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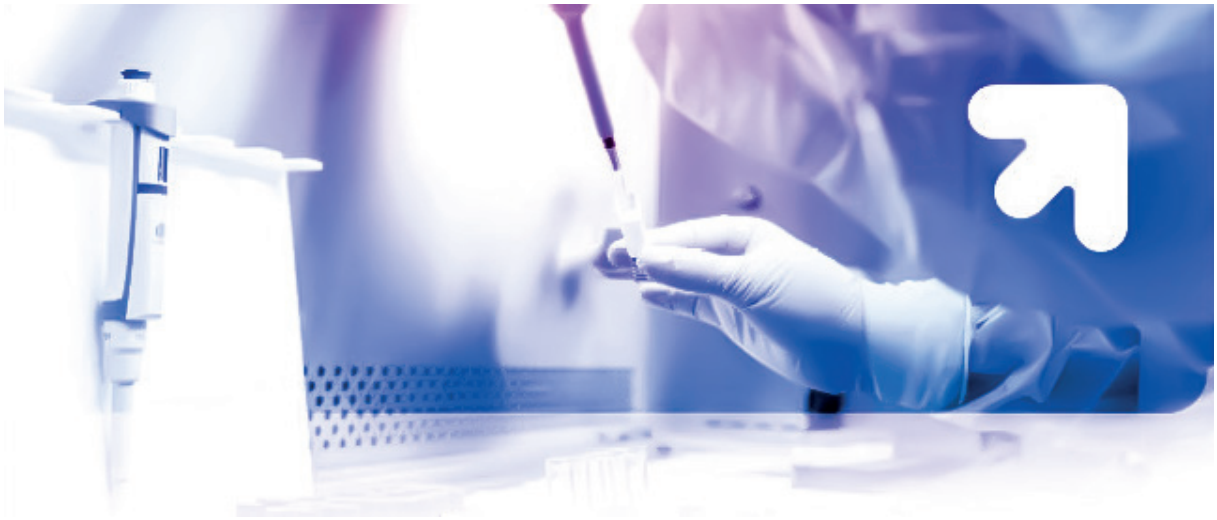

III KRAJOWA NAUKOWO-SZKOLENIOWA KONFERENCJA BIOBANKÓW POLSKICH

Badania populacyjne i omiczne a rozwój biobankowania materiału biologicznego

Łódź, 6-8 listopada 2019 r.

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PROGRAM KONFERENCJI

środa, 6 listopada 2019 r.

08:00-15:00 Hol	**REJESTRACJA**		
08:00-09:00 Sala 103	**Przygotowanie wniosku do komisji bioetycznej w kontekście biobankowania** Prowadzący: dr hab. Marek Czarkowski		
09:00-09:30 Sala 115	**QMS – Wykład: Współpraca naukowa** Prowadzący: dr Joanna Gleńska-Olender	09:00-10:00 Sala 102	**Bezpieczeństwo informacji – sytuacja w PSB** Prowadzący: dr Dominik Strapagiel, mgr Błażej Marciniak
09:30-10:00 Sala 115	**QMS – Wykład: Zarządzanie ryzykiem** Prowadzący: mgr Karolina Zagórska, mgr Patrycja Sitek		
10:00-11:00 Sala 103	**QMS 1 – Współpraca naukowa** Prowadzący: dr Joanna Gleńska-Olender, mgr Michał Laskowski	10:00-11:00 Sala 102	**Model finansowy Biobanku** Prowadzący: dr n. med. Łukasz Kozera
11:00-12:00 Sala 103	**QMS 2 – Zarządzanie ryzykiem** Prowadzący: mgr Karolina Zagórska, mgr Patrycja Sitek	11:00-12:00 Sala 102	**Formularz zgody** Prowadzący: dr hab. Jakub Pawlikowski
12:00-13:00 Sala 103	**Faza przedanalityczna – dobre praktyki w zakresie biobankowania** Prowadzący: dr n. med. Łukasz Kozera; mgr Paulina Zagórska-Szlufik	12:00-13:00 Sala 102	**Bezpieczeństwo informacji – sytuacja w PSB** Prowadzący: dr Dominik Strapagiel, mgr Błażej Marciniak
13:00-14:00 Sala 103	**Ochrona danych osobowych w biobankach – aspekty prawne** Prowadzący: dr Dorota Krekora-Zając	13:00-14:00 Sala 102	**Zastosowanie reguł wpływu społecznego w komunikatach na temat biobankowania** Prowadzący: dr Michał Wiechetek
14:00-15:00 Patio	**PRZERWA OBIADOWA**		
15:00-16:00 Sala 103	**QMS 1 - Współpraca naukowa** Prowadzący: dr Joanna Gleńska-Olender, mgr Michał Laskowski	15:00-17:00 Sala 102	**Praktyczne aspekty kontroli jakości w biobanku komórkowym** Prowadzący: dr Ilona Szabłowska-Gadomska, dr Anna Chróścicka
16:00-17:00 Sala 103	**QMS 2 - Zarządzanie ryzykiem** Prowadzący: mgr Karolina Zagórska, mgr Patrycja Sitek		
17:00-18:00 Sala 103	**Faza przedanalityczna – dobre praktyki w zakresie biobankowania** Prowadzący: dr n. med. Łukasz Kozera, mgr Paulina Zagórska-Szlufik	17:00-18:00 Sala 102	**Ochrona danych osobowych w biobankach – aspekty prawne** Prowadzący: dr Dorota Krekora-Zając

PROGRAM KONFERENCJI

czwartek, 7 listopada 2019 r.

07:30-10:00 Hol	**REJESTRACJA**	
08:15-08:30 Aula	**OTWARCIE KONFERENCJI**	Prowadzący: dr Dominik Strapagiel, dr n. med. Łukasz Kozera, prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł
08:30-09:00	**Wykład inauguracyjny** **prof. dr hab. Elżbieta Żądzińska** Prorektor ds. nauki Uniwersytetu Łódzkiego *History and present study of modern and archaic populations at the Department of Anthropology of the University of Lodz*	
09:00-10:50 Aula	**SESJA I: BADANIA POPULACYJNE**	Prowadzący: dr n. med. Łukasz Kozera, dr inż. Agnieszka Matera-Witkiewicz
09:00-09:30	**dr n. med Piotr Bandosz, dr hab. med. Tomasz Zdrojewski, prof. nadzw.** Gdański Uniwersytet Medyczny, Komitet Zdrowia Publicznego PAN *Potential gains from biobanking of biological samples collected in population surveys*	
09:30-09:50	**prof. dr hab. n. med. Karol Kamiński** Zakład Medycyny Populacyjnej i Prewencji Chorób Cywilizacyjnych, Uniwersytet Medyczny w Białymstoku *Reality bites – the development of biobanking strategy in multidisciplinary medical enviroment*	
09:50-10:10	**dr Wesley Oprzedek** Massachusetts Institute of Technology; Partner Firma LICONIC, Firma LKB-ARTIS Sp. z o. o. *Automated Sample storage and Biobanking solutions ranging from -20C to -196C*	
10:10-10:30	**dr hab. n. med. Katarzyna Zatońska, prof. nadzw.** Katedra i Zakład Medycyny Społecznej, Uniwersytet Medyczny im. Piastów Śląskich we Wrocławiu *PURE Poland study – design*	
10:30-10:50	**mgr Michał Karpeta** Regionalne Centrum Naukowo-Technologiczne *Screening tests in the Świętokrzyskie voivodeship*	
10:50-11:10 Patio	**PRZERWA KAWOWA**	

PROGRAM KONFERENCJI

11:10-13:30 Aula	**SESJA II: SESJA KONSORCJUM PROJEKTU BBMRI.PL ORAZ POLSKIEJ SIECI BIOBANKÓW**	Prowadzący: dr hab. med. Leszek Kalinowski, prof. nadzw., prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł
11:10-11:30	**dr n. med. Łukasz Kozera, dr Anna Chróścicka** Krajowy Ośrodek Wiodący – Sieć Badawcza ŁUKASIEWICZ – PORT Polski Ośrodek Rozwoju Technologii, Warszawski Uniwersytet Medyczny *Current Status of the Polish Biobanking Network*	
11:30-11:50	**dr inż. Agnieszka Matera-Witkiewicz** Biobank Uniwersytetu Medycznego im. Piastów Śląskich we Wrocławiu *QMS BBMRI.pl Expert Centre activity*	
11:50-12:10	**Matt Botterman** Brooks Life Sciences, Manchester, United Kingdom, Partner Bioanalytic Maciej Stopa *Starting small but aiming high*	
12:10-12:30	**dr hab. Jakub Pawlikowski, dr Michał Wiechetek** Uniwersytet Medyczny w Lublinie *Potential donors of biological material to biobanks – psychosocial characteristics*	
12:30-12:50	**mgr inż. Paulina Zagórska-Szlufik** Regionalne Centrum Naukowo-Technologiczne *Introduction of a uniform quality control system for all stages of the functioning of national biobanks*	
12:50-13:10	**dr Dominik Strapagiel** Pracownia Biobank, Uniwersytet Łódzki *Distributed IT systems within Polish Biobanking Network*	
13:10-13:30	**mgr inż. Jakub Szymanowski** Gdański Uniwersytet Medyczny *Central IT solutions for the Polish Biobank Network*	
13:30-14:20 Patio	**PRZERWA OBIADOWA**	
13:30-14:20 Hol	**SESJA PLAKATOWA (numery parzyste)**	
14:20-15:20 Aula	**SESJA III: BADANIA GENOMICZNE CZŁOWIEKA W MEDYCYNIE – Sesja pod patronatem Polskiego Towarzystwa Genetyki Człowieka**	Prowadzący: dr Dominik Strapagiel, prof. dr hab. n. med. Maciej Borowiec
14:20-14:40	**dr hab. n. med. Bartosz Wasąg** Katedra i Zakład Biologii i Genetyki Medycznej, Gdański Uniwersytet Medyczny *Molecular diagnostics in oncology – today and tomorrow*	

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PROGRAM KONFERENCJI

14:40-15:00 **prof. dr hab. n. med. Maciej Borowiec**
Zakład Genetyki Klinicznej, Uniwersytet Medyczny w Łodzi
Large-scale tests in the diagnosis of monogenic diabetes

15:00-15:20 **prof. dr hab. n. med. Rafał Płoski**
Zakład Genetyki Medycznej, Warszawski Uniwersytet Medyczny
Whole exome and whole genome sequencing for diagnosis and discovery of novel human disease

15:20-16:30 Aula **DEBATA OKSFORDZKA** Prowadzący: dr Anna Chróścicka, dr Joanna Wójtowicz

Temat debaty: ***Czy kolekcje materialnych próbek zostaną w przyszłości (w perspektywie 20 lat) zastąpione przez banki informacji?***

15:20-16:30 Eksperci:

dr Dominik Strapagiel
Pracownia Biobank, Uniwersytet Łódzki

dr hab. med. Leszek Kalinowski, prof. nadzw.
Gdański Uniwersytet Medyczny

dr Thierry van de Wetering
Pomorski Uniwersytet Medyczny w Szczecinie

dr n. med. Łukasz Kozera
Krajowy Ośrodek Wiodący – Sieć Badawcza ŁUKASIEWICZ – PORT Polski Ośrodek Rozwoju Technologii

prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł
Warszawski Uniwersytet Medyczny

prof. dr hab. n. med. Teresa Wierzba-Bobrowicz
Instytut Psychiatrii i Neurologii w Warszawie

16:40-17:40 Aula **SPOTKANIE POLSKIEJ SIECI BIOBANKÓW (za zaproszeniem - członkowie i obserwatorzy PSB)**

19:00-01:00 Restauracja PRZERWA **BANKIET POWITALNY – spotkanie integracyjne**
Restauracja PRZERWA ul. Wólczańska 128/134, Łódź

piątek, 8 listopada 2019 r.

08:00-10:00 Hol **REJESTRACJA**

09:00-11:00 Aula **PANEL DYSKUSYJNY – INTERESARIUSZE POLSKIEJ SIECI BIOBANKÓW** Prowadzący: dr n. med. Jarosław Skokowski

09:00-11:00 **MISJA I CELE POLSKIEJ SIECI BIOBANKÓW (PSB)**

- **dr n. med. Łukasz Kozera**

PROGRAM KONFERENCJI

09:00-11:00

PRZEDSTAWICIELE GRUP PACJENCKICH

- **Aleksandra Rudnicka**
  Polska Koalicja Pacjentów Onkologicznych
- **dr Maria Libura**
  Klub Jagielloński, Zakład Dydaktyki i Symulacji Medycznej Uniwersytet Warmińsko-Mazurski
- **Joanna Frątczak-Kazana**
  Fundacja Alivia

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- **dr Aleksander Sowa**
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  Narodowe Centrum Nauki
- **dr n. med. Beata Jagielska**
  Polska Koalicja Medycyny Personalizowanej, Centrum Onkologii
- **dr Adam Dawidziuk**
  Narodowe Centrum Badań i Rozwoju
- **dr n. med. Ewa Szkiłądź**
  Krajowy Punkt Kontaktowy Programów Badawczych UE
- **dr Aleksandra Wesołowska**
  Agencja Badań Medycznych

PANEL DYSKUSYJNY

11:00-11:35 Patio — **PRZERWA KAWOWA**

11:35-13:15 Aula — **SESJA IV: BADANIA OMICZNE** — Prowadzący: dr n. med. Jarosław Skokowski, dr hab. n. med. Bartosz Wasąg

11:35-11:55 **dr hab. Marcin Ratajewski, prof. IBM**
Instytut Biologii Medycznej Polskiej Akademii Nauk
Transcriptomics in the service of anti-cancer therapy. The case of SIRT2 and melanoma

11:55-12:15 **dr Gracjan Wątor**
Ośrodek Genomiki Medycznej OMICRON, Collegium Medicum Uniwersytetu Jagiellońskiego
Ultra deep sequencing of colorectal tumors: heterogeneity of somatic mutations in the context of lymph node involvement

12:15-12:35 **dr n. med. Bogusław Nedoszytko**
Katedra i Klinika Dermatologii, Wenerologii i Alergologii, Gdański Uniwersytet Medyczny
Occurrence of polymorphic changes in the genome of psoriasis patients using the GWAS technique in the Polish population - analysis of preliminary research results

12:35-12:55 **prof. dr n. med. Tomasz Wojdacz**
Pomorski Uniwersytet Medyczny
Discovery and clinical validation of methylation biomarker signatures in Chronic Lymphocytic Leukemia (CLL)

12:55-13:15 **dr hab. Michał Ciborowski**
Centrum Badań Klinicznych, Uniwersytet Medyczny w Białymstoku
From biobanking to metabolomics analyses – evaluation of small molecules in different aspects of non-small cell lung cancer

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PROGRAM KONFERENCJI

13:15-14:10 **PRZERWA OBIADOWA**

13:15-14:10 **SESJA PLAKATOWA (numery nieparzyste)**

14:10-15:00 Aula **SESJA V: DONIESIENIA KONFERENCYJNE**

Prowadzący: dr hab. Jakub Pawlikowski, dr Anna Chróścicka

14:10-14:20 **dr Jolanta Gromadzińska**
Instytut Medycyny Pracy w Łodzi
Association between urine phthalate metabolites and thyroid hormones

14:20-14:30 **dr Joanna Wójtowicz**
Narodowy Instytut Geriatrii, Reumatologii i Rehabilitacji w Warszawie
Research inland versus abroad, in public versus commercial institutions – what are Polish parents fears about pediatric biosamples sharing

14:30-14:40 **dr Justyna Jarczak**
Pracownia Biobank, Uniwersytet Łódzki
Genetic diversity of Polish male population based on the analysis of Y chromosome

14:40-14:50 **dr Joanna Banasiuk**
Wydział Prawa, Uniwersytet w Białymstoku
Commercialization of research results based on the use of human biological material and data

14:50-15:00 **mgr Paulina Borówka**
Katedra Antropologii, Uniwersytet Łódzki

dr Justyna Marchewka
Zakład Biologii Człowiek, Uniwersytet Kardynała Stefana Wyszyńskiego w Warszawie

Biobanking of bone material - the case study of an craniosynostosis individual

15:00-15:30 Aula **ZAKOŃCZENIE KONFERENCJI**

Prowadzący: dr Dominik Strapagiel, dr n. med. Łukasz Kozera, prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł

KOMITET ORGANIZACYJNY

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dr Elżbieta Gocek
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Natalia Zysiek, Gdański Uniwersytet Medyczny

dr Anna Chróścicka
prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł, Warszawski Uniwersytet Medyczny

dr Dominik Strapagiel
mgr inż. Błażej Marciniak
mgr inż. Sylwia Dobrowolska, Pracownia Biobank, Uniwersytet Łódzki

dr hab. Jakub Pawlikowski
dr Dorota Krekora-Zając, Uniwersytet Medyczny w Lublinie

dr inż. Agnieszka Matera-Witkiewicz
dr Joanna Gleńska-Olender
mgr Karolina Zagórska, Uniwersytet Medyczny im. Piastów Śląskich we Wrocławiu

mgr inż. Paulina Zagórska-Szlufik
Regionalne Centrum Naukowo-Technologiczne w Chęcinach koło Kielc

KOMITET NAUKOWY

Przewodnicząca: prof. dr hab. n. med. Małgorzata Lewandowska-Szumieł
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prof. dr hab. Elżbieta Żądzińska,
prof. Wawrzyniec Lawrence Dobrucki
dr hab. Jakub Pawlikowski
dr n. med. Łukasz Kozera
dr inż. Agnieszka Matera-Witkiewicz
dr Anna Chróścicka
dr Joanna Gleńska- Olender
dr Elżbieta Gocek
dr Dominik Strapagiel
dr Joanna Wójtowicz

EUROPEAN JOURNAL OF TRANSLATIONAL
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Scientific cooperation as the basic element for the Biobanks development

Joanna Gleńska-Olender[1,2,3], Michał Laskowski[1,2], Karolina Zagórska[1,2], Patrycja Sitek[1,2], Agnieszka Matera-Witkiewicz[1,2]

[1] Screening Laboratory of Biological Activity Test and Collection of Biological Material, Faculty of Pharmacy with Division of Laboratory Diagnostics, Wrocław Medical University, Poland
[2] BBMRI.pl Consortium
[3] Świętokrzyski Biobank; Regional Science and Technology Center, Chęciny, Poland

Abstract

National and international scientific cooperation plays an important role in the development of biobanks. It has an impact on improving the biobanking process, increasing the research potential, including the parent unit within which it operates. Biobanks are an important infrastructure for research supporting scientific progress and innovation. They help scientists, to collect high-quality samples and data for new experiments. Chapter 14 of the Quality Standards for Polish Biobanks contains guidelines for scientific cooperation. It describes the process of acquiring a partner, recommendations for entering into cooperation agreements, rules for sharing biological material for research and communication with a scientific partner. In addition, they describe the process of using research results in publications and scientific studies. The implementation of research cooperation with the participation of employees of both parties influences the development of the scientific, research and technical staff. Biobanks through high-quality biosecurity controls and data are known as safe and reliable scientific partners. Multicenter cooperation in which a biobank participates leads to reliable for research results, and is a source of development for the biobank itself.

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Citation

Gleńska-Olender J, Laskowski M, Zagórska K, Sitek P, Matera-Witkiewicz A. Scientific cooperation as the basic element for the Biobanks development. Eur J Transl Clin Med. 2019;2(Suppl.3):14.

Risk analysis for Biobank in practical terms – key issues, methodology and interpretation of results

Karolina Zagórska[1,2], Patrycja Sitek[1,2], Joanna Gleńska-Olender[1,2], Michał Laskowski[1,2], Agnieszka Matera Witkiewicz[1,2]

[1] Screening Laboratory of Biological Activity Tests and Collection of Biological Material, Faculty of Pharmacy with the Division of Laboratory Diagnostics, Wrocław Medical University, Wrocław, Poland
[2] BBMRI.pl Consortium

Abstract

Risk management is a systematic procedure based on elements such as: (1) identification of threats to the process and the probability of these threats becoming critical, (2) risk measurement, (3) risk guidence and (4) monitoring and control. An exchange of information between personnel involved in the risk management process to explain reasons and justifications proposed methods of operation is crucial in this process. Reliable risk management is the basis for ensuring the safety of biological material and data related to it from the moment of its generation to release. Properly selected risk management tools enable identifying areas that pose the greatest risk for the biobanking process. The basic tools of risk analysis are: 5Why method, FMEA and Ishikawa diagram. The subject of risk management is thoroughly discussed in Chapter 11 of the Quality Standards for Polish Biobanks regarding deviations and non-compliant products and services. Incorporation into the sample life cycle of the presented guidelines is a guarantee of maintaining the appropriate quality of services offered by Biobank.

Citation

Zagórska K, Sitek P, Gleńska-Olender J, Laskowski M, Matera-Witkiewicz A. Risk analysis for Biobank in practical terms – key issues, methodology and interpretation of results. Eur J Transl Clin Med. 2019;2(Suppl.3):15.

EUROPEAN JOURNAL OF TRANSLATIONAL
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History and present study of modern and archaic populations at the Department of Anthropology of the University of Łódź

Elżbieta Żądzińska, Wiesław Lorkiewicz, Aneta Sitek

Department of Anthropology, Faculty of Biology and Environmental Protection, University of Łódz, Łódź, Poland

Abstract

Since its foundation in 1945, the Department of Anthropology has conducted research on modern and archaic human populations. The research included: anthropological population pictures, auxological studies repeated every 10 years, anthropological expeditions to Asia and Africa, demographic research, clinical anthropology research, excavations exploring archaic and historical populations. Currently, the collection of the Department of Anthropology UL includes databases covering anthropometric features and socio-economic data for the Polish population from 1956 to the modern period. These databases have a total of over 25,000 records. Cephalometric databases for the population of Mongolia and Egypt have over 3,000 records. The Department's collections also contain skeletal series documenting the continuous occupation of the Kujawy region from the Neolithic to modern times. Osteological bases have over 3,500 skeletons. For their analyses, the Department of Anthropology in 2008 obtained the prestigious NOVUM grant of the Foundation for Polish Science. Recently, standard anthropological studies have been extended to include genetic analyses. Thanks to this, the population bases of the Department of Anthropology have increased by the PUPILS database containing anthropometric, socio-economic and genetic data of 960 people aged 6-13 and the aDNA database for skeletal series from Kujawy region with amount, at present, close to 500 records. Last years the Genetic Auxology Laboratory and the sterile aDNA Laboratory were established in the Department. The most important publications resulting from these studies present, among others: percentile grids for Polish newborns and children, analysis of risk factors for the development of excess and weight deficiencies (including genetic determinants of overweight and obesity), an indication of prenatal factors disrupting progressive ontogenesis, an indication of gene polymorphisms related to skin and hair pigmentation in the Polish population (invention entitled *A method of detecting a genetically determined predisposition to having a specific hair color* coverd by patent protection), discovery of large genetic contribution of autochthonous Mesolithic populations to agricultural societies of late post-Linear cultures in territory of today's Poland during the Middle Neolithic based of ancient genome-wide analyses.

Citation

Żądzińska E, Lorkiewicz W, Sitek A. History and present study of modern and archaic populations at the Department of Anthropology of the University of Łódź. Eur J Transl Clin Med. 2019;2(Suppl.3):16.

Potential gains from biobanking of biological samples collected in population surveys

Piotr Bandosz, Tomasz Zdrojewski

Department of Preventive Medicine and Education, Medical University of Gdańsk, Gdańsk, Poland

Abstract

During last decade, we prepared and conducted three nationally representative surveys of adults in Poland. There were: 1) NATPOL 2002 study (n = 2403) – study aimed to asses prevalence of cardiovascular risk factors, 2) NOMED-AF (n = 3014) – study on prevalence of hidden atrial fibrillation and 3) Polsenior-2 (n = 5600) – aimed to assess health problems of elderly in Poland. All three study samples were drawn using multistage and clustered random sampling. For the NATPOL study age range was 18+, for two other studies the age range was 65+ and 60+, with overrepresentation of eldest subjects. In all studies we performed detailed questionnaires, anthropometric measurements, blood pressure measurements and other tests (like Mini–Mental State Examination, long-term ECG monitoring). Venous blood samples were drawn at fasting state, and several biochemical parameters were assessed. Remaining biological specimen was frozen and stored in the repositories.

In this talk we will discuss potential future utility of collected specimens: to estimate population reference values for laboratory tests, use as control or reference groups in epidemiological studies or evaluation of the new potential risk factors of chronic diseases.

Citation

Bandosz P, Zdrojewski T. Potential gains from biobanking of biological samples collected in population surveys. Eur J Transl Clin Med. 2019;2(Suppl.3):17.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):18

Reality bites – the development of biobanking strategy in multidisciplinary medical environment

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Abstract

Medical research naturally combines in vivo and in vitro studies that complement each other in order to provide crucial data on physiological and pathological phenomena. One of the great challenges is collection of appropriate patient populations and their biomaterial. Structured biobanking program that will include cohorts of patients with particular diseases as well as prospective clinical research and population based studies may provide additional possibilities for multidisciplinary research. Moreover it creates opportunity for collaboration between academia and industry for the benefit of both parties. It is important to see biobank not only as a place to store samples, but an organisation of data and sample collection. Biobanking requires development and implementation of harmonised standard operating procedures for data and sample collection is a big challenge. Medical University of Białystok have started to develop a biobanking programme available for its scientists, with the emphasis on population research and cancer samples. This process long and full of challenges. Clearly defined milestones and involvement of enthusiastic scientists are crucial for the success.

Citation

Kamiński K. Reality bites – the development of biobanking strategy in multidisciplinary medical environment. Eur J Transl Clin Med. 2019;2(Suppl.3):18.

Automated Sample storage and Biobanking solutions ranging from -20°C to -196°C

Wesley Oprzedek

Massachusetts Institute of Technology, USA, Partner LICONIC, Partner LKB-ARTIS Sp. z o.o.

Abstract

The transition from manual traditional storage systems into advanced semi or fully automated biobanking systems confers many advantages including efficient and flexible sample access and control of sample integrity. Other important advantages include the elimination of human error and secure integration of metadata and LIMS systems, with the remote access capability. Long term running cost comparison and ROI (return on investment) are made between Manual and Automated systems. Solutions are available based on business's needs, space requirement, process flexibility, and future growth accommodations. Benefits of central biobanking storage facilities and global access capabilities. Various system types ranging from smaller portable systems to midsize processing biobanks verses larger and longer term storage systems constructed onsite. Options and requirements for migrating legacy sample collections from manual to automated processes will be discussed. Liconic Instruments provides some examples of implementation for systems that are built to address specific application requirements are provided. These include systems that are built for small dedicated processes; larger biobank operations that require high density storage with systems that can fit into any dimension space; systems that work well with legacy collections or unpredictable rack and tube dimensions; cryogenic storage; and hybrid temperature systems. These Liconic systems are serving various application needs across the world with over 60+ installations in the North America, Europe, Asia Pacific, and many new installations in the rapidly expanding Chinese biobanking network.

Citation

Oprzedek W. Automated Sample storage and Biobanking solutions ranging from -20°C to -196°C. Eur J Transl Clin Med. 2019;2(Suppl.3):19.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):20

PURE Poland study – design

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Abstract

Risk factors arising from environmental, genetic and psychosocial background lead to increase in incidence of noncommunicable diseases (NCD). Project's aim is to describe this processes and develop actions targeting major health challenges like increase of obesity, diabetes and cardiovascular diseases. PURE study is a global research project established to elaborate a strategy to link and analyze environmental factors with individual biological (metabolic, genetic and lifestyle) conditions. Participants were recruited from 21 countries, reaching in total over 150 000 people.

In Poland the project started back in 2007 at the Wrocław Medical University. Data was collected cyclically with 3 years intervals, the research is planned for 12 years (2007-2019). At present 9-year follow-up has been finished. Overall 2036 people aged between 30 and 85 years old were enrolled to the study. Project targets to examine households enrolled to the study and their communities: 1210 people with urban residence and 826 people with rural residence. At the first stage of the research participants were interviewed (face to face) with 6 study questionnaire forms: household, family, health (individual), international physical activity questionnaire (IPAQ), food frequency questionnaire, 24h dietary recall.

Citation

Zatońska K. PURE Poland study – design. Eur J Transl Clin Med. 2019;2(Suppl.3):20.

Screening tests in the Świętokrzyskie voivodeship

Michał Karpeta[1,2], Paulina Zagórska-Szlufik[1,2], Anna Sularz[1,2], Ilona Sychowska[1,2], Karolina Niebudek-Jach[1,2], Ewa Twardowska[1,2], Katarzyna Rozmianiec[1,2], Sylwia Knap[1,2]

[1] Regional Science and Technology Center, Poland
[2] BBMRI.PL

Abstract

At the beginning of March 2018, the Regional Science and Technology Center began screening tests of the Świętokrzyskie voivodeship inhabitants by implementing three projects. The aim of the first project is to assess the supply of vitamin D among the inhabitants of the Świętokrzyskie voivodeship. The obtained results will allow to determine the factors affecting the level of vitamin D and provide information on the extent to which vitamin D deficiency affects residents. So far, 1900 inhabitants have participated in the project.

The determination of the lipid profile is second screening test. Conducting research on a large number of participants will create a lipid profile of the region's population. So far, 700 inhabitants have participated in the project.

Last project was created due to the lack of keeping the national register of people suffering from celiac disease. Study participants in the age bracket between 3-5 and 12- 15 years old are tested for the occurrence of celiac disease. So far 400 people have used the project. In the case of 42 project participants, non-standard results were observed. Children were referred for in-depth diagnostics for celiac disease.

Citation

Karpeta M, Zagórska-Szlufik P, Sularz A, Sychowska I, Niebudek-Jach K, Twardowska E, Rozmianiec K, Knap S. Screening tests in the Świętokrzyskie voivodeship. Eur J Transl Clin Med. 2019;2(Suppl.3):21.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):22

Current Status of the Polish Biobanking Network

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Abstract

Access of Poland to BBMRI-ERIC was the starting point of a nationwide project which main goal is to create Polish Biobanking Network (PBN) and bring together entities interested in professional, harmonious and standardized biobanking of biological material.

Constant information campaign and numerous meetings with scientists across the country – resulted in 42 members and observers of the PBN. Polish biobanks collect various types of biological material. 69% of PBN members declare their willingness to share their collections. Most Polish biobanks are localized at universities (43%) and research institutes (26%) but there are also 5 units (12%) located in private companies. Majority of Polish biobanks take care of obtaining consents from donors (34 units) and bioethical commissions (31 units) for the use of biological material. They also have implemented or are currently in the process of implementation of QMS for collected samples (30 units).

The landscape of PBN is changing rapidly due to creation and involvement of new members.

This work was supported by Ministry of Science and Higher Education grant DIR/WK/2017/2018/01-1

Citation

Kozera Ł, Chróścicka A. Current Status of the Polish Biobanking Network. Eur J Transl Clin Med. 2019;2(Suppl.3):22.

QMS BBMRI.pl Expert Centre activity

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[2] BBMRI.pl Consortium
[3] Świętokrzyski Biobank, Regional Science and Technology Center, Chęciny, Poland

Abstract

ABBMRI.pl Consortium was established as a part of the European Research Infrastructure for Biobanking, BBMRI-ERIC. The organization currently includes 20 countries making it one of the largest research infrastructures in Europe. The main goal of BBMRI.pl is to build a platform for scientific-research cooperation and research development. One of BBMRI.pl activity is QMS Expert Centre organization where many tools for Polish Biobanking Network (PBN) have been developed to implement and improve quality management systems in theirs biobanks: - Audit programme (remote audits and directly dedicated visits with audit reports with recommendations and Corrective Action Plans preparation) - Workshops and trainings with practical 5 / 12 support (confirmed by a certificate) - Quality Standards for Polish Biobanks and Auditor's Manual Handbook preparation and sharing - Consulting and supporting activities for QMS implementation, ISO certification/accreditation The main assumption is to help Members and Observers of PBN to improve the quality of collected samples and data, thus the increasement of the visibility of polish researchers in international collaborations and projects can be achieved, where biological material and data are used. The team consists of experts and specialists having experience in the research area such as biomedicine and biobanking, pharmacy and biotechnology, management and quality assurance in healthcare entities, diagnostic laboratories, tissue and cell banks. They are representatives of the Polish Committee for Standardization in TC 287 Biotechnology and are working in Working Groups concerning Quality Management in BBMRI-ERIC.

Citation

Matera-Witkiewicz A, Zagórska K, Laskowski M, Sitek P, Gleńska-Olender J. QMS BBMRI.pl Expert Centre activity. Eur J Transl Clin Med. 2019;2(Suppl.3):23.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):24

Starting small but aiming high

Matt Botterman

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Abstract

A biobank for many labs consists of just a single freezer. Many scientists primary concern involves the research question and do not give as much consideration to the biobanking process. However, It's at this stage, with low sample numbers that future success is ultimately decided as samples stored incorrectly might not be suitable for future use. This presentation aims to give a detailed checklist for a new, manual biobank covering the 2 most important factors, the Process being employed, and the Equipment being used.

The process for biobanking system actually works in reverse chronologically. Starting with thoughts around how to collect and add samples to the biobank can lead to problems downstream. Instead we look at what come out of the biobank and what is import around this. From there we can work backwards through storage conditions, input into the bank, processing and collection.

Finally, we look at considerations for the equipment involved including collection, storage, transportation and monitoring. The aim is to ensure the sample is held at optimal conditions and chain of custody / traceability is maintained so that the maximal value can be extracted from the samples.

Citation

Botterman M. Starting small but aiming high. Eur J Transl Clin Med. 2019;2(Suppl.3):24.

Potential donors of biological material to biobanks – psychosocial characteristics

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[2] Department of Social Psychology and Psychology of Religion, The John Paul II Catholic University of Lublin, Poland

Abstract

The development of biobanks depends on the willingness of donors to cooperate. Effective communication with potential donors is particularly important for population biobanks. The aim of the presentation is to describe a psychosocial characteristics of potential donors of biological material based on the results of social research carried out on a representative group of 1,100 adult Poles. Results relate to: 1) willingness to submit samples to biobanks and motivational factors; 2) Social communication ways about biobanks; 3) psychosocial profile of potential donors. Results indicate that 47.5% of respondents declare the willingness to donate samples to biobanks at various levels. A significant relationship was observed between willingness to donate biosample and trust (in scientists and doctors) and preferred values (personal development, tradition and knowledge). The most effective ways of communications about biobanks seem to be a family doctor and the most motivating factors to donate biosamples are: reimbursement of travel expenses and the opportunity to know the results of research done on a given sample. The willingness to donate samples to the biobank seems to be the result of many psychosocial variables that can be taken into account in biobank management and social campaigns regarding biobanks.

Citation

Pawlikowski J, Wiechetek M. Potential donors of biological material to biobanks – psychosocial characteristics. Eur J Transl Clin Med. 2019;2(Suppl.3):25.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):26

Introduction of a uniform quality control system for all stages of the functioning of national biobanks

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[2] BBMRI.PL

Abstract

A uniform quality control system is a crucial step for controlling all stages of Biobank's operation. As part of the project implementation 4 Biobanks will collect a minimum of 500 samples of biological material from the region in which they operate. Quality control will take place in two stages. First stage will be performed by internal teams in each of the biobanks. Then, the National Lead Center will conduct an analysis of the quality control results from each unit. All critical parameters will be considered. A complement to this validation will be participation in the external laboratory quality control program for biobanks and biorepositories. The main results of the project will be standard operating procedures which will describe every step of proceeding with material from collection, through its 1/2 marking, transport, preparation and storage. In addition to the validation of developed solutions there will be published a report on the state of health of the population in individual regions of Poland and in relation to the entire country. The project is supported by the Polish Ministry of Science and Higher Education (DIR/WK/2017/01 and DIR/WK/2017/2018-01).

Citation

Zagórska-Szlufik P. Introduction of a uniform quality control system for all stages of the functioning of national biobanks. Eur J Transl Clin Med. 2019;2(Suppl.3):26.

Distributed IT systems within Polish Biobanking Network

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[2] BBMRI.pl Consortium

Abstract

Dynamic development of biobanking industry (both business and science) resulted in an increased number of IT systems for samples and data management. The most difficult and complicated case for the biobanking community was cooperation between institutions, equipped with different IT systems, in the field of scientific research, mainly data interchange and information flow. University of Łódź is responsible for developing tools for Biobanks like Information Management System, and distributed IT system for sample search and communication platform for sample users - BioFACE. We have developed BioSCOOP, a communication protocol in the form of a well documented JSON API, to harmonize and standardize the rules of communication between biobanks on the level of information about the donor together with information about the sample [1].

Our team is also responsible for information security audit program. All members of Polish Biobanking Network are subject to that audit and as the result there is prepared report about situation in particular biobank unit.

Citation

Strapagiel D, Marciniak B, Jarczak J, Lach J. Distributed IT systems within Polish Biobanking Network. Eur J Transl Clin Med. 2019;2(Suppl.3):27.

1. Jarczak, J., Lach, J., Borówka, P., et al. BioSCOOP – *Biobank Sample Communication Protocol. New approach for the transfer of information between biobanks* Database, Volume 2019, 2019, baz105, https://doi.org/10.1093/database/baz105

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):28

Central IT solutions for the Polish Biobank Network

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[2] BBMRI.pl Consortium

Abstract

In 2018, the team of the Department of Medical Laboratory Diagnostics - Biobank, Medical University of Gdansk, began work on the development of central IT tools for the BBMRI.PL and the Polish Biobank Network. The integrated platform includes elements related to the acquisition and processing of image data, data on biobanks, collections, samples, donors and research results with an integration module with national and clinical records. The development of such a solution is aimed at supporting communication and exchange of information on collected biological material between biobanks, biobanks and researchers and national nodes. The exchange of information and database 1/5 resources will not only allow for data standardization, but also will improve the quality of processed data, and through connection with national and clinical registers will open the new perspectives for clinical and omnic researches. The first version of the central IT system was released in September 2019. Access to the system is open to every registered biobank and researcher.

Citation

Szymanowski J. Central IT solutions for the Polish Biobank Network. Eur J Transl Clin Med. 2019;2(Suppl.3):28.

Molecular diagnostics in oncology – today and tomorrow

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[2] Laboratory of Clinical Genetics, University Clinical Centre, Gdańsk, Poland

Abstract

Implementation of modern molecular-targeted therapies and immunotherapy requires an assessment of molecular predictors in biological material from cancer patients. The emergence of next-generation sequencing (NGS) has revolutionized the approach to diagnostic procedures in personalized medicine. Recently, this method has been successfully implemented as a highly sensitive and cost-effective diagnostic tool to detect either germline or somatic mutations, even in DNA isolated from formalin-fixed paraffin-embedded (FFPE) material or in circulating tumor DNA (ctDNA). Current molecular methods used for diagnostics of oncological patients and future perspectives will be presented during the lecture.

Citation

Wasąg B. Molecular diagnostics in oncology – today and tomorrow. Eur J Transl Clin Med. 2019;2(Suppl.3):29.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):30

Large-scale tests in the diagnosis of monogenic diabetes

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Abstract

The course covers the latest news in the field of etiopathogenesis of diabetes in her broad terms, identifying both the substrate disease and therapeutic possibilities. Problems presented in the lecture relate to the genetic basis of disease, monogenic forms of diabetes identified in the pediatric population. The presentation will be presented previously known genetic basis of diabetes and possibilities of interpretation of the result, or "predictive" (providing consultative) affecting the behavior of the patient from the diagnostic and therapeutic e.g. the impact on the selection of therapy or verify (changes) have already implemented therapies patient and sick family members. At the same time the results of genetic identification in subsequent stages of the clinical - diagnostic in relation to the patient and his family plays a key role in genetic counseling and prognostic, not once drastically affecting the important decisions of family.

The lecture also aims to approximate the problem of modern genetics and its use in specialized laboratories from both the diagnostic and scientific and visibility of the problem of selection of appropriate tools of molecular biology to identify the problem from the side of "molecular genetics". The use of modern techniques, i.e. next-generation sequencing (NGS) as well as direct sequencing according to Sanger as an answer to the questions asked about the genetic background of the disease. Interpretation of his confirmation and documentation is at the present moment is also a big problem and presenting it in a readable form for the clinician, mistakes such a process are also discussed in the above lecture. They will also be presented exemplary results and interpretation indicating problems differences in clinical practice.

Citation

Borowiec M. Large-scale tests in the diagnosis of monogenic diabetes. Eur J Transl Clin Med. 2019;2(Suppl.3):30.

Whole exome and whole genome sequencing for diagnosis and discovery of novel human disease

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Abstract

In 2012 Department of Medical Genetics (Warsaw Medical University) has acquired Illumina HiSeq 1500 which allowed to establish whole exome sequencing (WES) as method for both research and diagnostic purposes. Since then we have performed > 1000 WES analyses, most of which aimed at finding diagnosis in patients suspected to suffer from rare disorders with a genetic basis. We also established shallow whole genome sequencing (WGS) based on Mate-pair libraries which we use to precisely map breakpoints in patients with symptomatic balanced chromosomal translocations. During the lecture selected findings will be presented illustrating how these approaches enable discovery of novel diseases (i.e. those caused by mutations in genes not yet associated with known human disorder).

Citation

Płoski R. Whole exome and whole genome sequencing for diagnosis and discovery of novel human disease. Eur J Transl Clin Med. 2019;2(Suppl.3):31.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):32-33

Occurrence of polymorphic changes in the genome of psoriasis patients using the GWAS technique in the Polish population – analysis of preliminary research results

Bogusław Nedoszytko[1], Marta Sobalska-Kwapis[8], Dorota Purzycka-Bohdan[1], Aleksandra Batycka-Baran[2], Paulina Barasińska[9], Rafał Czajkowski[4], Joanna Czerwińska[6], Magdalena Górecka- Sokołowska[4], Magdalena Krajewska-Włodarczyk[6], Leszek Kalinowski[7], Aleksandra Lesiak[9], Monika Matławska[6], Marta Macieja-Stawczyk[1], Joanna Narbutt[9], Agnieszka Owczarczyk- Saczonek[6], Waldemar Placek[6] , Anna Pasierb[3], Adam Reich[5], Lidia Rudnicka[3], Dominik Samotij[5], Marcin Słomka[8], Dominik Strapagiel[8], Justyna Szczęch[5], Jacek Szepietowski[2], Aneta Szczerkowska-Dobosz[1], Monika Zabłotna[1], Roman J. Nowicki[1]

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[2] Department of Dermatology, Venereology and Allergy, Wrocław Medical University, Wrocław, Poland
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[6] Department of Internal Medicine, School of Medicine, Collegium Medicum, University of Warmia and Mazury, Olsztyn, Poland
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[8] Biobank Laboratory, Department of Biophysic, University of Łódź, BBMRI Poland
[9] Department of Dermatology, Pediatric Dermatology and Oncology, Medical University of Łódź, Łódź, Poland

Introduction

Psoriasis is a disease with a complex multifactorial background in which genetic, environmental and epigenetic factors that regulate gene expression play a role. there are two basic types of psoriasis type I (first symptoms <40 years of age , strong genetic background) and type II (>40 ya, nongenetic). Genetic association, genomic and gene expression studies have shown the association of the type I disease with more than 80 chromosomal regions, and abnormal expression > 4,000 genes. Still, little is known about the genes located in psoriasis susceptibility regions (PSORS) and their epistatic effects in the development of different disease phenotypes. The aim of the study was to compare the incidence of SNP (single nucleotide polymorphisms) in the group of patients with psoriasis and the control group of healthy people determined by the GWAS (Genome Wide Association Study) method. The study was carried out in Poland for the first time. The study was conducted on a group of 618 patients with psoriasis (465 type I and 147 type II) and 5885 healthy people (POPOLOUS study)) as a control group. Genomic DNA was isolated from each person from saliva or peripheral blood, followed by SNP identification using an InfinumCoreExome (Illumina) matrix. Statistical analysis of the obtained results was carried out using the

PLINK 1.9 program, visualized in the Haploview and LocusZoom programs. According to the Bonferroni correction, statistically significant differences were assumed for $p < 10^{-7}$. From 558 231 SNPs in matrix after exclusion criterias, for the final analyse 233 644 SNPs were used. In patients with type II, no statistically significant differences were observed in the incidence of analysed SNPs compared to control ($p > 10^{-7}$). In contrast in type I psoriasis we found strong association of disease with 260 SNPs on chromosome 6 and *USP8* gene on chromosome 15. The high association of type I psoriasis with SNPs of the following genes on chromosome 6 has been observed: PSORC1, PSORC2 and PSORSC3, genes encoding proteins related to antigen recognition and presentation; *HLAC, HLA-DQA, HLADQB, HLA-DRB* and *TAP2*,coding for a C2 complement protein, encoding a protein associated with the regulation of endocytosis receptor: *NOTCH4*, transcription factors: RNF5, TCF19, POU5F1, NFKB inhibitor like 1, ser/thre kinase – *DXO*, structural epidermal proteins: CDSN, MUC22, tenascin XBand genes coding proteins associated with the activation of lymphocytes Tγδ, Tαβ, T CD8 + and NK cells: *MICA* and *MICB*. Our research confirms the differences in the genetic basis of type I and II psoriasis, confirms the strong association of type I psoriasis with the genes of the MHC I region. At the first time we have found the association of type I disease with *USP8* gene encoding enzyme involved in proteolysis, activation of EGFR and regulation of proliferation.

Citation: Nedoszytko B et al. Occurrence of polymorphic changes in the genome of psoriasis patients using the GWAS technique in the Polish population – analysis of preliminary research results. Eur J Transl Clin Med. 2019;2(Suppl.3):32-33.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):34

Ultra deep sequencing of colorectal tumours: heterogeneity of somatic mutations in the context of lymph node involvement

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Abstract

Intra-tumour heterogeneity (ITH) results from the acquisition of somatic mutations during tumour evolution. In spite of numerous research efforts, the implications of ITH are not fully understood nor applied for better diagnosis and therapy in cancer patients. We aimed to characterize genetic ITH of T3 size primary CRC tumours, using deep sequencing of 56 cancer-related genes. We compared lymph node positive with node negative patients using Renyi divergence between VAF (variant allele fraction) observed at different locations (peripheral and central regions of the tumour and healthy mucosa) and we have found that using this measure in the central region (p=0.028) we can distinguish node negative (NN) from node positive tumours (NP). Another important observation is that distribution of variants in tumour suppressor genes (TSG) differs between tumour fragments in central as well as in peripheral regions of NN and NP tumours. We also strongly support the idea that metastatic dissemination depends on clonal evolution driven by a subset of TSG – metastasis suppressor genes.

Citation

Wątor G, Seweryn M, Kapusta P, Świrta J, Wałęga P, Barczyński M, Wołkow P. Ultra deep sequencing of colorectal tumours: heterogeneity of somatic mutations in the context of lymph node involvement. Eur J Transl Clin Med. 2019;2(Suppl.3):34.

Transcriptomics in the service of anti-cancer therapy. The case of SIRT2 and melanoma

Marcin Ratajewski

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Abstract

Transcriptome profiling has been widely used to understand the heterogeneity of cancer cells, determinants of the response to therapy and even patient outcomes. Based on our previous research, we have identified sirtuin 2 (SIRT2) as potentially interesting target in therapy against melanoma. Thus, we downregulated SIRT2 expression in two melanoma cell lines representing vertical growth and metastatic phases. Detailed transcriptome analysis revealed that in cells with downregulated SIRT2, the expression of multiple genes encoding the tyrosine kinase proteins is affected. Among them were those identified as targets of multikinase inhibitor - dasatinib (e.g., EGFR, EPHA2, EPHB1, BTK, MAP3K4, MAP3K14). Indeed, cells with low expression of SIRT2 were more sensitive to dasatinib treatment compared with those with normal expression of this sirtuin. They also exerted impaired migratory function and altered phosphorylation status of downstream elements of the EGFR and EPHA2 pathways. In conclusion, we explained one of the possible resistance mechanisms to dasatinib and we propose usage of sirtuin 2 inhibitors in present regiments against melanoma to increase their efficiency.

Citation

Ratajewski M. Transcriptomics in the service of anti-cancer therapy. The case of SIRT2 and melanoma. Eur J Transl Clin Med. 2019;2(Suppl.3):35.

EUROPEAN JOURNAL OF TRANSLATIONAL
AND CLINICAL MEDICINE 2019;2(Suppl.3):36

Discovery and clinical applications of methylation biomarkers

Tomasz Wojdacz

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Abstract

DNA methylation is an enzymatic addition of methyl groups to cytosines in DNA strand. In general terms methylation of promoter downregulates gene expression. Regulation of the gene expression by this mechanism is one if the critical processes in development and tissue specification. At the same time, changes of normal methylation of genes contribute if not initiate malignant transformation. Thus, the phenotype of neoplasia (e.g. aggressiveness) to large extant depends on the methylation changes acquired during carcinogenesis. Identification of the physiological processes disrupted by methylation during carcinogenesis allows for the discovery of the new treatment targets and biomarkers for personalized medicine. The process of identification of the methylation changes typically begins with a study that utilizes a technology allowing comparison of the genome wide methylation pattern of pathologically changed tissues and their healthy counterpart. Discovered in those analyses, methylation changes are subsequently validated for the clinical utility. In my talk I will review methylation biomarker discovery process using examples from my research.

Citation

Wojdacz T. Discovery and clinical applications of methylation biomarkers. Eur J Transl Clin Med. 2019;2(Suppl.3):36

From biobanking to metabolomics analyses – evaluation of small molecules in different aspects of non-small cell lung cancer

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Abstract

Non-small cell lung cancer (NSCLC), the most common cancer in the world, is also characterized by the highest mortality rate. Although molecularly-targeted therapies for NSCLC patients are available, they often result in the development of drug resistance. Moreover, targeted mutations are present only in a limited number of patients. Therefore novel therapeutic targets and biomarkers (for early detection or prediction and evaluation of treatment efficacy) are needed for NSCLC patients. Metabolomics has the potential to achieve these goals. However, to obtain reliable metabolomics results, critical patients' medical and anthropometric data should be gathered. Moreover, the process of samples collection and storage should be properly planned and controlled. These crucial aspects of clinical metabolomics will be presented based on our experience from the MOBIT project. Additionally, some of the metabolomics results obtained within this project will be presented.

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Commercialization of research results based on the use of human biological material and data

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Abstract

The issue of commercialization of HBM(human biological material)-based scientific research results may be located in particular in two areas, namely the science sector and the medical sector, including the pharmaceutical one. With regard to the former, appropriate normative solutions of a technical and procedural nature were adopted on the basis of the Law on Higher Education. It should be noted that the relevant provisions relate to the course of the commercialization process as such and do not refer to the specific HMB issue. There are no similar regulations in the medical area.

Due to the lack of comprehensive legal solutions, it is necessary to refer to the general guidelines regarding the ban on commercialization of the human body. Nevertheless, there are some exceptions in this regard – it is possible to file patent applications based on HBM, in particular: I) an element isolated from the human body or otherwise produced by a technical method, including a sequence or a partial gene sequence, may be a patentable invention, even if the structure of this element is identical to that of a natural element, II) the blood and plasma in full composition or blood cells of human or animal origin are authorized without the authorization of an industrial process, excluding plasma processed in an industrial process. A number of HBM-based products authorized for marketing can be indicated, in particular: I) bone screws and acellular skin matrix II) collagen products, III) acellular skin matrix (which, although derived from human tissue no longer contains human cells),IV) fascia fragments injected from corpses, V) blood products, VI) products from live cell cultures, including human stem cells obtained from embryos or from adults, used for therapeutic or research purposes that may themselves become commercial goods , e.g. products from mesenchymal stem cells, VII) diagnostic products and VIII) non-medical uses of human tissue or products from human tissues, e.g. use of human collagen for cosmetic purposes.

Although, based on the case law of American courts, the donor of biological material is generally refused the right to ownership of this material, at the same time, it is possible to indicate the facts in which the will of the donor regarding the use of LMB has been taken into account.

Citation

Banasiuk J. Commercialization of research results based on the use of human biological material and data. Eur J Transl Clin Med. 2019;2(Suppl.3):38.

Molecular analysis of diet in three bat species of similar foraging tactics

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Abstract

Bats foraging on arthropods are perceived as food opportunists. Among bats, however, there are several main ways of food intake associated with its occurrence in the environment we classify them in “guilds”. One of these types of guilds are bats collecting food from the surface of leaves and trunks of trees and undergrowth. The food of bats representing this group is evenly distributed, and its availability in time is stable and predictable, so these species do not form large summer colonies. The body structure allows them to fly freely and maneuverably, which allows collecting, by means of the tail membrane or wing, victims sitting on plants.Until recently, the diet of bats was studied by morphological analysis of prey remains in their scats. This method usually does not allow to determine prey taxa below order or family level, thus making the more subtle mechanisms of resource partitioning among bat species undetectable.

Application of molecular methods allows to determine prey to the species level could give new insight into bat diet composition and show differences between species that hunt in a similar way. We present on our poster preliminary results of molecular diet analysis for three similar bat species, all foliage gleaners: Natterer’s bat Myotis nattereri, brown long-eared bat Plecotus auritus and Bechstein's bat Myotis bechsteinii. Guano pelets from 100 individuals were collected untill mist netting in six differetn localisation. We chose for further analysis 67 bat dropping and we isolated DNA from them. Isolation took place using a modified commercial procedure and produced very good results, the insulation failed with only a few samples. The primers used were selected in a way that allows good recognition of all groups of arthropods, both insects and arachnids. The first analyzes of the obtained sequences show us the descent to low taxonomic levels, which can help indicate specializations between the studied species. It is also interesting to jump over the taxonomic level and detect mites parasitizing plants or victims of scavenger beetles. We hope that further analysis will allow to indicate species in the composition of food constituting a strong separation of niches in seemingly similar species or will answer the question whether the phenology of insects can imitate ranges of occurrence of bat species

Citation

Bidziński K, Jankowska-Jarek M, Janik-Superson K. Molecular analysis of diet in three bat species of similar foraging tactics. Eur J Transl Clin Med. 2019;2(Suppl.3):39.

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Environmental monitoring during stem cells preparation in clean rooms

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Abstract

Environmental monitoring is a key element in clean rooms assuring low risk of the cell products contamination. The aim of this study was to qualitatively and quantitatively analyze the microbiological quality of the GMP laboratory environment. We present results of our 18 months environmental monitoring process during stem cells preparation. The microbiological analysis of 385 plates with microbiological infection were conducted and results of microbiological examination were collected. The plates were contaminated with 23 different microorganisms. The most commonly isolated were Bacillus spp., Micrococcus spp. and Staphylococcus spp. Almost all identified pathogens belong to group of physiological flora of human skin. Adhering to GMP rules and following the proposed procedures can be stated that such low level of contamination has no influence on the stem cells preparation in clean rooms. This is the reason why GMP laboratories are able to obtain uncontaminated specimen that can be used both in the clinic and for other biobank research purposes. This work was supported by STRATEGMED2/267976/13/NCBR/2015.

Citation

Bochyńska-Czyż M, Brzezicka J, Humięcka M. Environmental monitoring during stem cells preparation in clean rooms. Eur J Transl Clin Med. 2019;2(Suppl.3):40.

Biobanking of bone material – the case study of an craniosynostosis individual

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Abstract

Due to the variety of material type collected by biobanking facilities around the world, up to now, there is a lack of anthropological collection data and information availability in open directories. The bone tissue collected during the archaeological research, is an unusual research material that also states priceless information about 1/8 our identities and history. We present outcomes of the analysis of the bone material with unique features of an individual, with complex craniosynostosis – interpreted as the soldier, fallen in the fighting in the Bzura and Rawka region (1914-1915). Complex craniosynostosis is a disease most often interpreted as having congenital nature. According to estimates, about 10% of all craniosynostosis cases in today Poland, with the possibility of observing such changes on anthropological material is very rare. As the result of anthropological analyzes individual characteristics of the subject were determined The skeleton was interpreted as the remains of the male individual who died between 25-30 years old and he had unique special features such as significant deformation of the cranium. The shape of the skull of this individual, although without an extension of the anteroposterior dimension, is observed in the syndromes of Apert, Crouzon and/or Pfeiffer. In the case of the examined individual, a specific indication of any of these syndromes based on macroscopic examination was difficult due to facial damage and lack of finger bones. For this reason, based on the thesis, that the deformation of the skull did not significantly affect the functioning of the individual, it was decided to perform tests for Apert syndrome. The material collected for genetic testing included elements of the bone labyrinth, which is a source of DNA for sequencing in a low NGS method. Additional work was carried out to confirm the presence of SNPs associated with Apert syndrome in the FGFR2 (Fibroblast Growth Factor Receptor 2) receptor 2 gene. A variant of this gene – rs149200230 – was found in the analyzed sequence, which is pathogenic in nature, associated with a change in protein function.

Citation

Borówka P, Marchewka J, Królikowska K, Strapagiel D, Zalewska A. Biobanking of bone material – the case study of an craniosynostosis individual. Eur J Transl Clin Med. 2019;2(Suppl.3):41.

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Well mapped and supervised biological material life cycle as the key to the reliability of scientific research

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Abstract

In the era of biobanking, high-quality biological material samples have particular importance for science. Processing during biological material life cycle must be based on action in accordance with scientific, ethical and societal standards. To receive following assumption the biobanking procedure must be based on harmonized sample collection system, monitoring of transport temperature, as well as repeatable biological material processing. Crucial is to keep unified documentation to enable the samples make their way in a consistent and non-accidental manner from the patient to the place of material storage and further to the recipient. Obtained evidences in the form of unified documentation, e.g. reports from sample collection, records of transport temperatures, biological material processing protocols, confirm the quality of biological material, ensure their safety and reproducibility of the applied procedures. Defined, mapped and monitored biological 9/12 material life cycle results in continuous flow of samples in the biobank, thus ensuring the constant availability of high quality material for scientific purposes.

Citation

Burczyk M, Laskowski M, Wyderka M, Matera-Witkiewicz A. Well mapped and supervised biological material life cycle as the key to the reliability of scientific research. Eur J Transl Clin Med. 2019;2(Suppl.3):42.

Cryopreservation of the vimba bream (*Vimba vimba* L.) sperm

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Abstract

Cryopreservation protocols of genetic material such as fish sperm evolved from the second half of the twentieth century. In this group of animals effective methods of sperm freezing have been developed in about 250 species. Currently, methods are improved using extenders based on a simple composition, containing only sugar and a cryoprotectant. Recent studies show that beside DMSO, methanol is an appropriate cryoprotectant to freeze fish sperm.

The purpose of the present work was to improve the effectiveness of cryopreservation of the vimba sperm using methanol and optimizing freezing rate by high altitude of straws above the liquid nitrogen.

Selected sperm of vimba with high motility rate was frozen after mixing with cryomedium containing 10% methanol, 0.3 M glucose, 30 mM Tris (pH 8), in a ratio of 1:7. Three minute equilibration was applied. Sperm was frozen in 0.25 mL straws on frames floating 2, 3, 4 and 5 cm on liquid nitrogen in styrofoam box. The freezing time was 3 minutes. The straws were thawed in a water bath at 40°C for 5 seconds. The efficacy of cryopreservation protocols was assessed by analyzing spermatozoa motility parameters using a computerized-assisted sperm analysis system (CASA).

Thawed sperm has lower percentage of motile spermatozoa than that of fresh sperm. High-altitude freezing variants did not differ significantly motility rate range from 49.3 to 59.2%. In 2- and 4 cm high variants the total velocity of frozen spermatozoa was similar to that of fresh sperm. Values of LIN, STR, ALH, BCF parameters and duration of spermatozoa movement did not differ significantly in fresh and frozen/thawed sperm.

The optimal freezing rate of vimba sperm in 0,25 mL straw was achieved at 4 cm above liquid nitrogen.

Citation

Dziewulska K, Leśniańska J. Cryopreservation of the vimba bream (Vimba vimba L.) sperm. Eur J Transl Clin Med. 2019;2(Suppl.3):43.

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Distribution and frequency of the Y chromosome haplogroups in Polish population

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Abstract

The variability of Y chromosome in Polish population was analyzed over the years, in the studies regarding to different regions of Poland and in the studies of larger population groups, including Poland. Furthermore, the populations of Lithuanian, Belarussian and Tatar minorities living in Poland have also been studied. Most of the studies was based on the analysis of short tandem repeats (STR). The present study was conducted using microarray technology which allowed to study single nucleotide polymorphisms (SNP) located on Y chromosome to describe the genetic structure of male population. The comprehensive analysis of Ychromosome variability, based on the data from almost 3000 individuals from all polish voivodeships and most counties and with the use of clustering as the additional method of population grouping has been performed. By far the most frequent haplogroup in polish population is R which is 6 / 8 typical for West Eurasian populations. Less common but with still significant are I, N, E, J and G haplogroups. The observed differences between regions may indicate historical and cultural influences. Founding: DIR/WK/2017/01 and POIG Grant 01 01 02 10 005 08.

Citation

Grochowalski Ł, Urbanowicz M, Jarczak J, Borówka P, Lorkiewicz W, Sobalska-Kwapis M, Marciniak B, Słomka M, Strapagiel D. Distribution and frequency of the Y chromosome haplogroups in Polish population. Eur J Transl Clin Med. 2019;2(Suppl.3):44.

Association between urine phthalate metabolites and thyroid hormones

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Abstract

Phthalates are the group of widely used chemicals as plasticizers, adhesives, stabilizers, industrial solvents, insect repellent, in personal care products, textiles and pharmaceuticals. People are exposed to phthalates via inhalation, ingestion or dermal absorption as results of contamination of food chain, inhalation of contaminated house dust or occupational exposure. Phthalates are quickly metabolized and excreted with urine. In a limited number of studies was shown that exposure to some phthalates may be associated with altered thyroid function.

Aim of this study was to asses 6 phthalate metabolites levels in urine of healthy inhabitants of Poland (n = 111) and using multivariate linear regression analyses try to find any association of phthalate exposure with thyroid hormone levels. In investigated population we found statistically significant relationship between MEHP and MBzP concentrations with TSH levels.

Citation

Gromadzińska J, Mikołajewska K, Zieiński M, Szewczyńska M, Wasowicz W. Association between urine phthalate metabolites and thyroid hormones. Eur J Transl Clin Med. 2019;2(Suppl.3):45.

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Consents for biobanking during the social campaign Wyzwanie100

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Abstract

Human papilloma virus (HPV) cause nearly 100% of high-grade cervical dysplasia and cervical cancer. To date, approximately 200 types have been identified and some of them has been classified as oncogenic – types 16 and 18 with the highest oncogenic potential. In 2018, Cellgen Laboratory performed screening diagnostic tests as part of a social campaign Wyzwanie100. The aim was to examine women from Wrocław and its environs for HR-HPV (high-risk HPV) infection. Material for the test were cervical swabs, obtained by self-sampling device Evalyn Brush (Rovers). We performed an analysis showing the number of women consenting for biobanking of biological material. Data set for analysis comes from over 700 randomly applying women over 25 years. The results are satisfactory because most women were interested in storing the collected material and thus participating in scientific research.

Citation

Ilnicka A, Marciniak P, Fuławka Ł. Consents for biobanking during the social campaign Wyzwanie100. Eur J Transl Clin Med. 2019;2(Suppl.3):46.

Genetic determinants of the red hair occurrence in the Polish population based on GWAS

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Abstract

Red hair occurs naturally in 1-2% percent of the human population. GWAS leads to the identification of many loci associated with variation in human hair pigmentation in different populations. The aim of the following GWA study was to identify the SNP profile which may be a determinant of red hair in Polish people. Our research was carried as part of the POPC project (Operational Programme Digital Poland for 2014-2020) - that aims to collect and share 1/2 data from the field of genetics and anthropology of the Polish Europeans. A total of 5450 DNA samples, registered as POPULOUS collection at the Biobank Lab of The University of Łódź, were genotyped using the 24x1 Infinium HTS Human Core Exome. To identify SNPs associated with red hair in the Polish population, we performed case-control association analysis in the group of people with red hair (N = 57) versus dark and blond (N = 5393) using PLINK 1.07 toolset. From 280 935 analyzed SNPs, 10 were statistically correlated with red hair ($p < 2.71e-8$, BONF < 0.007). The range $\chi2 = 117.7–30.91$, $p = 2.02e-27–2.71e-8$ were observed for rs12931267 (FANCA gene), rs1805007 and rs1805008 (MC1R), rs258322 (CDK10), rs8051733 (DEF8), rs4785763 (AFG3L1P), rs3743860 (FANCA), rs4785759 (AFG3L1P), rs8058895 (FANCA) and rs3743829 (GAS8 gene). Both SNPs of the MC1R gene, rs1805007 and rs1805008, are missense mutations R>C and R>W, respectively. Our results help to understand the genetic basis of the unique red hair color and will improve the bioinformatic tools used to predict hair color based on the genotype. Projects no. POIG Grant 01.01.02-10-005/08 TESTOPLEK Projects no. POPC.02.03.01-00-0012/17 eCzlowiek.pl

Citation

Janik-Superson K, Sobalska-Kwapis M, Słomka M, Marciniak B, Strapagiel D. Genetic determinants of the red hair occurrence in the Polish population based on GWAS. Eur J Transl Clin Med. 2019;2(Suppl.3):47.

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Genetic diversity of Polish male population based on the analysis of Y chromosome

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Abstract

The aim of the present study was to define the variability of Y chromosome in Polish male population and to visualize the genetic relations between Poles. For the first time, the study of Polish population was conducted on such a large number of individuals (ar. 3000) representing administrative units of both levels of local administration in Poland (voivodeships and counties). Additionally, clustering was used 4 / 8 as a method of population subdivision. Performed genetic analysis, included FST and MDS plot. MDS plot clearly shows that almost all Polish population was seen as clustered together with only Łódź and Lower Poland different from the rest of Polish population and different from each other as well. Another MDS plot was constructed to visualize the relationships between generated clusters. In this case, a large group of almost all clusters was observed together, while clusters number: 8 (Brzeziny, Tomaszów Mazowiecki, Łowicz and Piotrków in Łódź voivodeship) and 12 (Kraków and Tatra region in Lesser Poland voivodeship) were separated. All of these allow us to treat Polish population as very homogenous. This comprehensive analysis confirmed that Polish population is characterized by the high homogeneity with the only slight genetic differences at the regional level. Using the clusters as the alternative for the intrastate administrative divisions, allowed to look deeper into the structure of the population with the real borders between the regional populations marked on the map of Poland. The differences between voivodeships could be misleading until the analysis of clusters is carried out. The present study together with the published data about mtDNA variability could be the basis for the further research forming the connection between the modern and ancient times of Poland considering human migrations or resettlements as well as historical and cultural influences. The study was financed by Polish Ministry of Science and Higher Education no. DIR/WK/2017/01: ”Biobank network in Poland, within the BBMRI-ERIC Research Infrastructure of Biobanks and Biomolecular Resources”. POPULOUS collection was financed by the Polish POIG Grant 01 01 02 10 005 08 TESTOPLEK from the European Regional Development Fund

Citation

Jarczak J, Grochowalski Ł, Urbanowicz M, Borówka P, Sobalska-Kwapis M, Słomka M, Lorkiewicz W, Marciniak B, Strapagiel D. Genetic diversity of Polish male population based on the analysis of Y chromosome. Eur J Transl Clin Med. 2019;2(Suppl.3): 48.

Screening for celiac disease of group Świętokrzyskie voivodeship inhabitants

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Abstract

The project was created due to the lack of keeping the national register of people suffering from celiac disease and the fact that the total number of patients with celiac disease (overt and latent) is not fully known and epidemiological data are only estimates. Planned number of study participants is 1000 inhabitants of the Świętokrzyskie voivodeship in the age bracket between 3- 5 and 12-15 years old for the occurrence of celiac disease (gluten intolerance). Project was carried out in cooperation with the First Clinic of Paediatrics of the Provincial Integrated Hospital in Kielce, outpatient clinic SPZOZ Nowiny, Local Government Center for Primary Health Care in Chęciny and outpatient clinic GALUS s. c. in Sukowo. 1/5 So far 312 people have used the project, all received results were consulted with the Medical Supervisor of the project. In the case of 42 project participants, non-standard results were observed. Children were referred for in-depth diagnostics for celiac disease.

Citation

Karpeta M, Twardowska E, Zagórska-Szlufik P, Sularz A, Sychowska I, Niebudek-Jach K. Screening for celiac disease of group Świętokrzyskie voivodeship inhabitants. Eur J Transl Clin Med. 2019;2(Suppl.3):49.

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Ethical and legal aspects of the functioning of breast milk banks

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Abstract

Female milk banks have been regularly established in Poland for several years. They are special institutions. The biological material stored in them can be transferred only by a unique group of donors. Of course, this is only about women whose body produces milk after delivery. The functioning of similar facilities is recognized by the public, representatives of state authorities, medical experts and patients themselves.

Female milk is certainly a "unique substance". Scientists are still working on a material whose properties would most closely resemble the composition of the milk that mothers have. Giving women their own milk and depositing it in a bank has a unique meaning. This substance has not only nutritional properties. It is commonly pointed out that this milk is a unique medicine, especially valuable during nutrition of premature babies, for example. Can undertaking similar actions result in significant ethical and legal dilemmas? Can access to breast milk be considered as exercising the patient's right to health services corresponding to the current state of medical knowledge? The proposed speeches aim to answer similar questions.

Citation

Kmieciak B. Ethical and legal aspects of the functioning of breast milk banks. Eur J Transl Clin Med. 2019;2(Suppl.3):50.

Central Biobank and its impact on the cooperation agreements, projects receiving and development of research area

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Abstract

Biobanks are known as organizations that strongly support research in the field of biomedicine and biotechnology. Their work results on effective exchange of biological material and data between units which collect samples and researchers. Centralized management of flow process concerning material and data from clinics to the researchers is the mission of biobanks that operate in large institutions, such as universities. Organization, regulation and supervision over the process of material and data collection in the clinics, its safety storage and distribution are the key to achieve this goal. Biobanking process centralization allows for more efficient usage of bioresources and ensuring their highest quality in the same time. It directly contributes to the increasement of the number of scientific research collaboration, including genomics, transcriptomics, proteomics, and metabolomics. Biobanks are crucial determinant which 7/12 intensify the cooperation between scientists. Biobanks activity in universities has a positive impact on research ability, resulting in participation in multi-center international research projects.

Citation

Laskowski M, Wyderka M, Gleńska-Olender J, Zagórska K, Matera-Witkiewicz A. Central Biobank and its impact on the cooperation agreements, projects receiving and development of research area. Eur J Transl Clin Med. 2019;2(Suppl.3):51.

EUROPEAN JOURNAL OF TRANSLATIONAL
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Circadian gene polymorphisms and breast cancer susceptibility

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Abstract

Breast cancer (BC) is a major civilizational problem, manifested by continuously increasing morbidity and mortality. Core 2/8 circadian genes may play an important role in cancer development and progression. To evaluate the effects of single nucleotide polymorphism (SNP) in circadian genes in breast cancer (BC) risk, six functional SNPs were genotyped in 321 BC patients and 364 healthy women. Selected SNPs were analyzed for the risk of BC, progression and the influence to genes expression in BC tissue pairs to demonstrate the functionality of genetic variants. The study showed a relationship between variant genotype of CLOCK rs12505266 and BC risk, especially estrogen-negative tumors. Additionally, we have observed an increased risk of estrogenpositive tumors under variant genotype of rs11894491 in PER2. We have demonstrated altered gene expression of CRY2, PER1, PER2, PER3 according to particular genotypes in BC tissue pairs. Our findings support the hypothesized role of circadian genes in breast carcinogenesis and indicate probable biomarkers for breast cancer susceptibility. This work was supported by the National Science Center (Grant No. 2014/15/N/NZ5/01671) and internal grant IMP 14.5.

Citation

Lesicka M, Jabłońska E, Wieczorek E, Pepłońska B, Gromadzińska J, Seroczyńska B, Kalinowski L, Skokowski J, Reszka E. Circadian gene polymorphisms and breast cancer susceptibility. Eur J Transl Clin Med. 2019;2(Suppl.3):52.

International cooperation between oncological Biobanks – three years' experience

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Abstract

International cooperation poses a challenge due to the many barriers. But it also opens great opportunities to exchange the knowledge and experience. We can take advantage of these capabilities also in the field of biobanking. Medical University of Bialystok (MUB), as a leader of MOBIT Project, aimed at establishing of Oncological Biobank in cooperation with global biobanking company Indivumed GmbH. Three Study Nurses and one biobanking coordinator were trained in the Indivumed's headquarter in Hamburg, Germany for two weeks and three months, respectively. As well, Standard Operating Procedures and Forms were prepared together with Indivumed's experts. In order to maintain control over quality and type of samples and data, coordinator is reporting the current biobanking status every two weeks. Indivumed also performs audits in MUB every six months to check if there is a need for improvements. Part of tissue and liquid samples collected in MUB are shipped to Hamburg for quality control and data are uploaded into IndivuNET system. Cooperation between MUB and Indivumed opened the possibility to establish biobank for collection of high-quality samples along with comprehensive database.

Citation

Michalska-Falkowska A, Reszeć J, Kiśluk J, Chludzińska S, Modzelewska P, Nikliński J. International cooperation between oncological Biobanks – three years' experience. Eur J Transl Clin Med. 2019;2(Suppl.3):53.

EUROPEAN JOURNAL OF TRANSLATIONAL
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Genetic determinants of skin and hair pigmentation in Polish children

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Abstract

The complex genetic system of pigmentation determines the phenotype that undergoes modifications through human ontogenesis. The observed pigmentation variation, both external- and intrapopulational, is the reason for research aimed at understanding the biological, evolutionary and genetic basis of this phenomenon. Advances in GWAS make it possible to identify gene variants involved in the pigmentation of different populations. This study focuses on examining the association of 34 known SNPs with skin and hair pigmentation in the group of Polish children using DNA microarrays as a genotyping method. The statistical analyzes allowed to detect the relationship of selected polymorphisms with the pigmentation parameters in terms of quantity for the skin and hair as well as the categorized hair colour. In addition, a covariance analysis was performed considering the significant influence of age and sex in the studied group. Based on the linear regression results, a significant effect of 6 SNPs (rs12203592 in 7/8 IRF4, rs7174027 and rs7495174 in OCA2, rs8028689 in HERC2, rs1805008 in the MC1R and rs1393350 in the TYR) for the skin pigmentation and 4 SNPs (rs1129038, rs12913832 and rs1667394 in HERC2 and rs7195066 in MC1R) for the hair colour were confirmed in the examined group. The values of the standardized correlation coefficient indicated a weak association of selected variants with spectrophotometric colour parameters. The obtained results of logistic regression for the categorized hair colour showed that 6 polymorphisms affect the probability of bright hair (rs8007923 in KIF26A, rs1667394 in HERC2, rs258322 and rs7195066 in MC1R, rs12896399 in SLC24A4 and rs12203592 in IRF4). The obtained results allowed to evaluate the association between particular genetic variants and pigmentation traits among Polish children.

Citation

Mozga P, Sobalska-Kwapis M, Żądzińska E, Sitek A, Strapagiel D. Genetic determinants of skin and hair pigmentation in Polish children. Eur J Transl Clin Med. 2019;2(Suppl.3):54.

Survey design method as an element of building the Polish Biobanking Network

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Abstract

Polish Biobanking Network (PBN) is creating to make cooperation between polish biobanks easier, what can be achieved by acquiring and then sharing information about biobanks and their collections. The type and scope of collected information should be unified, therefore most institutions responsible for biobanking material are based on *Minimum Information About Biobank data Sharing* (MIABIS) guidelines, this also applies also to BB-MRI-ERIC and BBMRI.pl. The aim of this work is to show how to design surveys that enable obtaining relevant information about biobanks.

Based on MIABIS two surveys were formed – First gives general information about biobanks, its localization, type of collected material. The second, more detailed one, was divided on five parts: biobank and its structure, collected material, sharing material, informatics system, final questions. Through appropriate recognition of the biobanks topic and correctly asked questions, significant information could be obtained. Based on the obtained results and their analysis, a full vision of Polish biobanks can be received.

Citation

Paluch A, Chróścicka A. Survey design method as an element of building the Polish Biobanking Network. Eur J Transl Clin Med. 2019;2(Suppl.3):55.

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Oxidative DNA damage and repair in Rheumatoid arthritis – a correlation with the key BER genes polymorphisms

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Abstract

Rheumatoid arthritis (RA) is a systemic, inflammatory disease of the joints and surrounding tissues. RA manifests itself with severe joint pain, articular inflammation and oxidative stress. RA is associated with certain types of cancer. We have assumed that increased susceptibility to cancer of RA patients may be linked with genomic instability induced by disturbed DNA repair of oxidative DNA lesions by BER. In the present work we determined the level of basal and oxidative DNA damage and the kinetics of removal of DNA damage induced by tert-butyl hydroperoxide in peripheral blood mononuclear cells (PBMC) of 30 RA patients and 30 healthy individuals. The data from DNA damage and repair study 5/11 were correlated with the genotypes of functional polymorphisms of the key BER genes including XRCC1, hOGG1, UNG, SMUG1, TDG, MBD4, MUTYH. DNA damage and repair were evaluated by alkaline single cell gel electrophoresis (comet assay). The genotypes of the polymorphism were determined by TaqMan SNP Genotyping Assays. We observed an association between RA occurrence, impaired DNA repair in PBMC and the polymorphisms of XRCC1, and hOGG1 genes. Therefore, our result suggest that polymorphism of the XRCC1, and hOGG1 genes may be linked with RA by the modulation of the cellular response to oxidative stress and these polymorphisms may be a useful additional marker in this disease along with the genetic or/and environmental indicators of oxidative stress. Acknowledgement: This work was supported by the National Science Center (Poland) - UMO-2017/25/B/NZ6/01358.

Citation

Popławska M, Pietrowska E, Brzezińska O, Galita G, Makowska J, Popławski T. Oxidative DNA damage and repair in Rheumatoid arthritis – a correlation with the key BER genes polymorphisms. Eur J Transl Clin Med. 2019;2(Suppl.3):56.

Motivations and trust of potential donors toward population biobanks in relation to transfers of biological samples and biodata

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Abstract

The project to create the framework for a national genomic biobank for the needs of scientific research in Poland requires an assessment of the level and quality of trust of potential donors.

The aim of the nationwide questionnaire survey carried out using CATI (computer-assisted telephone interviewing) was to study the motivation and trust of potential donors toward population biobanks in relation to transfers of biological samples and biodata. The study was conducted in 2018 (BST Group) on a sample of 1,100 people (women 52.3%, men 47.7%) representative of the Polish population in relations to gender structure, age groups and place of residence.

Regarding the question about the types of benefits to give biological samples to a biobank, the vast majority of respondents pointed to the possibility of making genetic testing for disease risk (45.8% - definitely yes, 33.1% – rather yes) and performing genetic genealogical research regarding the origin of ancestors (37.7% - definitely yes, 29.2% – rather yes). When respondents were asked about the type of institution to which hypothetically biological samples and biodata could be transferred by the population biobank without the need for additional donors' informed consent, the majority of them indicated Polish universities (definitely yes or rather yes – a total of 68.1%), universities in EU (58.4%), pharmaceutical companies conducting research in Poland (55.4%) and universities in the USA (54.7%).

Citation

Sak J, Pawlikowski J. Motivations and trust of potential donors toward population biobanks in relation to transfers of biological samples and biodata. Eur J Transl Clin Med. 2019;2(Suppl.3):57.

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Genetic diversity of Polish population based on the analysis of mitochondrial DNA

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Abstract

The aim of the present study was to define the mtDNA variability of Polish population and to visualize the genetic relations between Poles. For the first time, the study of Polish population was conducted on such a large number of individuals (5852) representing administrative units of both levels of local administration in Poland (voivodeships and counties). Additionally, clustering was used as a method of population subdivision. Performed genetic analysis, included FST and MDS plot. A MDS plot, on the basis of the pairwise FST values, was constructed to visualize the relationships between voivodeships. A group including Lublin, Lesser Poland, Greater Poland, Mazovia, Silesia, Kuyavian-Pomeranian, Opole, Lubusz, Subcarpathian, Lower Silesian and Podlaskie was observed while Łódź, West Pomeranian, Pomerania, Warmian-Mazurain, Holy Cross were observed as separate. Another MDS plot was constructed to visualize the relationships between generated clusters. In this case, a large group of almost all clusters was observed together, while clusters number: 16 (Rawa and Opoczno regions), 18 (Płock and Sierpc Lands), 23 (Kraków county), 31 (Mazovia region, counties Wołomin and Wyszków), 34 (northern Greater Poland, Szamotuły and Trzcianków county), 41 (Łowicz, Brzeziny and Tomaszów counties in Łódź voivodeship), 47 (Western Kuyavia), 49 (Ełk and Grajewo regions), 64 (northern Mazovia) and 71 (Choszczno and Drezdenko counties) were outside of this group. Although the level of differentiation within the Polish population was found to be low, the existing genetic differences can be explained well with geographic distances. Using a large set of data, it was shown that Poles can be considered as genetically homogenous but with slight differences, highlighted at the regional level. The structure of our study allowed us to confirm that intrastate administrative divisions are artificial formations and do not reflect the genetic diversity of specific populations. Spatial information-based clusters are more adequate and in similar studies, researchers should consider grouping available samples based on geographic location, enhancing the quality of analysis in comparison to division into voivodeships and counties.

The study was financed by Polish Ministry of Science and Higher Education no. DIR/WK/2017/01: Biobank network in Poland, within the BBMRI-ERIC Research Infrastructure of Biobanks and Biomolecular Resources. *PPOPULOUS collection was financed by the Polish POIG Grant 01 01 02 10 005 08 TESTOPLEK from the European Regional Development Fund.*

Citation

Słomka M, Jarczak J, Grochowalski Ł, Marciniak B, Lach J, Sobalska-Kwapis M, Lorkiewicz W, Pułaski Ł, Strapagiel D. Genetic diversity of Polish population based on the analysis of mitochondrial DNA. Eur J Transl Clin Med. 2019;2(Suppl.3):58.

Determination of the lipid profile of the inhabitants of Świętokrzyskie voivodeship

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[2] BBMRI.pl Consortium

Abstract

The determination of the lipid profile of the inhabitants of Świętokrzyskie voivodeship is carried out as a screening test. Its main purpose is to detect disorders and deviations in the early stage, among a diverse group of people, at the stage where existing abnormalities do not manifest any symptoms or when the symptoms are mild enough that the examined participant did not report to the doctor for this reason. Conducting research on a large number of participants will create a lipid profile of the region's population. This will enable the identification of high-risk groups and the early detection of possible irregularities. The obtained results will facilitate the development of a preventive and informational strategy in order to raise awareness of a healthy society. The project was carried out in cooperation with the Clinic of Gynecology and Obstetrics of the Provincial Integrated Hospital in Kielce, Saint Rafał Provincial Specialistic Hospital in Czerwona Góra and Gameta Infertility Clinic in Podzamcze. As part of the project, 650 people were examined. Patients were screened for cardiovascular biomarkers (total cholesterol, triglycerides, HDL, LDL).

Citation

Sularz A, Rozmianiec K, Zagórska-Szlufik P, Niebudek-Jach K, Sychowska I, Karpeta M. Determination of the lipid profile of the inhabitants of Świętokrzyskie voivodeship. Eur J Transl Clin Med. 2019;2(Suppl.3):59.

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Assessment of vitamin D concentration in blood, among inhabitants of the Świętokrzyskie voivodeship

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[1]Biobank Świętokrzyski Regional Science and Technology Center, Podzamcze, Chęciny, Poland
[2] BBMRI.pl Consortium

Abstract

The aim of the project is to assess the supply of vitamin D among the inhabitants of the Świętokrzyskie voivodeship. Current research studies indicate a common deficiency of vitamin D. The obtained results will allow to determine the factors affecting the level of vitamin D and provide information on the extent to which vitamin D deficiency affects residents of the Świętokrzyskie voivodeship. The role of vitamin D in the body is invaluable. Its shortages lead to i.a. osteoporosis, development of obesity, cardiovascular disease, hypertension, diabetes, cancer or autoimmune diseases and can also accelerate the aging process. The project is carried out in cooperation with the Regional Center for Blood Donation and Blood Treatment in Kielce. Since the beginning of the project, ie from March 2018, 1850 inhabitants of our province 4/5 have used the possibility of free check of vitamin D concentration.

Citation

Sychowska I, Knap S, Zagórska-Szlufik P, Niebudek-Jach K, Sularz A, Karpeta M. Assessment of vitamin D concentration in blood, among inhabitants of the Świętokrzyskie voivodeship. Eur J Transl Clin Med. 2019;2(Suppl.3):60.

Association studies between selected chromosomal regions 1q21.3, 5q21.3, 14q21.2 and 17q21.31 with the number of offspring in Poles – analysis of data from the POPULOUS database

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Abstract

This reproduction study attempts to reproduce results from two studies: (1) *A common inversion under selection in Europeans* published by Stefansson et al. (2005) and (2) *Genome-wide analysis identifies 12 loci influencing human reproductive behaviour* published by Barban et al. (2016). Both publications indicated associations between single nucleotide polymorphisms (SNPs) and the number of children per family (NEB). The aim of this study was to confirm hypotheses concerning association between various genetic alterations with NEB. Saliva from subjects was collected, isolated and genotyped by the Biobank in Łódz, and the data was transferred to Pomeranian Medical University. Four different statistical regression models were used. Each statistical model was performed on eight differently selected age/gender groups for four genetic models. None of the investigated regions showed association between SNPs in the different chromosomal regions with NEB. Conclusion: Bioinformatic tools have been correctly implemented, no association was found between NEB and SNPs within chromosome regions 1q21.3, 5q21.3, 14q21.2 and 17q21.31, in the entire geographical region of the Polish population.

Citation

van de Wetering T, Clark J, Ciechanowicz A, Strapagiel D. Association studies between selected chromosomal regions 1q21.3, 5q21.3, 14q21.2 and 17q21.31 with the number of offspring in Poles – analysis of data from the POPULOUS database. Eur J Transl Clin Med. 2019;2(Suppl.3):61.

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Cell banking in the manufacture of advanced therapy medicinal products

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Abstract

Regenerative medicine plays an increasingly important role in developed countries due to the aging of the population. Newlydeveloped advanced therapy medicinal products (ATMP) aim to restore the normal functioning of damaged organs. Facilities producing ATMP products are required by Polish and EU law to have laboratories with the authorization / status of *Cell and Tissue Bank*. Legal requirements relate to many aspects, including appropriate technical and apparatus infrastructure enabling documentation of monitoring environmental and biological factors and collecting reports on the results of tests carried out. In addition, all production work in the laboratory must be carried out in accordance with the implemented standard operating procedures (SOP), consistent with the principles of Good Manufacturing Practice (GMP). The objective and scope of the Center for Research and Implementation of Advanced Cellular Therapies currently includes the production of ATMP products containing: glial olfactory cells and fibroblasts (*Wrocław Walk Again*), stem cells isolated from dental pulp, and fibroblasts isolated from oral connective tissue.

Citation

Wiatrak B, Gębarowski T. Cell banking in the manufacture of advanced therapy medicinal products. Eur J Transl Clin Med. 2019;2(Suppl.3): 62.

Psychological factors modifying various dimensions of biobanking attitudes

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Abstract

Development of biobanks for scientific purposes is one of the greatest challenge for progress of biomedical science. However, in the public opinion, scientific biobanks are poorly recognizable, which can make difficult collaboration with research participants and complicate collection an adequate number of samples. The personal decision to give biospecimen to the biobank can be modified not only by sociodemographic variables (such as gender, age, level of education), but also by individual psychological factors (including personality, convictions concerning the determinants of human nature, temporal orientation or religiosity). The aim of the research was to analyze psychological variables that may play a role in the potential donors decisions. In the first step various dimensions of the social perception of biobanking phenomenon was described (i.e. opportunity, threat, altruistic action). In the second step the relationship between specific ways of perceiving biobanking and selected psychological variables was assessed. The research questionnaire include standardized tools and scales such as: BANKS scale (Wells et al., 2014), TIPI for personality (Gosling et al., 2003), Nature-Culture Questionnaire (Żmuda-Trzebiatowska, 2008), Perception of Time Scale (Cybis et al. , 2012) and DUREL (Koenig et al., 2010). Results indicate that some psychological variables of respondents' relate to more positive decision regarding donation to biobanks (e.g. openness for new experiences, conviction about determination of human nature, future orientation and religiosity) whereas others are connected with more negative decision.

Citation

Wiechetek M, Pawlikowski J. Psychological factors modifying various dimensions of biobanking attitudes. Eur J Transl Clin Med. 2019;2(Suppl.3):63.

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AND CLINICAL MEDICINE 2019;2(Suppl.3):64

Research inland versus abroad, in public versus commercial institutions – what are Polish parents fears about pediatric biosamples sharing?

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Abstract

Specific ethical and legal issues make pediatric biobanking challenging. In practice, however, parents willingness to donate own child biological material seems to play crucial role in the final collection, use and sharing of biosamples. Lack of knowledge, fears and superstitions may lead parents to disagreement or limited consent to pediatric research. 1/6 A nationwide survey about pediatric biobanking is taking place in Poland. General positive assessment of biobanking and agreement to relatively wide scope of use of the biosamples have been already provided by almost 500 parents. However, sharing of pediatric biosamples turned out to be very sensitive aspect. Parents would prefer local use of the biosamples in public research centers (80%). They would be reluctant (40%) to transfer biological material abroad or to private/commercial companies. Parents opinion on limited sharing of pediatric biosamples has to be taken into account by biobanks in Poland. It seems, on the other hand, that general educational campaign about pediatric biobanking is needed to promote its legitimacy.
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Citation

Wójtowicz J, Gietka P, Chróścicka A. Research inland versus abroad, in public versus commercial institutions - what are Polish parents fears about pediatric biosamples sharing? Eur J Transl Clin Med. 2019;2(Suppl.3):64.

Biobank certification as a confirmation of the quality standards fulfillment and its influence on reliability of scientific research

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Abstract

The high quality of biological material and research which are planning to be conducted on it, is mainly based on the 1/5 process management according to the highest standards. The implementation of unified procedures by using of the identical processing methods and samples supervision, guarantees the same quality of obtaining biological material and data. Presented proceeding gives the opportunity to trace sample life cycle and obtain reliable test results. Thus, it is possible to perform the experiment and also compare and confirm the results which have been previously conducted. Moreover, acting based on Quality Management System increases the trust of scientific community and also protects the donor's rights. Wrocław Medical University Biobank obtained ISO 9001:2015 certificate and is preparing for ISO 20387: 2018 accreditation. Thus, Biobank ensures that biological material and data possess high quality and guarantee the reproducibility and reliability of planning research wherever it will be used.

Citation

Wyderka M, Laskowski M, Gleńska-Olender J, Zagórska K, Matera-Witkiewicz A. Biobank certification as a confirmation of the quality standards fulfillment and its influence on reliability of scientific research. Eur J Transl Clin Med. 2019;2(Suppl.3):65.