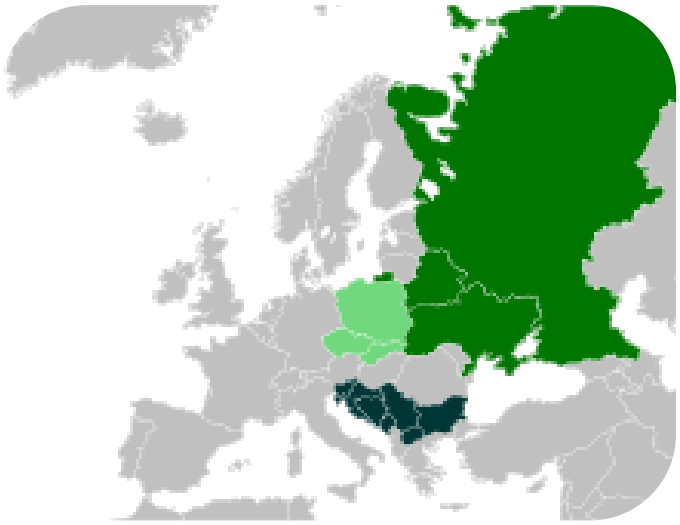
International Patient Organisation for Primary Immunodeficiencies

Access to care for PID patients in the Eastern region



Belarus is a country in Eastern Europe, with its population of 9, 464 million peoples.

The first immunological division was established in 2002 at the Belarusian Research Center for Pediatric Oncology and Hematology, Minsk region.

Before most of the first patients was diagnosed late in Germany, Israel, UK.

History

National registry for children with PID was established in November 2006 after 1st J-Meeting in Belarus under the patronage of professor László Maródi.

When we had no clinical immunologists
no experience in molecular diagnostics
no registry
….
Nobody believed us that PID were common disease!!!

But we had flow cytometry, PCR and sequencing lab (without sequencing experience, BMT unit and many patients with unusual clinical presentation).

But!!!

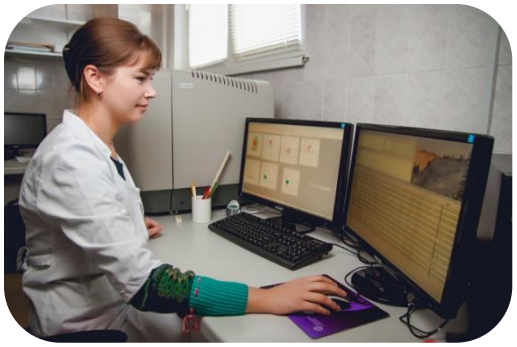
Mikhael Belevtsev, PhD
Research Director of
Belarusian Research Center for Pediatric Oncology, Hematology and Immunology, Minsk region, Belarus

Olga Aleinikova, MD, PhD, professor
Director of
Belarusian Research Center for Pediatric Oncology, Hematology and Immunology, Minsk region, Belarus

All medical care (diagnostics and treatment options) were organized in Belarus and paid by The Ministry of Health of the Republic of Belarus

2006 - 2014

Immunological laboratory for PID diagnostics



Genetic laboratory for PID diagnostics

Others laboratories for PID diagnostics

PID’s Meetings in regional centers (August – October 2013)

In each city, were present at the meeting of more than 50 pediatricians and immunologists

Creation Jeffrey Modell Diagnostic and Research Center for Primary Immunodeficiencies (February 2012)

CONTRACT ON SPONSORING

made by and between

Jeffrey Modell Foundation
780 Third Avenue
New York 10017
USA

(hereinafter referred to as "Foundation")

and

public institution "National Research Center for Pediatric Oncology, Hematology and Immunology"
223053, Belarus, Minsk district, Borovlyany,
Frunzenskaya str. 43,

(hereinafter referred to as "Center")

ДОГОВОР ПРЕДОСТАВЛЕНИЯ ИНОСТРАННОЙ БЕЗВОЗМЕЗДНОЙ ПОМОЩИ

заключен между

«Джефри Моделл Фаундейшн»
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Нью-Йорк 10017
США

(далее именуемый «Фонд»)

и

Государственное учреждение «Респуб
научно-практический центр детской о
гематологии и иммунологии
223053, Республика Беларусь, Минск
Боровляны, ул. Фрунзенская д

(далее именуемый «Центр»)

Minsk, Republic of Belarus
February 27, 2012

Jeffrey Modell Foundation

Frederick Modell
Co-Founder

Vicki Modell
Co-Founder

Public Institution
"National Research Center for Pediatric Oncology, Hematology and Immunology"

Olga Aleinikova
Director

Jeffrey Modell Foundation

Jeffrey Modell Diagnostic and Research Center for Primary Immunodeficiencies

Центр диагностики и изучения первичных иммунодефицитов имени Джеффри Моделла

Минск, Республика Беларусь
27 февраля, 2012 ода

«Джефри Моделл Фаундейшн»

Фредерик Модел
Соучредитель

Вики Моделл
Соучредитель

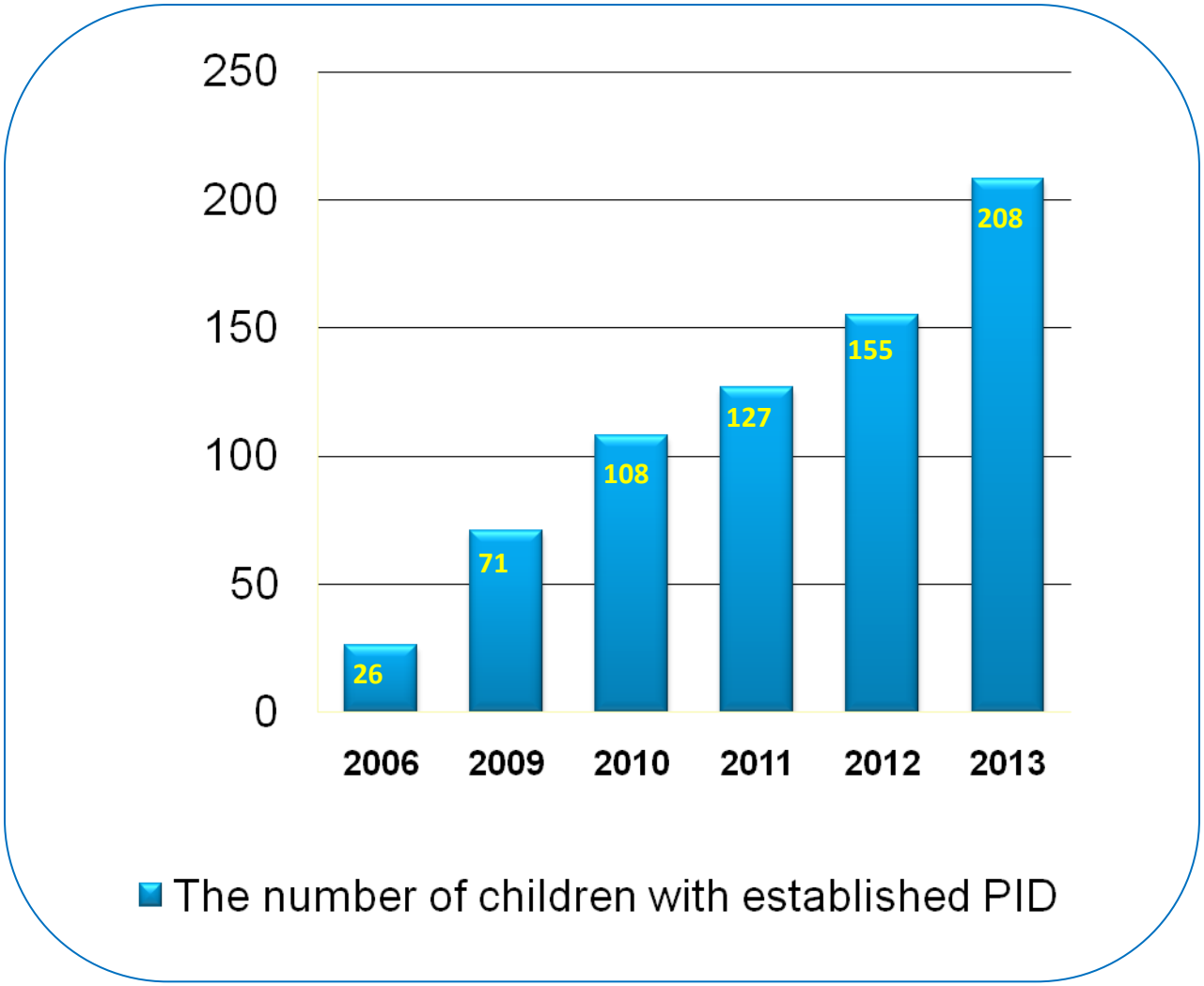
Государственное учреждение «Республиканский научно-практический центр детской онкологии и гематологии»

Ольга Витальевна Алейникова
Директор

As of February 2014

208 children (131 males, 77 females),

163 alive (78,4%) were registered

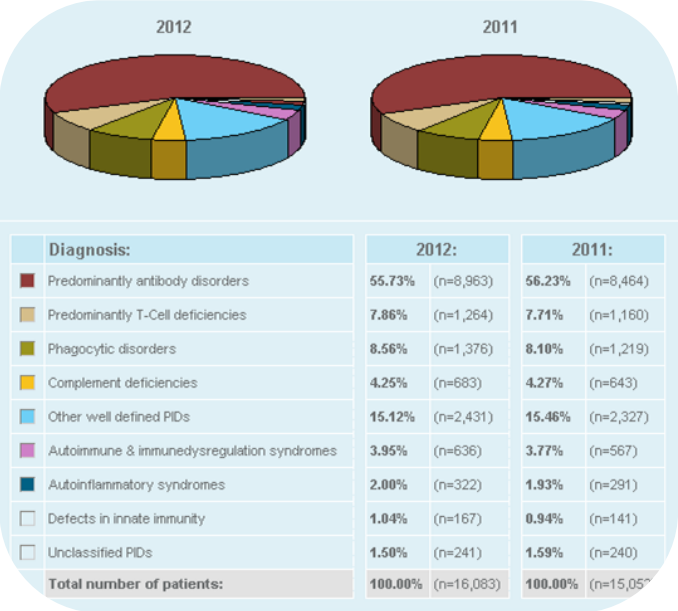


List of genes for PID diagnostics

1. *BTK gene*	XLA
2,3. *TACI and ICOS gene*	CVID
4. *CD40L*	X-HIgM
5. *IL7R*	AR –SCID
6. *γ chain of R-s for IL-2, IL-4, IL-7, IL-9, IL-15, IL-21*	X-SCID
7. *JAK 3*	AR -SCID
8. *FOXP3 gene*	IPEX
9. *AIRE gene*	APECED
10. *WAS gene*	WAS
11. *FAS gene*	ALPS I
12. *CYBB gene*	X-CGD
13. *C1INH gene*	Hereditary angiodema
14. *SH2D1 gene*	XLP I
15. *XIAP gene*	XLP II
16 *ADA gene*	AR-SCID
17, 18 *RAG1,2*	AR-SCID
19 *ATM*	A-T
20. *NBS*	Syndrome Nijmegen
21. *GATA2 gene*	MonoMac syndrome
22. ELANE	Congenital neutropenia

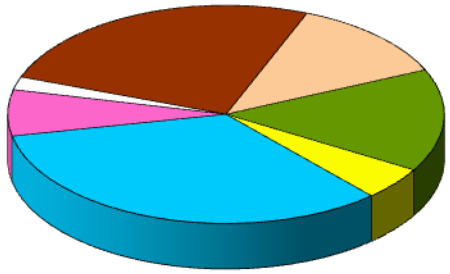
34 different genetics groups

2013



Diagnosis:	2012:		2011:	
Predominantly antibody disorders	55.73%	(n=8,963)	56.23%	(n=8,464)
Predominantly T-Cell deficiencies	7.86%	(n=1,264)	7.71%	(n=1,160)
Phagocytic disorders	8.56%	(n=1,376)	8.10%	(n=1,219)
Complement deficiencies	4.25%	(n=683)	4.27%	(n=643)
Other well defined PIDs	15.12%	(n=2,431)	15.46%	(n=2,327)
Autoimmune & immunedysregulation syndromes	3.95%	(n=636)	3.77%	(n=567)
Autoinflammatory syndromes	2.00%	(n=322)	1.93%	(n=291)
Defects in innate immunity	1.04%	(n=167)	0.94%	(n=141)
Unclassified PIDs	1.50%	(n=241)	1.59%	(n=240)
Total number of patients:	100.00%	(n=16,083)	100.00%	(n=15,05

http://www.esid.org/statistics.php?sub=2

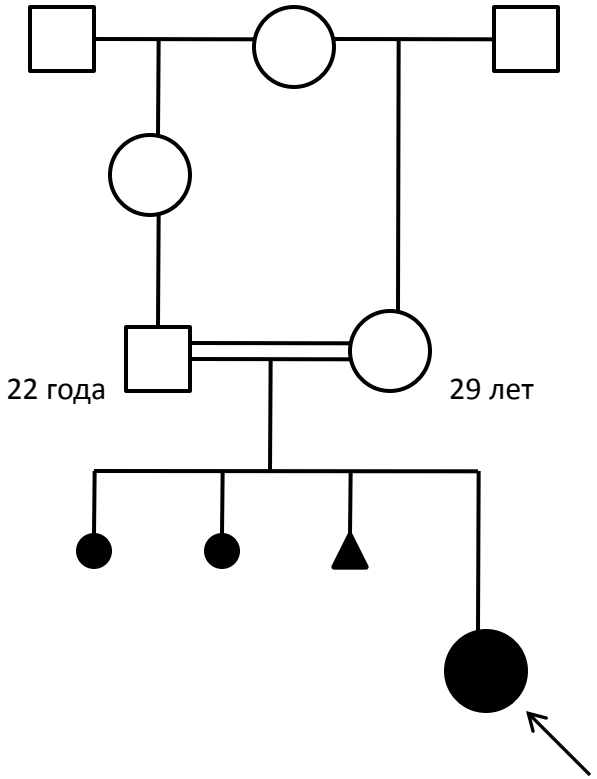


- Predominantly Ab disorders **25%** (n=52)
- Predominatly T-cell deficiencies **12,02%** (n=25)
- Phagocytic disirders **14,9%** (n=31)
- Complement deficiencies **4,8%** (n=10)
- Other well defined PIDs **32,7%** (n=68)
- Autoimmune&immunodysrequlation syndromes **6,7%** (n=14)
- Defects of innate immunity **1,9%** (n=4)

Total number 100% (n=208)

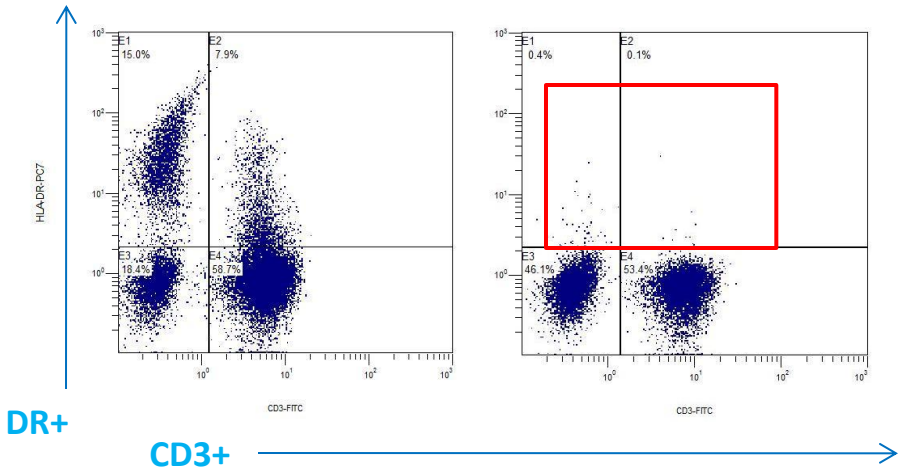
Consanguineous marriages are not typical in Belarus, 20% of PID is X-linked, among autosomal recessive forms Nijmegen syndrome is the most popular (16 patients).

MHC II deficiency. Clinical case



Female, at the age of 5 months – was admitted to our hospital.

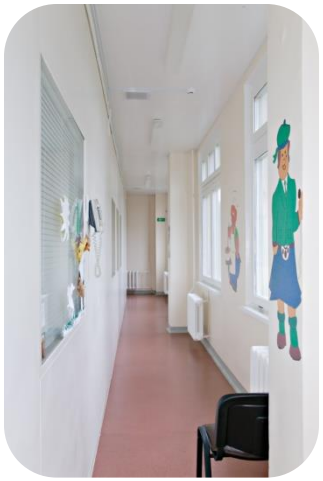
Hepatomegaly +3 cm, spleen +1 cm.
CMV-infection, low CD4+ T cells



BMT/HSC transplantation in Belarus

14 patients were treated with BMT/HSCT, 11 – alive.

2002 – Nijmegen syndrome – BMT – alive
2003 – X-CGD – BMT – alive
2008 – SCID – HSCT (haplo) – alive
2009 – XLP – BMT - dead
2009 – LADI – BMT (twice) – dead
2010 – Omenn-like – Cord blood SC – dead
2011 – WAS – BMT – alive
2013 – MHC-II, WAS, Nijmegen, congenital neutropenia (2 pts), SCID, X-CGD - alive



IVIg/SCIg in Belarus

-- **27 patients** received immunoglobulins (XLA, AR-agammaglobulinemia, CID, CVID, WAS)

--17 patients – IVIg
-- 10 patients - SCIg

-- **12 patients with X-CGD** treated with γ-IFN

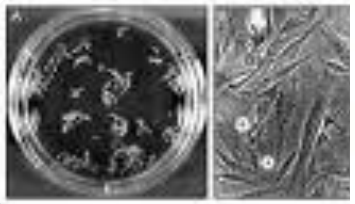
Prenatal diagnostics

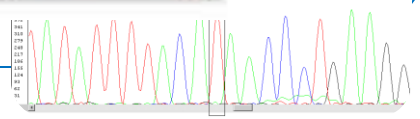
Together with specialists from Belarusian Medical Center "Mother and Child" Minsk, Belarus

Prenatal diagnostics was organized in Minsk in 2009

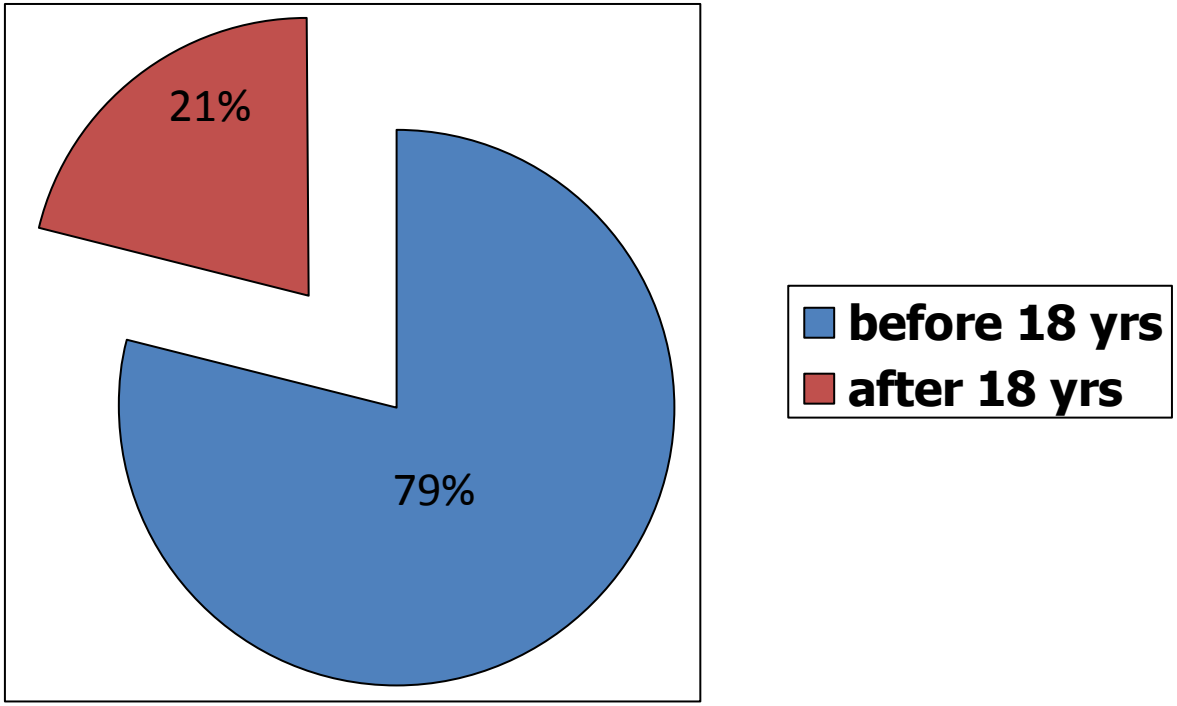
- *First CYBB* mutation detection in Family with X-linked CGD
- Now **7 PD** were performed in Belarus and more **12 families were genetically consulted**



Adult patients among all PID patients in Belarus (December 2013)

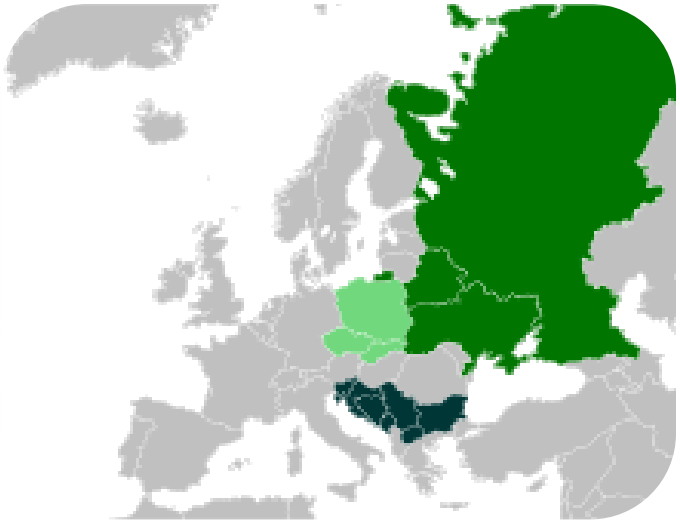


All adult patients treated in Minsk Diagnostic Center and Belarusian Research Centre for Radiation Medicine and Human Ecology, Gomel, Belarus.

2013 - 2014

First *ATM* diagnostics for Russian patient (paid by patients parents). (4 pts)

First diagnostic and treatment of patient from Ukraine (paid by charity foundations). (6 patients)



May 2010 The First Patients PID Meeting

- More than 70 patients and their parents visited The Fist PID Meeting;
  - August 2010 in the Ministry of Justice of the Republic of Belarus was registered social organization of patients with PID **“Save immunity” (Спасем иммунитет)**

2011, 2012 and 1st March 2014 were organised Patients PID Meeting
2013 rent Office for Patient PID Organization



IPOPI International Patient Organisation for Primary Immunodeficiencies

www.ipopi.org

Thank you for your attention