



RAZEM
MOŻEMY
WIĘCEJ



Presentation of the NBIA Poland Association

Maciej Cwyl

President of the NBIA Poland association



STOWARZYSZENIE
NBIA
P O L S K A



Characteristics of the members of the NBIA Poland association - after the meeting on 01.10.2022.

Basic information about the association:

- The association was registered in February 2019. We are the youngest of the associations in the NBIA Alliance group.
- From the latest research - NBIA Poland survey (research supported by CoA Therapeutics), we are one of the largest associations in terms of the number of members.

Lp.	Members of the NBIA Poland association	
1.	Liczba członków ogółem	93
2.	MPAN	53
3.	PKAN	20
4.	BPAN	14
5.	PLAN	2
6.	Bez badań genetycznych	2
7.	Bez chorego	1
8.	Do diagnozy prawd.MPAN	1



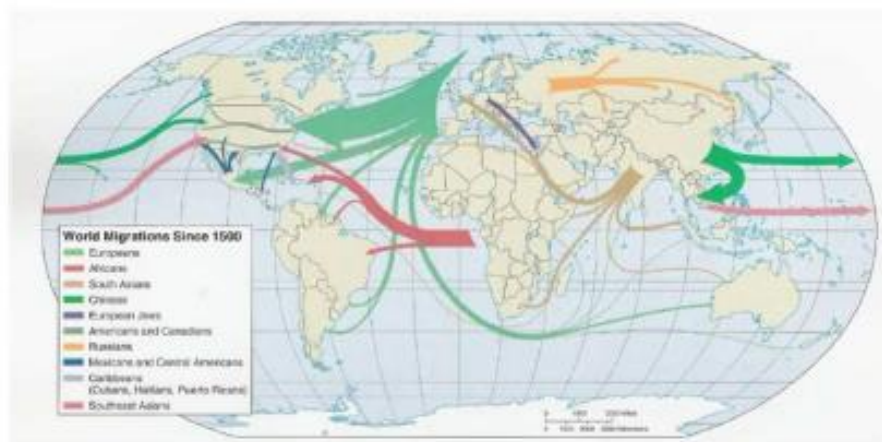
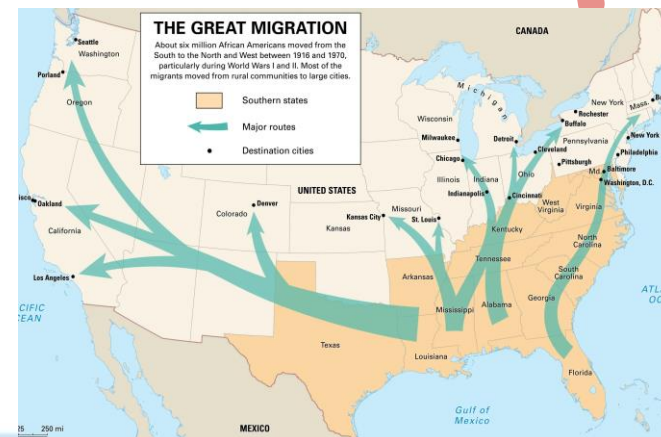
Selected information from research - surveys of the NBIA Poland association (with the support of CoA Therapeutics)

Please do not copy or disseminate the information presented - let us respect privacy - the materials will be published in the scientific article of the research team and the NBIA Poland association in 2022.

Main directions of population migration (selected maps) in the last ten centuries

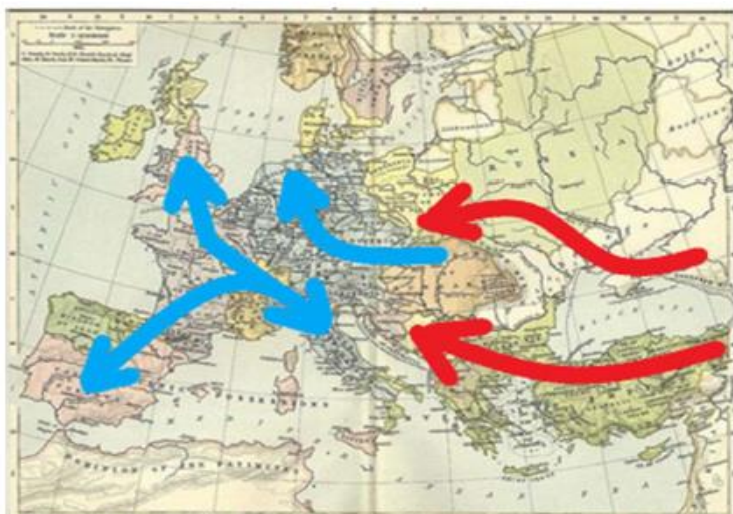


We check - where our ancestors come from :).



Rys. 2 Światowe migracje po XVIw. Migracje z Europy oznaczono kolorem zielonym.

Rys. 3. Zakres wojen Napoleońskich XIX w

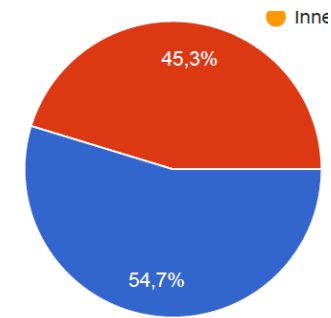


Rys. 1 Kierunki migracji w Xv. Główny kierunek napływu ludności (kolor czerwony) oraz migracje wewnętrzne w Europie.

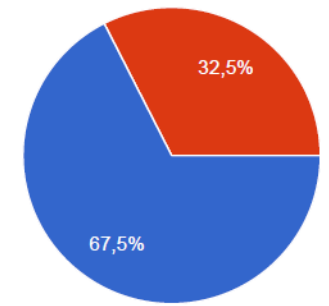


Selected results from the NBIA Poland survey

- Split of patients into women (blue), men (red).



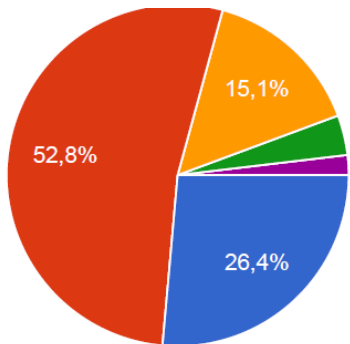
Polish-speaking group



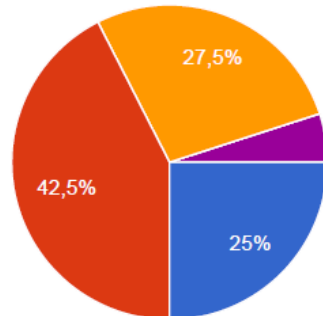
English-speaking group

- Percentage shares of particular varieties

Polish-speaking group

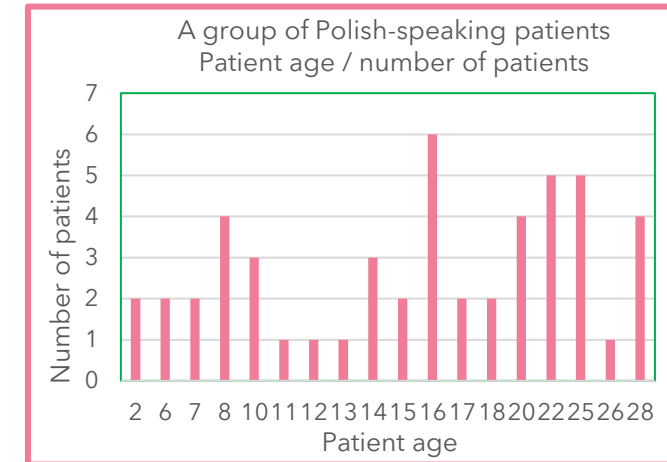
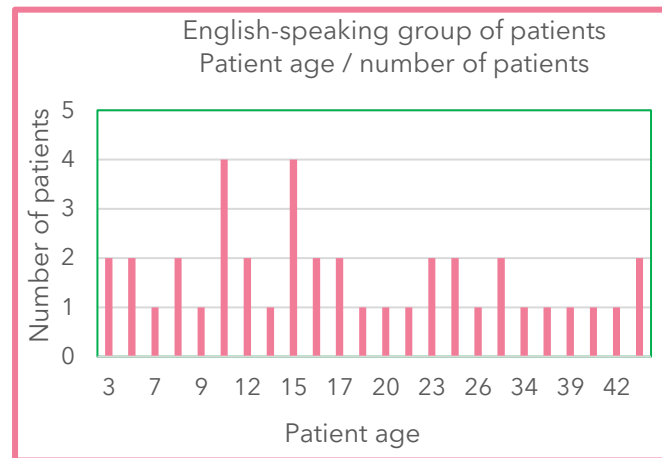


English-speaking group



- PKAN
- MPAN
- BPAN
- PLAN
- If other - please indicate the type of mutation in the next question

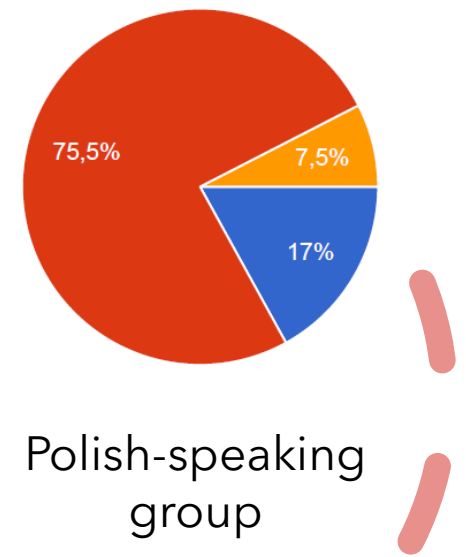
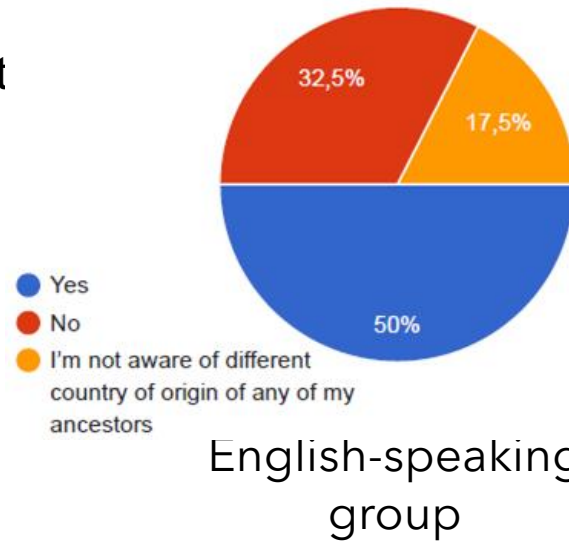
- Age and number of patients in each group





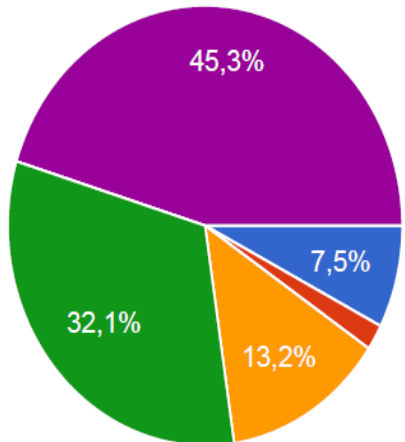
➤ Were the ancestors of the NBIA patient immigrated from another country?

Selected results from the NBIA Poland survey

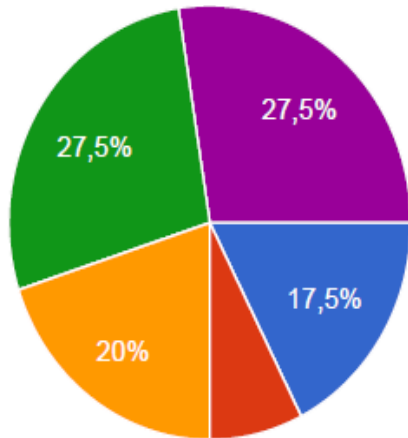


➤ Characteristics of patients according to the possibility of walking

Polish-speaking group

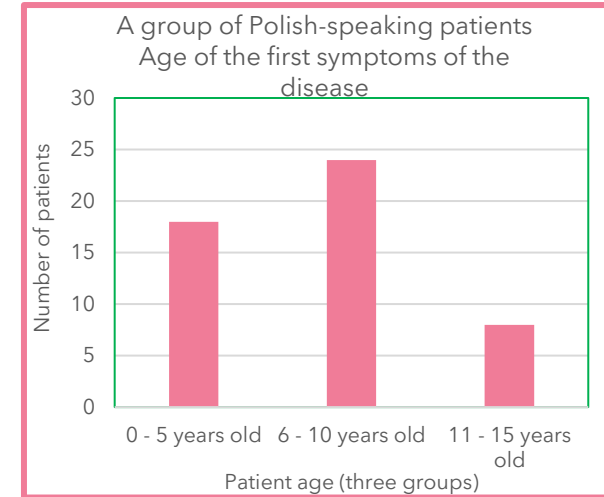
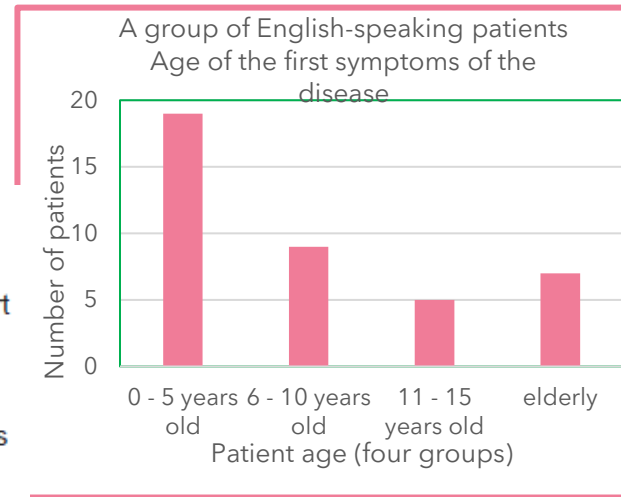


English-speaking group



- walks alone with no support needed
- minor walking problems
- moderate walking problems
- severe walking problems
- unable to walk

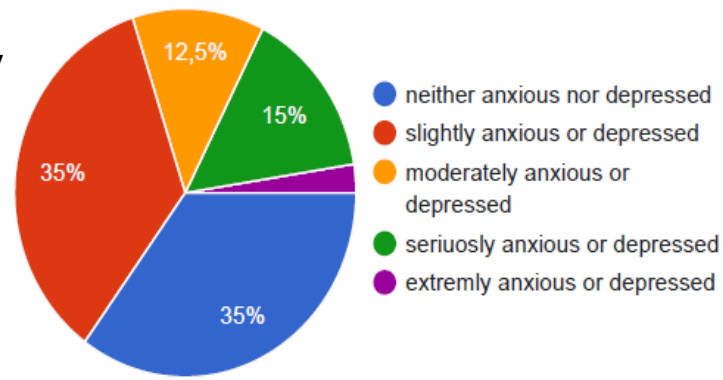
➤ Age of patients who recognized the first symptoms



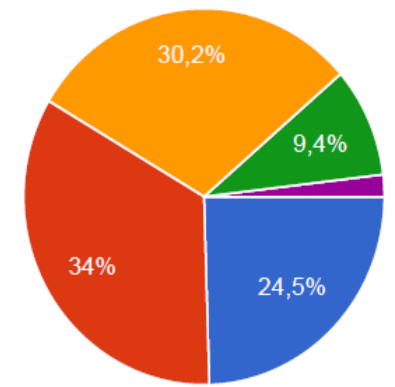


➤ The perception of mental health by NBIA patients

Selected results from the NBIA Poland survey



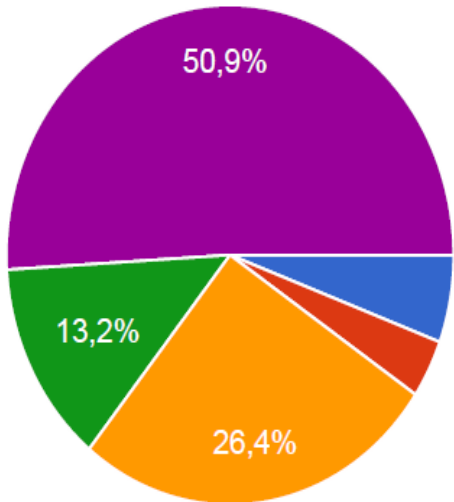
English-speaking group



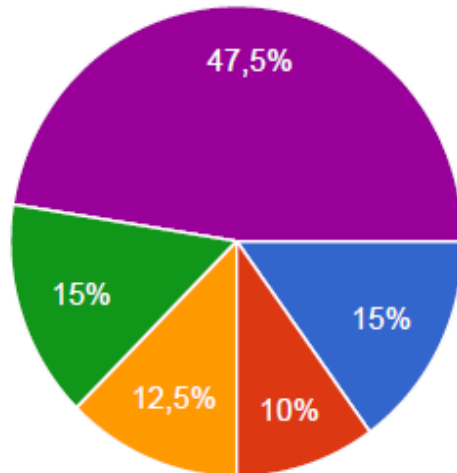
Polish-speaking group

➤ Characteristics of patients due to the possibility of performing simple household tasks / personal hygiene

Polish-speaking group

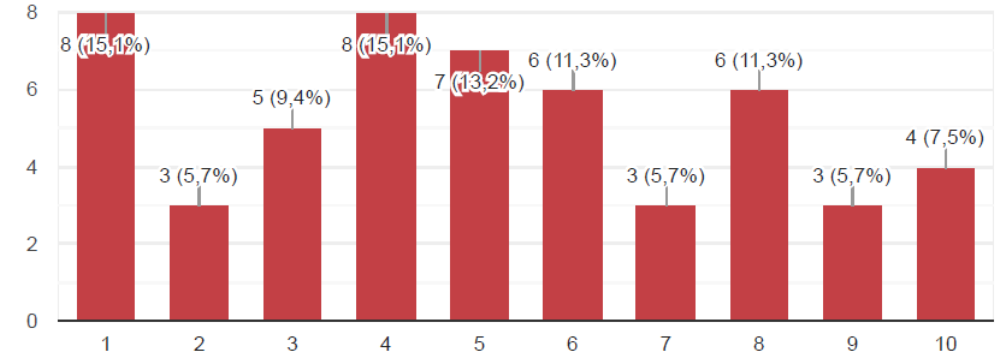


English-speaking group

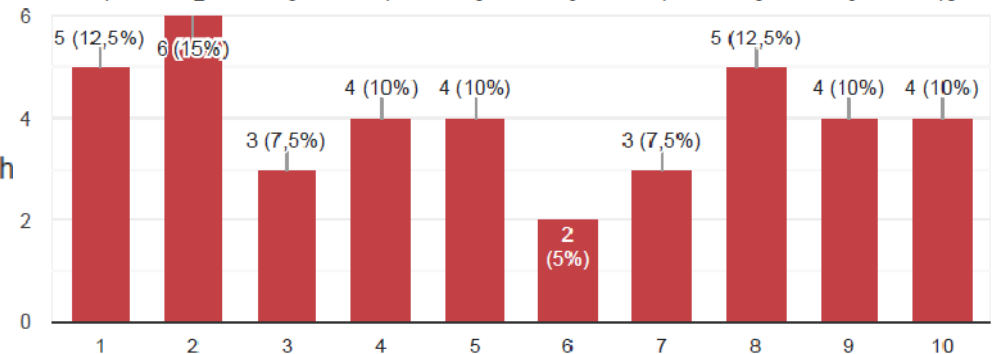


- fully independent
- minor problems
- moderate problems
- severe problems
- unable to dress and/or wash himself/herself

➤ Scale for assessing intellectual performance of NBIA patients



Polish-speaking group



English-speaking group

Selected results from the NBIA Poland survey



And many, many more questions in NBIA Poland Survey:

- We have characteristics of patients according to the types of medications taken (which form they prefer).
- We have the characteristics of patients in terms of the type of rehabilitation carried out.
- - We have characteristics of patients according to the type of equipment (strollers, walkers, orthoses, DBS, etc.) that helps them in their daily life.
- We have the characteristics of the families from which they emigrated, in which generation they emigrated, where are their roots (on the part of grandmothers, children and sometimes later generations).
- We have the characteristics of siblings; we know how many families have more than one NBIA child. We know how many patients have siblings without symptoms.

It is a pity that some other associations did not take part in this work! We would have shared data. We would have a joint publication! - as NBIA Alliance and not as NBIA Poland Association.

The research team will work on these data in detail. We will try to publish a full article from this research this year (currently the article is 30 pages long and too long for a single publication).



The statutory activity of the association:

- Gathering the patient database, organizing a family community. Exchange of information about the disease and about ongoing research.

- Collaboration with doctors and scientists.

Logistic assistance in the organization of blood and biological material tests. Assisting families in carrying out such tests.

- Cooperation with rehabilitation centers.

Organizing rehabilitation stays. Cooperation with rehabilitation specialists, providing information on types of exercises, equipment, methods of rehabilitation - proper rehabilitation is particularly important - especially when will be therapy programs.

- Raising funds for research programs. Cooperation with scientists and doctors. **Co-financing of research programs.**



Programs implemented / financed by the NBIA Poland Association in 2022.

We call on us to create a **database of programs in which NBIA associations** participate.

We call to create a **database of patient numbers in individual associations**, detailing the types of NBIA, so that doctors / scientists can better plan clinical trials and research. One NBIA Alliance list, updated every 1 year.

Creating a **list of research teams and clinics that work with our associations** to facilitate contact between teams of physicians. One list of collaborative research centers for the NBIA Alliance.

Presentation of the results to the database from the NBIA Polska association on the following slides



1



Current programs with the participation of the NBIA Polska Association.

Title:

Program INVESTIGATING C19orf12 REDOX FUNCTIONS of METABOLISM...

Program duration:

1/08/2020 to 31/08/2022. Currently extended to 02/27/2023.

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

Financing unit

Research Unit :

Helmholtz Center München, Germany

HelmholtzZentrum münchen

German Research Center for Environmental Health

Project leaders:

Dr A. Iuso, Dr A. Messiah

2



Title:

ENZYMATIC THERAPY project

Program duration:

The program started in 01/01/2021 and is open continuously

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

Co-financing unit

Research Unit :

- Institute of Psychiatry and Neurology in Warsaw II Department of Neurology,
- University Children's Hospital in Lublin,
- Warsaw University of Life Sciences
- Children's Health Center (IPCZD). Locomotor System Diagnostics Laboratory,
- OLINEK Intensive Therapy Center in Warsaw

Project leaders:

Dr Marta Skowrońska; Prof. I. Kurkowska-Jastrzębska; Prof. M. J. Syczewska
Prof. M. Chrościńska-Krawczyk; Dr J. Janicka-Rachtan; Mgr A. Stęplowska



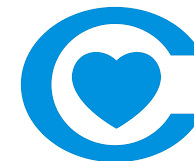
Instytut Psychiatrii i Neurologii
w Warszawie



UNIWERSYTECKI
SZPITAL DZIECIĘCY
W LUBLINIE



SZKOŁA GŁÓWNA
GOSPODARSTWA
WIEJSKIEGO



Olinek
Centrum Intensywnej Terapii



3



Title:

Studies of metabolic disorders in patients with neurodegeneration with deposition of iron in the brain.

Program duration:

from 04/29/2022 to 05/01/2024.

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

Financing unit

Research Unit :

- Institute of Experimental Biology M. Nencki of the Polish Academy of Sciences (PAN)
- Institute of Psychiatry and Neurology in Warsaw II Department of Neurology (cooperation)
- **Project leaders:**

Prof. M. Więckowski, Dr M. Skowrońska



Instytut Psychiatrii i Neurologii
w Warszawie



instytut biologii doświadczalnej
im. Marcelego Nenckiego PAN

4



Title:

Effect of experimental therapy with the use of autophagy modulators on inflammatory processes in the CNS in basic NBIA variants..

Experimental drug program for BPAN patients (first group - research based on the consent of the ethics committee)

Program duration:

from 20/11/2022 duration: 1 year.

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

Financing unit

Research Unit :

- University of Gdańsk Faculty of Biology Department of Molecular Biology
- University Children's Hospital in Lublin, Department of Children's Neurology - Department of Children's Neurology

Project leaders:

Prof. G. Węgrzyn, Prof. M. Chrościńska-Krawczyk



UNIwersytecki
SZPITAL DZIECIĘCY
W LUBLINIE



5



Title:

Studies of Lipid Pathway Metabolism in Patients with NBIA.....

The program is financed by the Ministry of Education and Science in Poland

Program duration:

Project duration 2 years from April 29, 2022

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

The social partner of the program

Research Unit :

- Institute of Experimental Biology M. Nencki of the Polish Academy of Sciences (PAN),
- Institute of Psychiatry and Neurology in Warsaw II Department of Neurology,

Project leaders:

Prof. A. Dobrzyń; Dr M. Skowrońska; Prof. M. Więckowski;
Prof. I. Kurkowska-Jastrzębska



**NAUKA DLA
SPOŁECZEŃSTWA**



**Instytut Psychiatrii i Neurologii
w Warszawie**



**Ministerstwo
Edukacji i Nauki**



**instytut biologii doświadczalnej
im. Marcelego Nenckiego PAN**

7

**Title:**

Establishment of a biobank of cells and biological material of NBIA patients

Program duration:

Project started 01/09/2022 and is open continuously



Instytut Psychiatrii i Neurologii
w Warszawie

NBIA Aliance participating in the program:

NBIA Poland Association

Function of the NBIA Polska Association:

Co-financing unit

Research Unit :

- Institute of Psychiatry and Neurology in Warsaw II Department of Neurology,

Project leaders:

Dr M. Skowrońska;

IMPORTANT!

The biological material for research is freely available to the research teams of the NBIA. The condition for obtaining biological material is:

- obtaining the required consent of the biotechnology commission to conduct research,
- participation in research / access to research results for the team of scientists of Dr. Marta Skowrońska (IPIN Polska), and access to research results for the NBIA Poland Association.

For commercial entities, access to biological material on the basis of a separate cooperation agreement with the team of Dr Marta Skowrońska (IPIN Polska)

Characteristics of the members of the NBIA Poland association - after the meeting on 01.10.2022.



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1.	Liczba członków ogółem	93
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5.	PLAN	2
6.	Bez badań genetycznych	2
7.	Bez chorego	1
8.	Do diagnozy prawd.MPAN	1



STOWARZYSZENIE
NBIA
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Research units cooperating with the NBIA Poland Association (as of 2022).

Lp.	Nazwa jednostki prowadzącej badania z zakresu NBIA	Lider Projektu	Zakres badań
1.	Instytut Psychiatrii i Neurologii w Warszawie II Klinika Neurologiczna	Dr hab. n. med.. M. Skowrońska; Prof. dr hab n. med. I. Kurkowska-Jastrzębska	Badania Podstawowe i kliniczne; Diagnostyka
2.	Uniwersytecki Szpital Dziecięcy e Lublinie Klinika Neurologii Dziecięcej - oddział Neurologii Dziecięcej	Prof. dr hab. n. med. M. Chrościńska-Krawczyk	Badania Kliniczne; Diagnostyka
3.	Centrum Zdrowia Dziecka (IPCZD)	Prof.dr hab.n.med. M. J. Syczewska; Dr n. med.. T. Kmieć	Diagnostyka
4.	Centrum Intensywnej Terapii OLINEK w Warszawie	Mgr A. Stęplowska	Rehabilitacja
5.	SGGW Warszawa	Dr J. Rachtan - Janicka	Badania w zakresie dietetyki
6.	Uniwersytet Gdański Wydział Biologii Katedra Biologii Molekularnej	<u>Prof. dr hab. G. Węgrzyn</u>	Badania Podstawowe
7.	<u>Instytut Biologii Doświadczalnej im. M. Nenckiego PAN</u>	Prof. dr hab. A. Dobrzyń; Prof. dr hab. M. Więckowski	Badania Podstawowe
8.	Helmholtz Center München, Germany	Dr A. Iuso Dr A. Messias	Badania Podstawowe



Dears

Thank you for your attention.

Maciej Cwyl.
President of NBIA Poland
maciej.cwyl@pw.edu.pl

<https://nbia-polska.org/>

Special thanks to Bogusław Kiejnowski, member of the management board, for preparing the materials for the presentation.

Many thanks to all members of the NBIA Poland Association for their work on these programs - all activities of the association are charitable - for the benefit of NBIA patients

Lozana 2022

