downstream applications.

Discussion and Conclusion

The study's findings underscore the importance of pre-analytic measures in maintaining sample integrity and reproducibility. An established OMICS-QC model can be recommended in the process of specimen qualification before the multi-omics assays for reproducibility and replicability in research.

Keywords

omics; quality control; plasma; serum; RNA Intergity

O9: Evaluating biobank processes – a proficiency test concept to enhance sample quality

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Description

Quality-assured access to human biosamples and the associated phenotype data are essential factors for reproducible and high-qualitative biomedical research, particularly for omics technologies. Thus, QC and QA as well as standardization of biosample handling processes are of major importance in biobanking and translational research.

QM concepts play a crucial role in monitoring sample and process quality. In particular, proficiency tests (PTs), e.g. in the context of laboratory diagnostics in the healthcare sector, are well-established instruments for evaluating analytical performance and optimizing critical process steps. Unfortunately, proficiency-testing programs for biobanks and their core processes are only partially established, primarily focusing on quality of nucleic acids and PBMCs. Essential biobank processes such as e.g. entry control, aliquoting of body fluids and shipment of samples are usually not taken into account. The purpose of our proficiency-testing concept, developed within the German Biobank Alliance, is to fill the gap between established PTs and the need to control crucial biobank processes. Therefore, we have developed PTs for body fluids to check entry control, processing time, homogeneity and volume accuracy of sample aliquots. Moreover, compliance of appropriate shipping temperatures on dry ice in accordance with international shipping standards and proper packing of potentially infectious biospecimens were also evaluated.

Here we present the results from two PTs, conducted with 21 national and 18 international biobanks. The proficiency-testing concept developed by us serves as a basis for the assessment of biobank processes and will be refined in the future to improve harmonization and minimize process variability between biobanks.

Keywords

Proficiency Test, Sample quality, Sample homogeneity, Entry control, Volume accuracy

O10: Pre-analytical process for biobanked biomaterials poorly reported: a cause of replicability crisis in clinical biomarker research?

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Introduction

Biobanks form a rich source of biomaterials for clinical biomarker research. Pre-analytical processing of biomaterials stored in biobanks determines their fit-for-purpose. Underreporting of pre-analytical methods in scientific articles carries risk of non-valid conclusions. To improve insights in the scope of this problem, we assessed pre-analytical reporting quality in recent clinical biomarker research using biobanked plasma and serum.

Methods

We selected 294 articles published between 2018-2023 that used biobanked plasma or serum samples for clinical research. Data was extracted from these articles on how well pre-analytical processes were reported. Articles were analysed based on pre-analytical elements derived from the Sample PREanalytical Code (SPREC) combined with the Biospecimen Reporting for Improved Study Quality (BRISQ) criteria.

Results

Many pre-analytical processes were underreported in most papers. This was also the case in the most used biobank (7.8% of articles), the UK biobank. Fasting (time) (31.0%), freeze-thaw cycle (22.8%), transport (8.5%), pre- and post- centrifugal delays (17-22%), and centrifugal settings (20-30%) are all rarely reported on. Demographics (97.3%), long-term storage temperature (81.0%), blood tube additives (82.7%), and inclusion period (72.4%) are generally well described. We found no relation between reporting proportion or year of publication and journal impact factor, and only small difference in reporting pre-analysis elements for different analysis purposes.

Conclusion

The pre-analytical process is not or poorly described in recent literature. This is a potential threat to research replicability. We advocate raising awareness about importance of reporting pre-analytical processes for biobanking in scientific articles, and to optimize and implement pre-analysis reporting guidelines.

Keywords

Biomarker, biobank, pre-analysis, blood, serum, plasma, research waste, replicability, clinical research.

T6. The road towards ISO 20387 accreditation

O23: Finland: Auria Biobank

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O24: Balancing Quality and Costs: Challenges in Implementing ISO 20387 in Biobank Operations

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Introduction

Biobanks, essential for scientific research, grapple with diverse regulations and standards, with ISO 20387:2018 attempting to standardize operations. However, ensuring high quality poses funding challenges for these institutions, demanding specialized and expensive equipment for safe biological sample storage. As this system's intricacies are poorly understood, our analysis aims to guide biobanks seeking to implement ISO 20387.

Materials and methods

The University of Szeged's Biobank, ISO 20387:2018 certified for two years, boasts advanced infrastructure, including industrial cold storage, surveillance, and smart technologies for environmental monitoring. Over the past two years, we analyzed costs primarily associated with maintaining ISO 20387 and ensuring operational processes, calculated over a complete annual cycle.

Results

Results reveal that salaries constituted 22.87%, LIMS, IT, and software 27.59%, laboratory equipment 14.63%, materials and consumables 3.81%, service costs 35.45%, quality management system costs 11.08%, other services 4.59%, and reimbursement costs 1.86%. Significantly, the maintenance of ISO 20387 increased our costs by 29%.

Discussion and conclusion

Our two-year ISO 20387-certified biobank operations saw annual expenses rise by nearly 30%. This cost escalation, while ensuring high quality, presents a disadvantage in a competitive market where potential partners prioritize cost considerations for biobank storage and services.

Keywords

ISO 20387, biobank, cost, market

O25: Implementation of ISO 20387 standards: Transitioning with human tissue authority (UK) requirements

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Background

In preparation for ISO 20387 accreditation, the KHP Cancer Biobank has undertaken an assessment of how to implement the ISO standards alongside existing Human Tissue Authority (HTA) requirements. The information will be used to plan resource requirements to achieve accreditation.

Methods

Individual ISO standards were audited against the Biobank quality management system and a gap analysis created. Identified shortfalls were raised with the UKAS Biobank Steering Group to establish if these were unique to the KHP Cancer Biobank or a more general issue that other members had experienced. Regular meetings were held with the Biobank staff to obtain input and engagement and a phased schedule of ISO standard upgrades implemented.

Results and discussion

ISO standards are more specific and surpass those required for HTA licencing. Reviewing differences by sector was the most strategically valid option and partitioning the work led to better team engagement. The use of themed meetings to present information and encourage discussion led to improvement in assimilation of new policies and procedures. Higher success rates were achieved where processes were

widely discussed by the team, rather than arbitrarily imposed due to quality reasons. Indications are that implementing ISO standards will build upon those already in place for the HTA licenced Cancer Biobank.

Conclusion

An efficient transition revolves around tactical approach and knowledge of the regulations. HTA standards create a solid base to develop an accreditation plan but success lies with full staff engagement.

Keywords

ISO, ISO 20387, quality, standards

O26: SBP labels, the Swiss Quality program to support national biobanks towards the implementation of the ISO 20387:2018

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Description

In its mission to support high-quality biobanking, Swiss Biobanking Platform (SBP) aims at engaging biobanks adopt a Quality strategy and work according to best practices. As the research infrastructure funded by the Swiss National Science Foundation, SBP oversees the coordination of human and non-human biobanking at national level.

Following the publication of the ISO 20387 in 2018, the importance to establish robust QMS became prominent. In Switzerland, SBP responded by developing a stepwise quality program aligned with the ISO 20387 requirements to support biobanks developing their quality strategies and implementing this professional standard.

SBP has thus developed a comprehensive service using the biobank SQAN online tool to evaluate biobank practices. This assessment measures compliance with the applicable legal framework, and the ISO 20387 requirements (operational processes, resources, QMS). Once fulfilled, three distinctive labels can be awarded: the VITA label (governance), as a prerequisite to appear in the national and BBMRI Directory and the NORMA and OPTIMA labels (Quality) to demonstrate the minimum requirements in terms of process standardization and quality assurance, respectively. While governance evaluations are conducted remotely, onsite audits are required for quality assessments.

By guiding Swiss biobanks through a stepwise implementation of the ISO 20387, SBP empowers them to increase the overall quality level of their activities to support (inter)national research. Furthermore, SBP is laying the groundwork to assist when ready the Swiss Accreditation Service in launching a nationwide accreditation program.

Keywords

Quality program, ISO 20387, Practice harmonization, Accreditation, QMS

T9. Modelling and regenerative medicine – artificial cellular models

O34: Biobanks as the Cornerstone of Future Medicine

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O35: 3D organoids and ex vivo cultivation of tumor tissues in oncological research and personalized medicine

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Introduction

Current in vitro model systems do not fully reflect the biological and clinical diversity of tumors. Organoids are 3D in vitro cell cultures that can better recapitulate the heterogeneity of the disease and preserve the characteristics of the original tumor. Short-term ex vivo tissue culture may also facilitate drug testing in personalized medicine. In our work, we focused primarily on prostate cancer.

Material and methods

For organoid culture, we processed both tumor and normal tissue from 50 patients. In addition, we used the ex vivo tissue culture technique and performed a short-term experiment using

gemcitabine and the Chk1 inhibitor MU380 in samples from 10 patients.

Results

We established organoid culture from 58% of tumor (29/50) and 69% of normal tissues (20/29). Immunohistochemical staining of two representative cases revealed cellular positivity for pan-cytokeratin confirming the presence of epithelial cells. However, overexpression of AMACR and ERG proteins in tumors was not maintained in organoids. In ex vivo cultures, tissue samples from prostatectomies mostly showed a low rate of Ki-67 positivity. Only one case showed a high proliferation rate for drug testing, and tumor tissue was present in all samples tested.

Conclusion

We managed to establish cultures of organoids and tissue fragments from primary prostate tumors. However, the expression of tumor markers was not preserved in the obtained organoids. Inconsistent morphology and low proliferation made drug test results difficult to interpret in most cases. Nevertheless, these approaches may hold promise when using tissues from metastatic castration-resistant prostate cancer.

Keywords

organoids, ex-vivo tissue culture, personalized medicine, prostate cancer

O36: Decoding Cellular Plasticity and Microenvironment Crosstalk in Gastro-Esophageal Adenocarcinoma through a Living Biobank of Patient-Derived Organoids and Cancer-Associated Fibroblasts

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Introduction

Biobanks play a pivotal role in advancing healthcare research, providing critical infrastructure for personalized medicine. We aim to explore the impact of cancer specific biobanking, emphasizing its potential to expedite a focus on molecular mechanism of tumor cell plasticity and cancer-associated fibroblast in gastro-esophageal adenocarcinoma (GEC).

Materials & Methods

SOP was designed and optimized for both liquid biopsy and tissue biopsy processing. EDTA-plasma, serum and PBMC samples were collected. Treated native tumor specimens were collected and dissociated for either frozen storage or tissue culture downstream applications. Both direct and in-direct co-culture systems were utilized in this study.

Results

Over the last 7 years, Cologne Surgical Oncology Biobank has recruited over 1400 esophageal cancer (EC) and more than 300 gastric cancer liquid biopsies. Among EC, 74% cases are esophageal adenocarcinomas (EAC). Paired EAC tumor-derived organoids and cancer-associated fibroblasts were established in vitro offering a viable approach for studying tumor-stroma crosstalk. We tested chemotherapeutic drugs, targeted therapy and immunotherapy reagents in 3D co-culture model to evaluate the therapeutic effects.

Discussion and Conclusion

Our findings underscore the transformative potential of living biobanks (patient-derived organoids and cell lines) in GEC cancer research. Through a translational study of GDF-15, we demonstrate the significant contributions of biobank to advancing biomedical research, research fundings and interdisciplinary collaborations, which eventually may improve the patient outcome with individualized strategies.

Keywords

living biobank, patientd fibroblasts, liquid biopsy, gastro-esophageal adenocarcinoma

O37: A review of the ELSI challenges of organoid biobanking

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Introduction

From human biological material, using modern techniques, it is possible to generate organoids that reproduce the functioning of healthy organs, tumors and, recently, also the embryo structure . The applications of organoid research will be many, and possible applications for regenerative medicine are already being discussed. In this context, the role of biobanks is central to addressing the ethical, legal, and social implications of these innovative methods.

Method

As part of the horizon scanning activities of the WP2 - ELSI of #NextGenerationEu 'Strengthening BBMRI.it' project, in the absence of a comprehensive regulatory framework on organoids, an effort to reconstruct the role of biobanks was made by analyzing the emerging debate and systematically considering the general rules applicable to biological materials from which organoids are generated, the general rules on biobanking and research activity with samples and data.

Findings

The debate is still in its initial stages, but major challenges can already be identified:

- The role of the biobanks: from mere custodian to 'administrator' of biological material.
- The legal nature and 'moral' status of the organoid and its consequences.
- How to involve patients/participants: rethinking consent.

Discussion and conclusions

The role of biobanks is evolving as techniques and processes innovate. This evolution will bring changes in the relationship of the biobank with biological samples and with patients/participants that will have to be adequately considered when they give their consent to biobanking as the act of establishing the fiduciary relationship between biobank, patient/participant and the samples derived from them.

Keywords

organoids, ELSI challenges, role of biobanks

T17. Getting more from less – liquid biopsies, biopsies and fine needle aspirates for molecular technique

O64: Minimally invasive fine needle aspiration-based molecular diagnostics in support of precision cancer medicine

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Background

Diagnostic tissue biopsies are required to select therapy for patients with solid tumors. However, core needle biopsies can cause complications and may be difficult to repeat longitudinally. Sampling via fine needle aspiration biopsy (FNA) is globally established, minimally traumatic and can be repeated during treatment.

We have developed standard operating procedures for collection, biobanking and sample preparation of minimal FNA materials, compatible with clinical routines and targeted analysis of mutations, as well as gene and protein expression. Expression levels of proteins (leftover material only) were profiled (olink.com), and data were analyzed with statistical tools provided by e.g. Qlucore Omics Explorer (qlucore.com) and machine learning strategies to characterize the tumor microenvironment (TME), detect target molecules and to identify tentative predictive biomarker signatures.

We will describe the development of FNA-based atraumatic molecular cytology for precision cancer medicine (PCM) and the identification of tentative biomarker signatures, proteins related to the immune TME and to resistance to immunotherapy. The methodology is highly sensitive and reproducible and permits multi omics profiling with assessment of biomarkers for diagnosis, therapy selection and monitoring of therapy.

Keywords

biomarkers, fine needle aspiration biopsy, immune signalling, machine learning, prostate cancer, proximity extension assay

O65: Rescue of Low-Yield DNA Samples for Next-Generation Sequencing Using Vacuum Centrifugal Concentration in a Clinical Workflow

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Description

The implementation of next-generation sequencing (NGS) in clinical oncology has enabled the analysis of multiple cancer-associated genes for diagnostics and treatment purposes. The detection of pathogenic and likely pathogenic mutations is crucial to manage the disease. Obtaining the mutational profile may be challenging in samples with low yields of DNA—reflected by the type of biological material, such as formalin-fixed paraffin-embedded tissue (FFPE), needle biopsies, and circulating free/tumor DNA, as well as a sparse tumor content.

Moreover, standardized strict procedures for the extraction of DNA in a clinical setting might contribute to lower amounts of DNA per µL. The detection of variants in low-yield DNA samples remains a challenge in clinical diagnostics, where molecular analyses such as NGS are needed. Here, we performed vacuum centrifugation on DNA extracted from five FFPE tissue blocks, with concentrations below 0.2 ng/µL. Through NGS analysis, we found that low-yield DNA samples could be concentrated to sufficient levels, without compromising the mutational profile.

Keywords

Vacuum Centrifuge, Low-yield DNA, Linear regression models, Next-generation sequencing, cancer, targeted therapeutics, clinical diagnosis.

O66: Verification of a Method for Generating Single-Cell Suspensions from FFPE Cervical Tissue for Flow Cytometry Analysis and Cell Sorting

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Description

Formalin-fixed paraffin-embedded (FFPE) tissue blocks are abundant and valuable resources for cancer research, but tumour heterogeneity and limited tumour content are problematic especially for smaller tissue samples. Strategies to enrich tumour cell content such as single-cell suspensions may overcome such challenges. Presented here is a method verification to generate FFPE tissue-derived single-cell suspensions and their application in flow cytometry and cell sorting.

Cervical squamous cell carcinoma (CSCC) tissue (n = 10) was used as study model. The Miltenyi FFPE Tissue Dissociation Kit and gentleMACS Dissociator with heaters, were used to prepare single cell suspensions from 100 µm tissue sections. The Amnis Image Viewer, the Beckman Coulter Flow Cytometer and BD FACSmelody were used for single cell analysis and cell sorting. Conjugated antibodies for Cytokeratin-FITC and Vimentin-APC were used to identify cancer and non-cancer cell populations, respectively. FlowJo and DX Flex software were used for analysis.

Single-cell suspensions were obtained, with cell yields ranging from 0.645 x 106 to 3 x 106 cells. Cytokeratin+/DAPI+ cells depicted cancer cell populations (median: 26.72%), whilst Vimentin+/DAPI+ indicated the proportion of non-tumour cell populations (median: 34.73%). The Cytokeratin+/DAPI+ and Vimentin+/DAPI+ populations were sorted for each sample.

In conclusion, we verified the method for FFPE-derived single cell suspensions using the Miltenyi gentleMACS system and FFPE dissociating kit. Cancer versus non-cancer cell populations were identified with flow cytometry and sorted. Single cell suspensions not only provide a method for tumour enrichment, but

also expands the secondary use of FFPE tissue blocks for molecular applications.

Keywords

FFPE- Formalin-fixed Paraffin Embedded, CSCC-Cervical squamous cell carcinoma

O67: Advanced Biobanking for Adult Adipose tissues-derived Stem Cells

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Introduction

Regarding the project "Strengthening of the Biobanking and Biomolecular Resources Research Infrastructure of Italy (BBMRI.it)", a biobank 4.0 will be established for adipose tissue derived stem cells (ADSCs) isolated from adult biopsies. This is of highly interest for scientific communities to study the onset and progression of metabolic, neoplastic and degenerative diseases.

Material and Methods

ADSCs will be isolated from adipose tissues following an enzymatic digestion with Collagenase. To guarantee the quality of samples, the isolated cells will be characterized by analyzing the three minimum criteria stabilized by the International Society for Cellular Therapy (ISCT) as: the growth in adhesion, the presence/absence of surface markers by FACS and the ability to differentiate in adipocytes, chondrocytes, and osteoblasts.

Results

The results expected will be focused on the creation of a wide organized collection of high-quality biological samples and data information associated, including genomic profile and the analysis of growth factors, cytokines and chemokines secreted by cells (Sequencing and Pyrosequencing systems, high-throughput genotyping, multi-ELISA processing). The advantages of the use of ADSCs isolated from adult biopsies is due to the less invasive and more accessibility of adipose tissue.

Discussion and Conclusion

The availability of high-quality biological materials provides the basis for more reliable and reproducible research in the context of predictive features studies for regenerative therapy. ADSCs can be useful for the development of new biomarkers for diagnosis and the discovery of new therapeutic approaches in the field of personalized and translational medicine. As such, biobanks represent the cornerstone of any research infrastructure.

Keywords

biobanking, biobanks 4.0, adipose derived stem cells (ADSCs), adipose tissue, high-throughput genotyping, BBMRI.it

T21. Tools to achieve quality – (new) standards for biobanks

O82: The Woven Web of ISO 20387, ISBER Best Practices, and Interrelated Tools

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O83: SBP SMPL: Helping biobanks visualize and configure their daily practice

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Description

Biobank information management systems (BIMS) have become a necessary tool towards proper documentation and traceability of biological samples. Unfortunately, deploying such a solution is often prohibitively complex, time consuming and expensive. The biggest challenge is the simultaneous complexity and uniqueness of processes used in each biobank protocol, which prevents availability of off-the-shelf solutions, resulting in the need to externalize many hours of custom development.

In its mission to promote interoperability and access to well-documented samples at the national level, Swiss Biobanking Platform (SBP) has developed an innovative event-based

method for accurately modelling any biobanking workflow through visual building blocks.

This method offers the following benefits:

1. It allows biobank administrators and principal investigators to make most custom changes themselves, without relying on dedicated IT services.
2. It facilitates day-to-day operations and reduces the need for dedicated training.
3. It readily integrates SBP's harmonized datasets and recommendations for improved data quality.

This method has been implemented into SMPL, a plugin module currently built for DiData BIMS, and deployed for the use of eight pilot biobanks. SMPL has been challenged with various workflows, including tissue, liquid, and animal-based biobank protocols. An associated business model ensures biobanks are provided an affordable solution.

By normalizing the description of biobank workflows, SMPL furthers possibilities for standardization and interoperability in the field, ultimately improving data quality. Next steps include the porting of SMPL to other platforms, and further integration with SBP's growing ecosystem.

Keywords

interoperability, workflows, data management, data quality, information visualization, bioinformatics

O84: Ensuring and Sustaining Data Quality in the Biobank Information Management System

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Introduction

Biobank Antwerp is organised in a decentralised way where researchers can opt to perform their own sample registrations. This brings a challenge to the biobank which has to guarantee that registered data meet quality standards. Our BIMS system (Agilent), facilitates maintenance of high data quality and helps to align with the fundamental quality criteria being accuracy, relevancy, completeness, timeliness, and consistency.

Results or findings

Various checks were put in place to ensure user-registered data meet quality requirements. Consistency is stimulated via upfront translation of the study protocol in the BIMS system, not allowing researchers to deviate. Accuracy, relevancy and completeness are pushed through the implementation of rules, value expressions, automated email notifications, fixed/dynamic field options, required field attributes, highlighting of questionable data, non-conformities, and BI reports. Timeliness is more difficult to monitor, but will be partly streamlined through an automated connection with the electronic patient file in the Antwerp Hospital were a majority of samples come from.

These elements are captured within three lines of control. 1) daily by users, 2) weekly by quality assurance officers and 3) monthly by the quality manager. Processes are monitored through Key Performance Indicators and internal audits.

Discussion and conclusion

Despite these extensive efforts violations to the five main data quality criteria persist. A continuous investment in training to empower users and more intensive cooperation with major research groups already resulted in notable improvements in data quality. However, correct sample registration by researchers rather than biobank staff, remains a challenge.

Keywords

Biobank; Sample Registration; Researchers; Research; Sample Quality; Quality Standards; Quality Control; BIMS

O85: Advancing Reliability and Trustworthiness of Research Results with the Common Provenance Model for Biological Material and Data

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Introduction

With the increasing importance of data and specimen sharing, the need for processes that enable reliable, non-repudiable, cross-organizational documentation that supports the traceability of both physical and digital research objects and their metadata has become imperative. In addition, for biological materials used in research and development, specimen collection must be in compliance with regulatory requirements and benefit sharing must be addressed. As a result, there is an urgent need for standardized documentation of biological materials and data processing.

Material and Methods

Experts from ISO TC/276 “Biotechnology” together with BBMRI-ERIC and EOSC-Life WP6 have initiated a project for the development of a standardized data model for biological material and data provenance.

Results

The specification covers the entire lifecycle of biological specimens and data, from specimen collection, processing and storage to final analysis and data collection, processing and analysis, including both primary and secondary use. The Common Provenance Model is designed to enable:

- Backward and forward traceability of the physical or digital objects and their derivatives, including their provenance;
- Evaluation of experimental results and data analysis;Automated monitoring and control of specimen and data quality;
- Assessment of fitness for purpose of specimen and data;
- Confidentiality and privacy.

Discussion and Conclusion

The data model provides a framework for capturing, in a structured and standardized way, information and metadata generated during the lifecycle of biological samples and data processing and analysis. By linking the information from each process step, a coherent chain of provenance information is created that can be traced and queried.

Keywords

Provenance, Biological material, Standardization, Data Quality, Fitness for Purpose

TRACK 3: Stakeholders in the Spotlight

T3. Training and education for quality improvement and knowledge generation

O11: Learning by doing in biobanking – Czech-Bavarian cross-border, interdisciplinary experience"

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O12: The first Swiss educational program on Biobanking for a large audience to unlock biobanking from basic knowledge to specialized professionals

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Description

The Certificate of Advanced Studies (CAS) in Biobanking, a collaborative effort between the University of Geneva, Swiss Biobanking Platform, Institute Pasteur, and HSeT Foundation, is a pioneering training initiative in Switzerland. This advanced program addresses the critical need to equip individuals and institutions interested in biobanking and research, with essential knowledge and proficient skills for effective biobank establishment and management.

The meticulously designed curriculum focuses on elevating standards in quality, management protocols, and interconnections within clinical laboratory medicine and cutting-edge research. The program adopts a progressive three-module structure, facilitating a stepwise acquisition of knowledge from beginners to highly specialized professionals. Employing a blended learning approach, incorporating e-learning and online lectures, the teaching language is English.

Module 1 provides foundational insights into biobanking and is tailored for those seeking to grasp the basics, including PhDs, physicians, clinical researchers, ethics committees, and patient organizations. Module 2, about the implementation of biobanking processes, targeting professionals with roles in biological sample management and offering intermediate-level insights. Module 3 focuses on professional management and caters to advanced professionals.

Starting on July 2024, interested participants can obtain further details at www.unige.ch/formcont/en/courses/cas-biobanking and contact casbiobanking@unige.ch for inquires. This comprehensive CAS in Biobanking serves as a valuable resource for individuals and institutions seeking harmonized knowledge and competencies in the dynamic field of biobanking, ensuring a progressive and tailored approach to skill development for diverse professionals.

Keywords

biobanking, training, sample management, quality standards, education, governance, processes

O13: Educational program for biobank personnel and interested persons

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Description

Successful banking and use of samples requires the collaborative effort of various stakeholders, e.g. patients, study nurses, biobank personnel (project management, informaticists, laboratory technicians etc.) as well as researchers. They all need to know different aspects of biobanking at different levels. The German Biobank Alliance has focused on how a comprehensive biobank educational program for different stakeholders could look like.

Surveys of German biobank staff were used to analyze their training needs and to develop content and learning methods tailored to different target groups. In addition, the usefulness of the tools was regularly evaluated to integrate improvements over time. The focus of the educational program was on biobank personnel, especially technicians, and students, the researchers of tomorrow.

The program has 4 pillars: 1) A combination of online preparation and hands-on technician training has been successfully implemented for DNA isolation from biospecimens. 2) A German adaptation of the Canadian edX course “Biospecimen research methods” serves as onboarding course for new biobank personnel and as an introduction to biobanking for students. 3) A webinar series on relevant biobanking topics has also been established. Experts from the GBA community contribute to webinars, which are recorded to enable timely self-paced learning.4) A biobank board game was developed. It has already been used in student courses and public outreach event for the lay public. The feedback we received from users has been very positive.

This educational program meets the needs of the German biobanking community. Other national nodes are encouraged to adopt (elements of) it.

Keywords

Education program; stakeholder; biobank education

O14: Starter kit: a practice-oriented guideline for the establishment and operation of professional biobanks

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Background

The establishment and expansion of professional biobanks is in rapid progress both nationally and internationally. Previously no national or European comprehensive, practice-oriented instructions particularly for the establishment and operation of biobanks exists. However, there is a high need and demand for such a compact guideline.

Material & Methods

Within the framework of the German Biobank Alliance (GBA), biobank experts work in topic-specific working groups with the aim of developing new generic biobank standards and tools. The Starter Kit working group has defined thematic blocks essential for the establishment and operation of biobanks and compiled currently relevant guidelines and practices recognized by the Biobank community in a compact form.

Results

The Starter Kit has been published on the GBA platform ((GBN/GBA eLearning on) OpenILIAS). The thematic blocks address governance & organization, financial management, human resources, ELSI, IT, laboratory & repository, quality management, communication, training & public relations, as well as study- and project management. Each block has the same structure: a topic-specific introduction, links to related GBN/GBA webinars and a checklist with further links and literature. GBA members have free access to the tool. In addition, academic biobanks outside GBA can be approved for admission.

Conclusion

Starter kit was initially developed as a practical guideline for the establishment and expansion of younger professional biobanks. It widely used and implemented in Germany. An adaptation for the Europe Biobank Society is planned in the near future.

Keywords

Biobank, Starter Kit, Education

O14b: Biobanking education: the project “SCIence outreach: The example of BIObanks in Europe” and its progression so far

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Introduction

Public involvement is essential in biobanking for medical research, as these endeavors greatly rely on public trust and citizen participation. To achieve these, raising public awareness and keeping the interest alive, biobanks with the aid of researchers and students need better community engagement strategies. The “SCIence outreach: The example of BIObanks in Europe” (SCIBIOEU) project, conducted by seven BBMRI-ERIC node partners in five

European countries (Austria, Cyprus, Finland, Greece, Italy), addresses this need.

Materials & methods

To map existing educational resources, online courses and serious games for students, young researchers and citizens on biobanking were searched and summarized in an inventory. In addition, focus groups involving the public and selected stakeholders (including students, professors, researchers, developers and designers, biobanking professionals), were conducted. An online course is engineered using Moodle, while the serious game is developed in Unity, with WebGL enabling online browser accessibility.

Results

The focus groups provided insights into target group's needs and important elements for the development of the course and serious game. An online multimedia course, incorporating texts, videos, and user self-evaluation mechanisms, is developed. Additionally, a 3-D online serious game that navigates users through a virtual biobank, is designed. This interactive experience is enhanced with the assistance of an AI service, providing multiple stages and customization options.

Discussion

SCIBIOEU, an ERASMUS+ funded initiative, seeks to bridge the communicative divide between scientists and the public about biobanking. It equips scientists with effective strategies for scientific outreach and boosts public knowledge of biobanking, through an online course and a serious game.

Keywords

biobanking, serious game, e-learning, gamification, public engagement, science outreach

T7. Information security and data privacy

O27: "Challenges for the Biobanking Community in Light of the New EU Digital Package of regulations, with a Focus on the European Health Data Space (EHDS)"

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O28: Donor first – how to foster usage of banked clinical samples by leveraging large language models and AI technologies to analyze Informed Consent Forms

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Description

Biobanks collecting residual biospecimen from clinical studies contribute to novel clinical research in companion diagnostics, clinical assay development and biomarker development for precision medicine. In a multi-centric clinical study these collections may potentially include different populations originating from various geographies. When an individual donor decides to participate, various ethical, cultural, legal regulations and site-specific policies may come into effect regarding the use of human biosamples for clinical research. The donor's agreement is usually obtained through Informed Consent Forms (ICF).

Consequently, informed consent collected in multi-centric studies may display a significant range of variations. This variability in consent poses compliance risks when it comes to broad reuse of residual samples obtained from these studies. Therefore, it is crucial to have an in-depth understanding of the diverse consent requirements. For instance, a larger phase III study including protocol and procedural amendments could easily encounter over 500 different consent variations, depending on site- and country-specific regulations.

Here we showcase a novel approach that harnesses the power of large language models (LLMS) and machine learning to extract key features for ensuring the compliant usage of clinical biosamples. By leveraging LLMs and advanced algorithms, we can accurately identify different types of consent and detect pertinent features that facilitate the reuse of clinical samples. Our analysis involves more than 2500 ICF documents collected from diverse clinical studies. The utilization of LLMs and machine learning techniques proves to be instrumental in generating insights and streamlining the extraction of consent-related information, all while ensuring regulatory compliance.

Keywords

ICF, Informed Consent Form, AI, Artificial Intelligence, LLM, Large language models

O29: The exchange of health data through the cloud. How to overcome the possible risks for patient

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Description

The use of new technologies, such as cloud computing, can be a very useful tool for storing and exchanging research information.

However, it can lead to reduced user control. This can be even more problematic when it comes to Universities, Hospitals, Research Institutions putting health data into the cloud.

The risks are greater when the cloud service is outsourced, as the Hospital, University or Research Infrastructures would not be exempt from the responsibility given to it by the European legislation on the personal data protection.

It is essential to analyze the risks for the security of patient privacy when researchers use the cloud paradigm as the latter is a technology that is increasingly used, also considering the lower costs compared to other data storage solutions, the elasticity and the scalability it typically allows.

An assessement of the measures that users must take to protect the data subject and comply with the obligations laid down in the accountability principle.

Keywords

New technologies, health data, risks, accountability principle, infrastructure management costs

T10. ELSI topics in biobanking - friend or foe

O38: Unlocking Potential, Addressing Concerns: Use of AI in Biobank-Based Research through the Lens of Research Ethics Committees

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Introduction

Recently biobanks are extensively involved in research using large volumes of data and applying AI for analysis. Introduction of new technologies in the field of biomedical research usually raises novel ethical issues and pose challenges to the process of ethical review. The presentation will analyse challenges posed to the ethics review process by employing AI in biobank-based research.

Materials & methods

The presentation is based on state-of-the-art literature analysis.

Results

The challenges posed by AI use in biobank-based research include epistemic challenges and their ethical implications, (e.g., ethical aspects of AI knowledge production, understanding of novel risks, potential of bias, technological reliability, epistemic justice, and transparency) and challenges of AI use for implementation of the research ethics principles (e.g., evaluation of risk/benefit ratio, protection of confidentiality and privacy, informed consent, ensuring public benefit and building trust).

Discussion

RECs members often lack knowledge and criteria for evaluating use of AI. Also, the traditional guidelines used by RECs currently do not sufficiently address the complexity of AI. Lack of criteria, guidelines and AI expertise in the review process might lead to delays to the progress of research.

Conclusion

Some of the recommendations for facing the new challenges include strengthening RECs capacities by training in AI ethics, involving external experts, and building a dialogue with

researchers using AI tools for biobank-based research.

Keywords

research ethics; research integrity; trustworthy AI

O39: Informed Consent for Minors – Empowering the Future of Paediatric Research

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Introduction

Since 2007 Biobank Graz - a partner of BBMRI.at - has been collecting informed consents from adult patients at the University Clinic Graz. This allows researchers to use samples acquired during routine processes in many different projects concerning therapies, medication etc. after ethical approval. Nevertheless minors have not been included in the consent form, which limited the usability of their samples. To address this, Biobank Graz collaborated with paediatric clinicians to implement a consent for patients aged 0 – 17 years allowing the collection of their samples and fostering research in this crucial area.

Material and Methods

Biobank Graz engaged in discussions with paediatric clinicians and the ethics committee of the Medical University of Graz to understand their needs regarding an informed consent for minors. Additionally, an info clip was developed to simplify the medical briefing for children explaining the concept of biobanking in an easy and entertaining way.

Results

In close cooperation with the paediatric clinicians and the ethics committee, Biobank Graz implemented three informed consents adapted to the different age groups (0 -7, 8-13, 14-17 years). Reconsenting is only required once patients reach legal age. The info clip explaining biobanking for minors is available on Biobank Graz's website and is shown to minors during their medical briefing.

Discussion/Conclusion

The implementation of the Biobank informed consent for minors allows the utilization of valuable samples not only in the scope of the study specific consent but also for future research questions. Biobank Graz contributes to advance the future of health research in the paediatric field.

Keywords

informed consent for minors, paediatric research, biobanking

O40: Hands on old collections: the biobank as a fair vector of integration in the scientific ecosystem

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O41: Navigating New Waters: Implications and Challenges for Biobanks in the European Health Data Space

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Introduction

The European Health Data Space Regulation (EHDS) aims to increase the control of individuals over their electronic health data, while also making more data available for healthcare and scientific research. The law introduces new roles and responsibilities, most notably the concepts of health data access bodies (they will be responsible for authorizing access to data), data holders (they have the right or obligation to make data available), and data users (they can have access to data). However, the integration of biobanks as data holders within this framework introduces a complex layer of consideration, given the current heterogeneity of biobank regulations across Member States.

Material & methods

Legal desk research.

Results

Our presentation will unfold in four key sections: the overview of the EHDS, a comparison of biobank regulations across Member States, the potential benefits of EHDS for biobanking (such as promoting standardization in data access and reuse, thereby encouraging cross-border research), and an exploration of the possible risks (for instance, uncertainty regarding how the EHDS framework will align with existing research oversight regulations in Member States, particularly concerning ethical review processes and civil society involvement in governance).

Discussion & conclusion

Uncertainties remain regarding the interaction between the EHDS provisions and current national biobank regulations. Careful consideration is required to avoid further fragmentation of the European biobank framework. For biobanks, the transition into this new regulatory landscape offers both opportunities and challenges, which our contribution aims to untangle.

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Keywords

EHDS, Health data space, Regulatory alignment, fragmentation, cross-border research

T14. “Be aware of biobanking” – outreach and communication in and for biobanks

O53: A Decade of Outreach: Communication Strategies tailored to Different Stakeholders - Best Practices of the German Biobank Node

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O54: Communication – a tool for awareness and implementation of a new Biobank Act

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O55: Danish patients are willing to contribute to genetic research - knowledge gained from the Copenhagen Hospital Biobank.

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Introduction

Since 2009, excess biological material used for blood type and screen testing in the Capital Region of Denmark, has been preserved in the Copenhagen Hospital Biobank for future research purposes.

While the storage of samples is not consented, patients are informed that remaining blood or tissue samples may be stored in biobanks for future research. Additionally, patients are informed about the general opt-out possibility, allowing them to exclude their samples from research use through the National Database of Non-Consent to the Use of Tissue Samples for Scientific Purposes.

Material & methods

To raise awareness of our sample storage, we informed in 2020 all patients with a sample in the biobank by electronic or traditional mail. Subsequently, we inform newly enrolled patients every six months. In 2020 we established a call center and built a new website (www.regionh.dk/biobank) to address patient inquiries.

Results

Presently, the information has been distributed to 368.999 patients.
Merely, 0.5% requested the destruction of their sample, 0.1% provided positive feedback and 0.1% had more detailed questions.

Discussion

Only 0.5% of the contacted patients opted not to take part in the biobank, highlighting the

supportive and willing participation of Danish patients in research.

Keywords

Biobank, information, communication, call center

O56: Bringing biobanking and pathology closer to children and adults

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Introduction

The number of biobanks is increasing, accompanied by growing awareness among researchers. However, public knowledge about biobanks, at least in Austria, is still low. Therefore, BBMRI.at partners actively engage in public events, such as the Austria-wide Long Night of Sciences, regional initiatives for students/pupils, open-house-events, and biobank tours.

Material & methods

BBMRI.at partner Medical University of Graz developed a concept for “biobanking and pathology workshops” for citizens - particularly for children. Aim is to familiarize them with the terms biobank and pathology and make them tangible in hands-on parts where they “become” biobankers and pathologists.

Results/findings

The concept consists of

- an interactive introduction about: i) the workflow in a university-hospital (patient anamnesis, surgery, pathology, biobank, research leading to the development of new therapies) and ii) the organs of our body and their functions.
- a practical, hands-on part where children and adults can perform classical work steps of a biobank and pathology laboratory in different stations. This includes pipetting blood (raspberry juice), freezing it in a ‘biobank’, snap-freezing tissue (sausage or chicken liver) in liquid nitrogen, embedding tissue in paraffin, staining paraffin (mouse) tissue sections from different organs and viewing them under microscope.

During public events this is accompanied by guided biobank tours offered by Biobank Graz.

Conclusion

The workshops are an excellent engagement model for citizens and for paediatric patients to introduce biobanking and pathology. They are a suitable for different age groups, event settings and durations, and are a pleasure for children, teachers, parents and involved staff.

Keywords

pubic engagement, education & outreach, biobanking, children, paediatrics

T18. Partnerships towards real world data – opportunities, needs and hurdles

O68: European Open Science Cloud

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O69: Biobanks samples catalogue: challenges and opportunities

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Introduction

One challenge of researchers at public institutions or private biotech’s is the difficulty to find biosamples. Most biorepositories have the information of their samples mainly available by biobank personnel who perform searches using their Laboratory Management Systems (LIMS). Lately there have been efforts to make more accessible this information through the development of catalogues

available through biobanks or biobank network webpages (BBMRI, Plataforma de Biobancos (Spain)).

Methods

Our biobank repository was initially analyzed to determine completeness, integrity and standardization of data. A plan was designed to improve all these parameters and then the first version of our biobank catalogue was created.

Results

THE HUVH Biobank is constituted by almost 70.000 donations recruited by clinicians at different hospital services and with registries of different complexity and accessibility. The clinicians collaboration and access to hospital registries was needed to collect data. Our LIMS was changed to include a minimum dataset for diagnostic and protocols for validation of the data were implemented.

The first version of our Biobank catalogues was created containing 65.609 donations, of them 4,81% are rare diseases, 67% are immune-mediated diseases, 6% are infectious diseases, 11% are COVID-19 and 8% are oncological diseases. The COVID-19 and rare diseases catalogue has been integrated within the *Plataforma de Biobancos* catalogue. Both catalogues are searchable catalogues that allow users to query based on some parameters.

Conclusion

Biorepositories sample accessibility through catalogues is a useful tool that requires centralization of information, data protection measures, data standardization and verification and multidisciplinary collaboration.

Keywords

data standard; data quality; catalogue

O70: Collaborative Efforts in Developing a Future-proof Biobank Information Management System for Pediatric Oncology: a FAIRy tale from Hungary

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Introduction

The growing need for a substantial pool of high-quality and multimodal FAIR data in paediatric diseases necessitates collaborative partnerships to jointly overcome many obstacles.

Methods

Our extensive software development initiative united eight Hungarian pediatric oncology clinical centers and several IT service providers in one interdisciplinary alliance to build a prototype of a next-generation biobank information management system (BIMS).

Results

Our web-based BIMS 1.0 version features a suite of specialized modules and functionalities for ethical and interoperable end-to-end biobanking while following standardised data models. Tracking samples and aliquots through their entire life-cycle, managing transfer logistics through the whole chain-of-custody, and fit-to-purpose distribution control are ensured separately. A unique hallmark is a double integration with the Hungarian Pediatric Cancer Registry, and the database application of the Central Diagnostic Facility, allowing for automated synchronization of donor clinical and molecular genomic data. Additionally, a privacy-preserving data-sharing technology is implemented in the BIMS, enabling the generation of encrypted output datasets that are ready for secure distribution to requesting researchers worldwide.

Conclusion

We aim to set an example of bringing great minds of IT-experts, biobank specialists, and healthcare professionals together to design sustainable innovations for biobanking. We are currently working on the 2.0 version, which will feature additional unique characteristics and full audit support. Our solution demonstrates that even a small biobank can contribute substantially to advancing paediatric cancer research by providing high-quality biospecimens and detailed, yet well-structured FAIR datasets.

Keywords

paediatric biobanking, BIMS, FAIR data sharing, innovative partnerships

O71: Clinical, Biological, Imaging and Genetic Repository (C-BIG), An Integrated Approach to Biobanking in the Context of Open Science

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Description

The Montreal Neurological Institute's Clinical, Biological, Imaging and Genetic (C-BIG) collection and patient registry has recruited almost 4000 participants across Canada with neurological conditions as well as healthy controls since 2016. Using the web-based LORIS open source database, the open biobank integrates patient and sample data to help scientists around the world to run cutting-edge research projects within an Open Science framework, to advance the understanding of neurological diseases and discover new therapeutic ways to help fight neurological disorders.
The main objective is to collect de-identified biological material as well as clinical, imaging and genetic information from patients and controls to enable innovative research projects that will advance the understanding of neurological diseases and human health under the Open Science principles. So far, C-BIG has collaborated with more than 90 academic and industrial partners. Each partner has to give a summary data report on the use of samples within an adjustable period of time if no publication has arise.

The Neuro's C-BIG repository is using an open version of LORIS (Open Portal) for data access, including a Data Query Tool for scientists. 3 different levels of access are available: Open, Registered and Controlled.

The C-BIG Repository hopes to improve and facilitate the material and data collection and sharing under the Open Science principles. The long-term goal is to reinforce the recruitment of participants across the world, by integrating the most information possible of these participants in the multimodal database to have a broader picture of neurological disorders, and accelerating research.

Keywords

biobanking, neuroscience, Open Science, open sharing, multimodal, data basing

O72: Cost recovery model in biobanking – from theory to practice. Success stories of model implementation in three EU funded projects.

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Description

The model of cost reimbursement for providing human biological samples and/or data is a difficult task to implement as it should depict all activities and elements affecting the exact cost of human biological samples or a data set preparation.

Many biobanks globally collect, process, store, and distribute human biological samples from healthy donors and patients suffering from civilization and rare disease. Although all biobanks are working towards improving and widening the access to biological samples, there exists no generally accepted cost recovery model for sample/service.

We have adopted previously published calculation scheme developed by the international expert group from six EU countries, to evaluate costs for collections of various types of biological samples (Clemont et.al, Sci Trans Med, 2014). The cost recovery model includes calculation of all costs of collection, preparation and storage of clinical samples/data and largely depends on the level of advancement of the collection. It can be also used to calculate the cost of scientific service provided by the expert centre, when performing analysis on samples released from biobank. The model is divided into several blocks allowing each biobank to choose elements illustrating their involvement in sample preparation, data collection or analysis.

Our adopted model has been implemented in three European projects: HE ISIDORe, HE CanSERV and H2020: CY-biobank. We have successfully used it to estimate costs by

biobanks participating in those projects and releasing samples for non-commercial partnership.

Keywords

sample cost recovery model, sustainability, economics

T22. Green biobanking and sustainability

O86: BIOSEK - A cost-effective freezer facilities provided free of charge for researchers within the Copenhagen Hospital Region

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Description

Scientists within the Copenhagen Hospital Region, like many others, encounter challenges in securing cost-effective, secure, and accessible storage solutions for their biological specimens. These research samples are frequently distributed across various smaller and larger biobanks, held by individual researchers. These biobanks contain valuable samples useful for general research and personalized medicine studies. However, local storage limits the optimal utilization of these resources.

As a service for researchers and to promote research collaboration, the Copenhagen Hospital Region has established joint freezer facilities (BIOSEK). The aim is to gather regional biobanks into a few regional freezer farms, offering safe and free storage and creating a better overview of the biological material available for research. Consequently, the region benefits from the sustainability achieved by creating these designated freezer locations with large scale operation, where energy efficient water-cooling systems can be established.

To ensure the quality and visibility of all biobanks and samples stored in BIOSEK, researchers must undergo an application process and provide essential data connected to the samples when submitting them for storage. Currently the storage capacity in BIOSEK comprise of three freezer farms with a total of 136 -80°C freezers and one -20°C freezer robot, located throughout the Region. In total, the storage capacity in BIOSEK currently is about 8,5 million samples, but the demand is still rising and the need for expansion is on-going.

Keywords

sustainability

O87: A Generic Concept for Biobank Services Reimbursement

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Introduction

The ability to reimburse costs for biobank-services provided represents a cornerstone of biobank sustainability.

Material and Methods

A generic cost compensation model has been developed within the German Biobank Community that consists of a set of basic services and consumables required to register, transport, process, and store biosamples within a clinical setting. The model assumes that each biosample type is processed according to a standard and well-defined workflow, which is characterized by a subset of basic services and/or consumables. Both, services and consumables may be scaled within the process to account for increased efforts or multiple consumables, respectively. Finally, four different rates specify the level at which the fee for each process is calculated (Budget funds, public third party funding internal/external, and industry partners). Further options exclude fees for specific services from being accounted or consumable fees from being scaled among different rates.

Results

As a proof-of-concept, we evaluated the methodology in a spreadsheet format regarding its flexibility and generic usability. Individual biosample fees are calculated by just counting the number of primary samples and their corresponding aliquots and multiplying them by the primary component fee and the aliquot component fee, respectively. Internal researchers are now able to estimate biosample processing and storage costs for upcoming projects via a web-based Sample Cost Estimator.

Conclusion

The here developed generic model for biobank reimbursement is a generic and robust tool to reimburse biobank services for research projects. Next, we plan to incorporate the model in our Biobank Information Management System.

Keywords

Sustainability, Reimbursement, Biobank Services

O88: Construction of a sustainable biorepository

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Description

Presenting the state of the art fully automated DZNE LN2 Biorepository: an overview from the first concept to the completion of the building.

The main focus of the project was to adhere as closely as possible to sustainability guidelines while offering a tailor-made home for a large-scale automated sample storage setup. In this presentation, it will be shown how crucial it is to construct the building around the selected sample storage system instead of shoehorning a system into a preexisting construction object.

Examples of sustainability can be found in the airflow-, the electric-, the heating & cooling-system as well as the building structure itself. The decision of a biobanking system has strong influence on the building systems. To fulfill additionally all government requirements results in an useful structure with energy saving walls, collecting generated heat and guiding it back into the system while reducing the cooling energy levels. The incoming energy sources are topped off with geothermal energy coupled to highly modern reversible heat pumps. The necessary power is mostly supplied by a photovoltaic system. This low-energy building offers itself as an exemplary model for optimizing infrastructural operating costs in other similar projects.

Keywords

Sustainability, construction, building, biorepository, biobanking, automation, energy saving

O89: Visualising biobank costs

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Introduction

Within the BBMRI.be sustainability working group the Belgian member biobanks, all academic or hospital associated, developed a tool that visualises biobank associated costs enabling their reporting to researchers and institutional management.

Methodology

Costs were grouped into categories such as storage, sample management system, staff etc. For each category, an extensive list of associated costs was developed, aimed at identifying hidden costs. As an example, storage not only comes with the costs of the storage unit, but also includes the content of the storage unit, its monitoring system, its power supply, a back-up system, insurance, etc. All these costs for each of the categories were included in an intuitive calculator tool. Inclusion of the number of projects and the number of samples allows to calculate an average resource cost. The tool is currently under validation by the involved biobanks.

Conclusions

Many biobank associated costs are easily overlooked. Their inclusion in a calculator tool on the one hand allows to optimise cost recovery from researchers while on the other hand allows optimised reporting of total costs to institutional management. Whether a certain cost category is included in the calculation depends on internal biobank politics as well as to what extend costs are financed institutionally. In the latter case, within our calculator tool, biobanks can easily opt not to include such costs when calculating cost recovery costs for internal researchers. Overall, our calculator makes biobanking-related costs more visible and transparent and allows for more sustainable biobanking.

Keywords

sustainability, calculator

O90: Novel Dry Storage Approach for DNA Preservation at Room Temperature

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Description

In the era of precision medicine, accurate and standardize assays are needed for high quality NGS data. Pre-analytical variations such as collection, processing, storage and transportation need to be considered to minimize their influence on the outcome data. Current gold-standard method for DNA preservation is -80ºC which implies high maintenance costs, large spaces, constant energy supply and safety measures for Biobanks.

We evaluated quality and suitability of DNA processed and stored with an innovative sustainable solution based on precision freeze-drying of DNA samples. We stored purified DNA in parallel under 3 conditions: -80C; dried and stored at 22C; dried and stored at 60C. We evaluated integrity and purity of DNA samples during 18 months of real storage using TapeStation, Long Multiplex PCR and absorbance ratios.

21 years of RT-equivalent storage did not significantly affect the DNA integrity and the detection of the 17,5Kb band in multiplex Long PCR (Fig.1). Additionally, Whole Exome Sequencing of the RT storage samples showed a recall of 99% in all time points analyzed, when compared with the -80C gold-standard (Fig.2). Finally, a Cytoscan 750K analysis of DNA samples just after its extraction and at the end of the storage period at different conditions revealed the same alterations, showing same loss or gain patterns observed at extraction (Fig.3).

Based on these results, the technology here evaluated demonstrated to preserve DNA quality, allowing its use with high demanding genomic methods. A standardized methodology like this may enormously contribute to the safety and sustainability of long-term sample storage.

Keywords

long-term, freeze drying, DNA, sustainability, integrity

O91: Winning the Freezer Challenge Clinical Biorepository Award 2023

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Introduction

Long-term storage of materials for biomedical research at ultra-low temperature (ULT) significantly impacts the environment due to high energy consumption. **T**he International Laboratory Freezer Challenge, an initiative of My Green Lab and the International Institute for Sustainable Laboratories (I2SL), aims at increasing **energy efficiency in cold storage**. The Biobank core facility of Amsterdam University Medical Center, which owns about 150 ULT freezers for the storage of biobank and study collections, participated in the Freezer Challenge 2023.

Methods

We focused on temperature tuning and moved the set point of 128 ULT freezers to -70°C instead

of -80°C. We assured ourselves of support from the board of directors and started the project as pilot. We did not justify the -70°C temperature but asked researchers to motivate eventual exceptions from the new set point.

Results

Within 6 months, we neither met resistance and nor did we face problems in relation to the shorter response time related to the adjusted temperature. Consequently, this temperature adjustment has been adopted as a standard at Amsterdam UMC. By saving 650,576 kWh over the year, we won the Clinical Biorepository Award of the Freezer Challenge competition 2023.

Conclusion

The participation of Amsterdam UMC Biobank in the Freezer Challenge 2023 illustrates how institutional biobanks can use their potential to efficiently reduce the carbon footprint of health research. It serves as a model for other institutions, showing that targeted freezer management changes can offer substantial environmental benefits without compromising research.

Keywords

ultra-low temperature freezers, energy saving, freezer challenge

TRACK 4: Insights into Healthcare: Patient Engagement, Rare Diseases, and Organisational Profiles

T4. Organisational profiles

O15: Introducing the EvolveBBMRI project – Accelerating datafication for support of EU health priorities, greening of biobanks and integrated Research Infrastructures' approach to "One Health"

Prof. Jens Habermann (1), **Jana Pavlic-Zupanc** (presenter) (1)
(1) BBMRI-ERIC

Introduction

Since its creation in 2013, the Biobanking and BioMolecular resources Research Infrastructure - European Research Infrastructure Consortium (BBMRI-ERIC) has become one of the largest RI for health and life science research in Europe. BBMRI-ERIC is now facing a unique moment to develop its strategy for the next decade. This presentation will introduce the EvolveBBMRI project, aiming at further developing BBMRI-ERIC for the benefit of its scientific communities, increase its capacity to serve EU priorities and deepen industry collaboration.

Material / methods

Having started in 2024, the EvolveBBMRI project will run for three years, including BBMRI ERIC and 10 National Nodes. EvolveBBMRI is structured in four pillars: 1) Acceleration of datafication of biobanks and biomolecular resources to enable reproducible advanced medical research in support of EU health priorities; 2) Teaming with industry towards greener biobanking; 3) Long-term sustainability measures addressing RI landscape gaps; 4) Strengthened approach for career paths, training and outreach activities to maximise impact.

Results

The project will ensure implementation of specific results on six key target groups: a) scientific community; b) National Nodes, biobanks in Europe/beyond; c) industry/private foundations; d) national/European decision-makers; e) European/international networks; f) donors, patients/citizens.

Discussion

EvolveBBMRI activities will strengthen the European research infrastructure landscape by focusing on collaborative "One Health" approach to health challenges across different Research Infrastructures. Ultimately, the project will contribute to increased scientific excellence to better tackle scientific and societal challenges and to foster the role of biobanks in the future of healthcare research.

Keywords

One Health, Sustainability, Health Research, Biobanks, Greening, Research Infrastructures

O16. Biobank network in Japan to accelerate progress towards genomic medicine.

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Description

In Japan, the Government has drawn up a health and medical strategy for genomic medicine as the national growth strategy, and research and development to realize genomic medicine including drug development utilizing biobanks is being promoted.

A programme for the utilization of biobanks called B-cure (Biobank - Construction and Utilization biobank for genomic medicine REalization) is currently underway to realize genomic medicine. In the B-cure programme, we developed a network of 14 major biobanks and a system for utilizing biobank samples and data that will facilitate research and development of genomic medicine in Japan.

We then developed a biobank cross search system on biospecimen and data stored in our biobank network. Research institutions can find biospecimens and data they need for their own genomic medicine research and development. To implement a cross-search service for biospecimens and data, we standardized the metadata of biospecimen and data.

Now our biobank network stores 1,420,000 biospecimen and 320,000 molecular data provided by 540,000 donors. We also setup the web-based coordination system of fast access to biospecimen and data to meet the requests by academic/commercial users using the biobank cross-search system. Our biobank network provides essential infrastructures for different types of health research including basice medical science to accelerate progress towards personalized medicine.

We are addressing drawing up an ecosystem of stakeholders to realize personalized medicine and discussing how to maintain and develop this infrastructure along with the health and health care strategy in Japan.

Keywords

Biobank network, Japan, infrastracture, stakeholder, ecosystem

O17: Strengthening the Italian National node of BBMRI, BBMRI.it

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Description

BBMRI.it is a distributed infrastructure including 97 biobanks located throughout Italy in 19 universities, 33 research hospitals, and 45 hospitals.

BBMRI.it was awarded the Strengthening BBMRI.it project through a competitive NextGenerationEU competitive call for grants launched by the Ministry of Research for the improvement and digitalisation of the national nodes of the ESFRI RIs.

The project focuses on four main topics. The first topic concerns the strengthening of the infrastructure backbone: fourth-generation biobanks of organoids, microbiomes, iPSCs and extracellular vesicle biofluids, and digital imaging biobanks will be created, with an important focus on biobank ISO accreditation and the implementation of contingency plans for sustainability. In addition, a network of Core Facilities offering -omics services will strengthen the portfolio of the RI biobanks. The 2nd topic will reinforce the BBMRI infrastructure Common Services (CS) and enhance the provision of quality, IT and ELSI services with the development of a Digital Informed Consent and introducing Digital Common Conditions of Use to all biobanks. The 3rd topic will address the implementation of Open Science and Data: the project is implementing a service catalogue marketplace and a service for FAIRificating of the current datasets, to improve standardization and accessibility of the sample data. The last topic addresses issues regarding access and use cases.

In addition, a variety of training opportunities will be offered to young generation of biobankers and quality managers who will promote biobanking in Italy and abroad for the years to come.

Keywords

fourth-generation biobanks, organoids, microbiomes, iPSCs, digital imaging biobanks, Core Facility, CS IT, CS ELSI, Digital Common Conditions of Use, Open science, FAIR data, standardisation, training, italy, bbmri.it

O18: Fostering genetic studies in Ukraine by developing biobanking network: challenges and future perspectives

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Biobanks (2) Ukraine Association of Biobank, (3) Ministry of Health of Ukraine, (4) Bogomolets National Medical University, (5) Ukrainian Association of Research Biobanks, (6) Medical Laboratory CSD, (7) Bogomolets National Medical University

Introduction

Biobanks are essential in advancing precision medicine, discovering novel biomarkers and new treatment strategies. Ukrainian biobanking faces multiple obstacles limiting the progress of genetic research. This study assessed the existing barriers and defined the future perspectives of biobanking in Ukraine.

Materials

The analysis of national policies on research with biosamples and existing biobank activities was performed.

Results

Few existing documents regulate biobank operations. One of them is the resolution of the Cabinet of Ministers of Ukraine on licensing conditions for Banks of umbilical cord blood, other human tissues and cells. However, no national policies exist for clinical data collection, sample handling, use, and sharing. The lack of educational programs on biobanking defines the shortage of qualified staff and low biobank involvement in fostering genomic medicine. In 2021, the Ministry of Health (MOH) of Ukraine called for the development of national regulations on biobanking, addressing core ethical issues, including respecting human rights, protecting participants, transparency and accountability. Despite the ongoing war, the policy was developed and circulated among experts in the field for revision, improvement, and further approval. MOH, the Ukraine Association of Biobanks, and the Ukrainian Association of Research Biobanks have joined efforts to build a network of private and academic biobanks, meeting the stakeholders` needs in education and standardization of biobanking for fostering research infrastructure and precision medicine in Ukraine.

Conclusion

Building a national network of biobanks in Ukraine and approving national policies on biobanking are vital to strengthening education and research infrastructure for fostering genomic medicine in Ukraine

Keywords

biobank, genomics, policy, precision medicine, research infrastructure

T11. Rare diseases

O42: Boosting rare disease research capacity through national and international networking

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O43: Mitochondrial Complex 1 deficiency; a case study

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Introduction

A Maltese girl born to healthy, non-consanguineous Maltese parents was suspected of having a Complex I respiratory chain mitochondrial disorder based on a muscle biopsy. She developed: gradual motor regression; nystagmus; dystonia; loss of sitting balance, spasms, facial dyskinesia, jaw stiffness, mouth opening, a global developmental delay, bilateral hip dysplasia. Died under 10 years.

Methods

The whole exome, mitogenome and the full transcriptome were analysed. The effect of a rare SNV (c.308C>T, rs749249430, MAF=0) in NADH:ubiquinone oxidoreductase complex assembly factor 3 (NDUFAF3) on mitochondrial Complex I protein expression was investigated in HEK293 cells with an ELISA assay for Human NADH dehydrogenase (CI). The differential expression of NDUFAF3 in transfected cells was measured. A 3D protein model was visualised to understand the structural consequence of the c.308C>T mutation on NDUFAF3.

Results

WES revealed the patient inherited mutations in 4 nuclear genes which cause mitochondrial CI deficiency (mtCID): NDUFS1, NDUFA10, NDUFAF3 and NDUFB9, the last predicted to be pathogenic in silico. 3 mitochondrial mutations that cause Leigh syndrome were also found. 7 transcription factor binding sites and 2 enhancer gene targets were differentially expressed. NDUFAF3 was up-regulated in both mutant and wild-type transcripts. There was no statistically significant difference in the mean concentration of NADH dehydrogenase between the mutant and wild type NDUFAF3 vectors in HEK293 cells, p = 0.62. Protein modelling showed a (p.Ser103Phe) change did not affect the structure of NDUFAF3.

Conclusion

This biobank-led research showed the complex genetic heterogeneity of mitochondrial diseases which involve both nuclear and mitochondrial genes.

Keywords

mitochondrial diseases, omics, protein expression, protein modelling

O44: Swiss AutoImmune Liver Disease (AILD) Biobank

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Introduction

In 2017, the Swiss Autoimmune Liver Disease Cohort Study was initiated, representing a nationwide registry of Autoimmune Hepatitis (AIH), Primary Sclerosing Cholangitis (PSC), and Primary Biliary Cholangitis (PBC). The primary objective is to gather high-quality retrospective and prospective data on these rare diseases to elucidate their epidemiology, natural history, local treatment practices, response to treatment, and outcomes. The registry includes a biobank, enabling basic and translational local or collaborative research projects.

Material & Methods

This multi-center project involves 21 centers across Switzerland, including 9 paediatric units. Clinical data are collected at baseline and scheduled annual follow-ups. Blood samples are collected for biobanking once a year and stored centrally.

Results

The study has enrolled 762 patients, comprising 334 (40%) AIH, 371 (45%) with PBC, and 124 (15%) with PSC . Sixty-seven patients (8.8%) were diagnosed with AIH and either PBC or PSC (AIH variant syndromes). Thirty-four paediatric patients were included. A total of 4977 serum samples, 1580 mRNA samples (PAXgene tubes), and 122 peripheral blood mononuclear cell samples have been collected. Several collaborative nested projects using data and/or biosamples have been carried out since 2017.

Discussion and Conclusion

Swiss paediatric and adult hepatologist are interested in joining the Swiss Autoimmune Liver Disease Cohort Study, which provides researchers with valuable resources to advance knowledge in the field of liver autoimmunity in a collaborative way.

Keywords

Autoimmune Liver Disease; rare disease; biobank; cohort

T15. Citizen & Patient Involvement: Emerging best practices?

O57: Involvement in biobank - rare disease patient experience and perspective in Latvia. Patient perspective

Sinica, Sanita (1)
(1) Patient representative

Description

Presentation will contain information about people involving methods in biobank, especially for patients of rare diseases in Latvia. What kind of challenges and perspectives there are for rare disease patients and researches in Latvia. Personal experience story of participation in the Latvian Population Genome Reference Project.

Problem – how to get enough rare disease patient involvement in biobank to provide quality researches for rare diseases. Why patients don't participate – no trustable information or fair?

Survey for rare disease patients will be presented (data from Latvian Alliance of Rare Diseases patient organisations and other rare diseases patients in Latvia). Also existing experience of Latvian Biomedical Research and Study Center including rare disease patients in biobank and researches.

The main contribution – the role of patient organizations (NGO) to improve involvement in biobank and researches. Patient organization – the bridge between science, government and patient.

Discussion and conclusion – how can we share success stories with other countries. What can be next steps to involve patient organisations to biobank development? What we still don't know about biobank and precision medicine or – what people are afraid of? Answers from patient perspective.

Keywords

Biobank, rare diseases, patient organisations

O58: Better collaboration, user research and co-creation with patients

Clareborn, Anna (1), Riggare, Sara (1)
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Description

Samka! is a Swedish project funded by Vinnova and managed by Uppsala University in partnership with Genomic Medicine Sweden (GMS), the Network against Cancer, the National Association for Rare Diseases, the Swedish Brain Tumor Association, the Forum for Emerging Patients, and Combined. The initiative aims to help create the right conditions for both patient and next of kin representatives to contribute to healthcare and medical research, in order to achieve the best possible health and well-being over time.

The Samka! project helps individual researchers, institutions, and others achieve this by actively working with six central recommendations for collaboration. The recommendations are based on the report *Patient and next-of-kin collaboration for better research and healthcare* from 2023. The report builds on nearly 8000 scientific articles evaluated through a collaborative effort by a number of Swedish life science and patient organisations.

This presentation will address the recommendations, but also give practical examples of how they have been applied, with a particular focus on how they have allowed patient and next of kin representatives to give input on everything from AI ethics to educational content, and what the outcomes have been so far.

Keywords

collaboration, PPI, co-creation, user research, patients

O59: National Patient Participation within Biobank Sweden

Marija Armus (1), Linda Lindskog (1), Alexander Hertzberg (1), Sonja Eaker Fält (1)
(1) Biobank Sweden

Introduction

The Swedish Biobank Act is recognized globally for its strong protection of citizen integrity as sample donors. To support sustainable biobanking and compliance to laws and regulations, Biobank Sweden (a national biobanking infrastructure) is starting a new initiative to increase patient involvement within the network. The initiative emphasizes the importance of health literacy through proper and distinct information and communication.

Methods

The initiative involves comprehensive mapping exercises and explores various forms of collaboration between stakeholders. The conclusions in the report “Collaboration 2.0: Sustainable collaboration for value and innovation” (2023) will be used as guidance. An initial phase includes the involvement of the European Patients´ Academy on Therapeutic Innovation (EUPATI), development of patient-oriented information materials and engagement with patient representatives and organizations.

Results

Dialog meetings and workshops will be conducted to identify relevant content for education and communication. Patient representatives will be actively involved in reviewing materials to ensure accessibility and relevance. A new education program will be integrated into a digital learning platform and launched for healthcare, academia, industry and patient organizations. The work also aims to include the patient perspective in research on individuals unable to make decisions for themselves at the point of care and improve processes for better patient-centered outcomes.

Discussion

By actively involving patients in the biobanking segment, Biobank Sweden aims to increase awareness and understanding of the significance of human biological material in research and the development of healthcare. Patient participation and patients´ health literacy is vital for ethical and sustainable biobanking practices and improved patient outcomes.

Reference list:

Claerborn, A., Degsell, E., Kannisto, K., Haag, M., Friedman, M., Riggare, S., & Juran, S. (2023). *Patient and next-of-kin collaboration for better research and healthcare. Collaboration 2.0: Sustainable collaboration for value and innovation* (Report Samverkan 2.0). Riksförbundet Sällsynta diagnoser, Nätverket mot cancer, Regionalt cancercentrum Stockholm-Gotland, Forum spetspatient, Biobank Sverige, Genomic Medicine Sweden och ATMP Sweden. Patient and next-of-

kin collaboration for better research and healthcare (biobanksverige.se)

Keywords

Biobanking, Health literacy, Patient-centered outcomes, Patient Involvement, Patient Participation

T19. Pitch your innovative idea

O73: MSHC: An innovative top-down multi-omics spatial technology with single cell resolution to unlock biomolecular information from FFPE samples archived in biobanks

Verhaert, Peter (1)
(1) ProteoFormiX

Description

Mass Spectrometry Histochemistry (MSHC) is developed as a spatial top-down mass spectrometry platform technology that takes molecular images of histological sections from FFPE samples, such as those archived in tissue biobanks.

MSHC makes the millions of pathologically well-documented wax-embedded tissues archived in human patient and donor biobanks accessible for molecular imaging.
MSHC employs atmospheric pressure MALDI in combination with high resolution (HR)MS (orbitrap) technology to detect metabolites and small (endogenous) peptides, making it an ideal translational biomarker discovery platform in human diseases, without the need for pre-clinical disease models.

Our mission is to compile the "Tissue Atlas of the *Homo sapiens* body (healthy & diseased)" which overlays classical histological images as those displayed in traditional light microscopy-based histology textbooks with biomolecular information obtained with MSHC of FFPE samples. Thanks to the Human Protein Atlas Initiative (www.proteinatlas.org), we are well on our way on this big endeavour.

Our human MSHC database is steadily growing which we expect to become an invaluable tool for pathologists or anybody studying specific human diseases in the not too far away future.

The Figure shows a histological section (5 micrometer thickness) through FFPE human pituitary adenoma using HRMS (ThermoFisher Scientific Exploris 480, equipped with an AP/MALDI UHR source (MassTech)), illustrating multiomics MSHC imaging. Pixel size is 5x5 um. Different colours represent different peptides and metabolites.

Keywords

Top-down mass spectrometry imaging; peptides; metabolites; multi-omics; FFPE samples; Human biobanks

O74: Empowering Genetic Counseling in Biobanks: The Rise of Chatbots and the Case of Alva

Van der Graaf, Fennie (1), **Loudini, Naila** (1), Marinelli, Giuseppe (1)
(1) Aevai Health

Description

This abstract presents a literature review conducted to investigate the use of chatbots for genetic counseling in the context of biobank participants. It highlights the main challenges, limitations, and potential advantages of genetic counseling chatbots and introduces our novel chatbot, Alva, which employs advanced techniques for structured questioning and for accurate genetic counselling.

A comprehensive literature review was conducted from 2018 to 2023 using Google Scholar. The search yielded 15 relevant articles to the topic of genetics chatbots, however none specifically pertained to biobanks. The search was expanded to include industry use cases of genetic chatbots in biobanking, and 2 use cases in the United States were discovered.

The 15 articles reviewed highlight genetic chatbots, like in the IMPACT-FH study and Mayo Clinic Biobank, as scalable tools for genetic counseling, enhancing family discussions on genetic risks. These bots assist in communication, scheduling, and counseling delivery, addressing both negative and actionable genetic findings. Critiques exist regarding their accuracy and empathy. Alva addresses these through a hybrid design, combining rule-based questioning with privacy-

preserving LLMs for natural responses, and using a retrieval augmented generation approach for up-to-date accuracy.

Genetic chatbots hold significant promise in genetic counseling within population biobanks, contributing to research and understanding in diverse populations. However, ethical, regulatory, and evaluative considerations must be addressed for their responsible integration. More literature is needed to further explore the possible applications of genetic counselling chatbots for biobank participants.

Keywords

Genetic counseling, biobanks, chatbots, large language models (LLMs), ethics

O75: Digital informed consent/assent matrix based on Common Condition of Use Elements: Coproducing an APP to track the process, return results, re-contact

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(1) CNR, (2) BBMRI, (3) UNIBO, (4) Unimib

Description

In 2017, the BBMRI.it community co-produced an informed consent matrix for the biobank with citizens, patients, and ethics committees, shared as CC04 https://repository.bbmri.it/s/stC8Lc4kPDn2qQt. The aim was to harmonize the informed consent as a participatory process, addressing the challenge of returning results, reviewing consent preferences, and re-contacting participants within a nationally sustainable accountability framework.

The recently funded “Strengthening BBMRI.it" project has as a step forward the objective of co-producing/developing a digital pathway for accessing informed consent and the return of individual results integrated into the electronic medical record or in a dedicated APP.

Eight biobanks were selected based on geographical distribution, biobanking scope, ELSI and digital maturity, and were involved with their respective information systems in the structuring of a standard digital consent matrix based on the Common Condition of Use Elements developed in EJPRD.

Sample characterization with consent preferences is a priority to facilitate third-party access and biobank interaction with participants. This matrix provides an APP, a digital interface between the participant-citizen and the biobank, facilitating the double flow, from the biobank to the participant regarding the return of results and re-contact, and from the participant to the biobank regarding review of consent preferences.

In addition to the basic information provided by the ethical-regulatory framework, the APP will offer paths and tools for in-depth and documented consent preferences and will facilitate re-contact.

The co-design process engages all the players, especially mature minors for the digitized assent.

Work is underway, the prototypes will be made available as a common good.

Keywords

common conditions of use elements CCEs, matrix, digital informed consent, digital informed assent, returning individual result, re-contact

O77: Laser-based state of the art cryotube labelling

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Description

Cryotubes featuring 2D codes at the vessel bottom prove advantageous for handling within fully automated biorepositories. However, they present a challenge for most laboratory analyzers and technicians receive no human-

readable information, complicating manual processing and increasing the likelihood of mix-ups.

To address these issues, we adopt a dual-labeling approach for our 2D coded cryotubes. In addition to the bottom 2D code, we include a paper-based side code containing a 1D barcode as well as human-readable information such as study name and/or sample material (e.g., EDTA plasma). Due to the increased outer diameter, paper-based labeling can pose challenges during frost or when used with high density racks.

A Tube Laser Marker (TLM) from LVL - a user-friendly tool with customizable programming - allows customers to define the content of the side code without paper. The TLM has proven reliable, exhibiting no failures in its inaugural year and successfully labeling around 10,000 cryotubes. Picking rates remain unaffected and the laser labels demonstrate resilience against mechanical or chemical agitation.

Additional side coding for cryotubes is a well-established practice. The limitations associated with paper-based side barcodes can be entirely overcome by employing laser-based labelling, which boasts impeccable labeling performance, reading characteristics and does not have any adverse impact on picking rates.

Keywords

Cryotube labelling, fully automated biorepositories, Tube Laser Marker