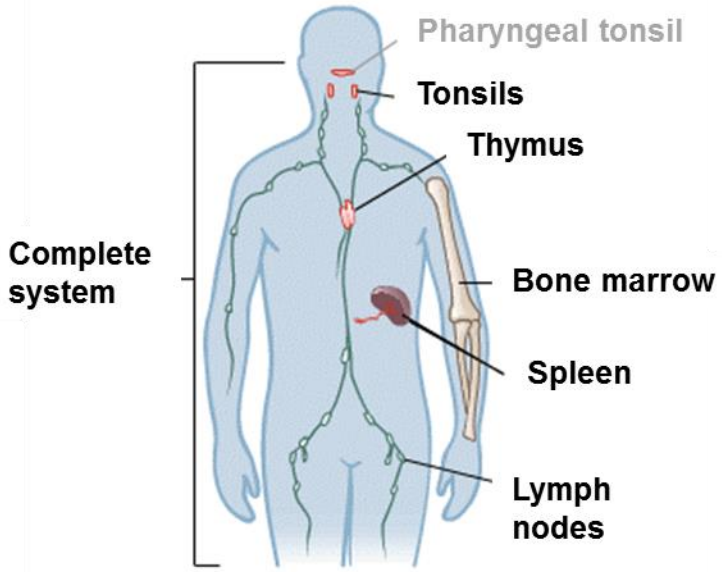
BARCELONA PID FOUNDATION

IMMUNOLOGY & INFECTION

Fundació Barcelona per a les Immunodeficiències Primàries

Primary Immune Deficiencies (PID)

WHAT ARE PID?



Heterogeneous group of diseases caused by quantitative and/or functional defects in the immune response.

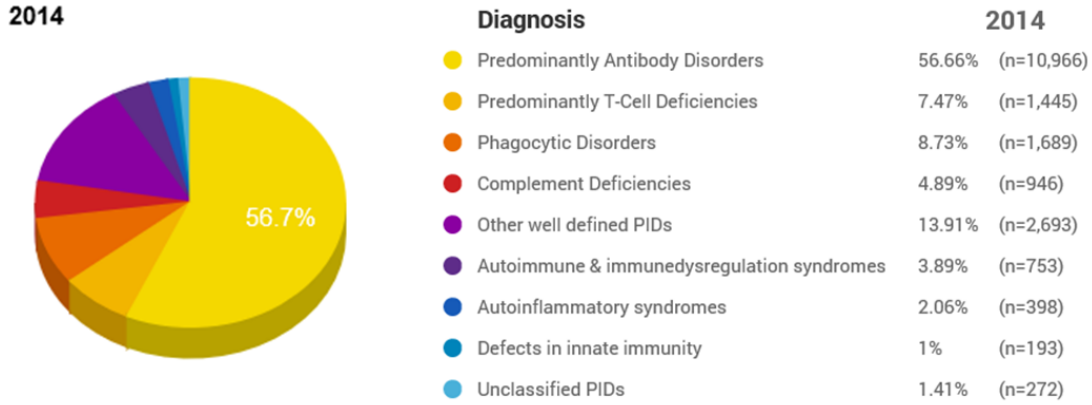
Affect 1 in 2000 live births

Martín-Nalda A, Soler-Palacín P, Caracseghi F, Hernández M, Martínez-Gallo M, Figueres-Nadal C.
Immunodeficiències primàries: cal pensar-hi.
Pediatr catalana 2011; 71: 23-29

Primary Immune Deficiencies (PID)

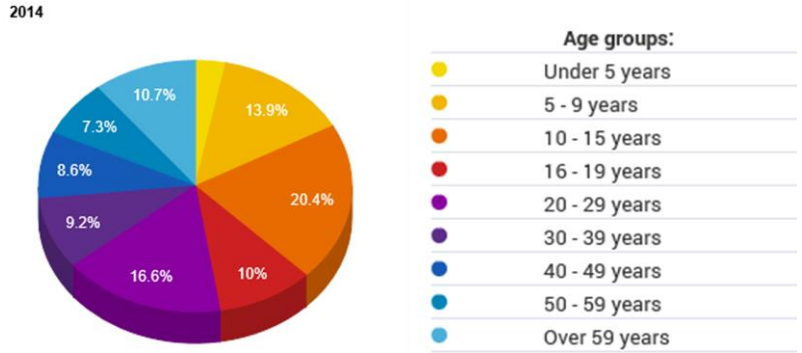
CLASSIFICATION

- Includes more than 200 disorders
- Classified into 8 groups according to their characteristics



Major Immunodeficiency Groups. June 2014. ESID Database Statistics

- More than 60% of patients are diagnosed in childhood
- 10% of cases - the most severe – can be diagnosed at birth (newborn screening)

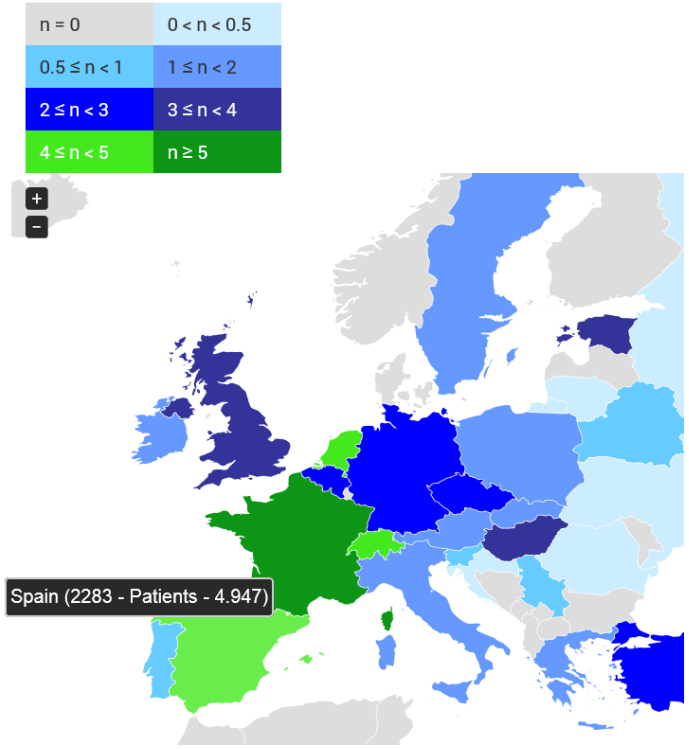


Age Groups. June 2014. ESID Database Statistics

Primary Immune Deficiencies (PID)

PREVALENCE

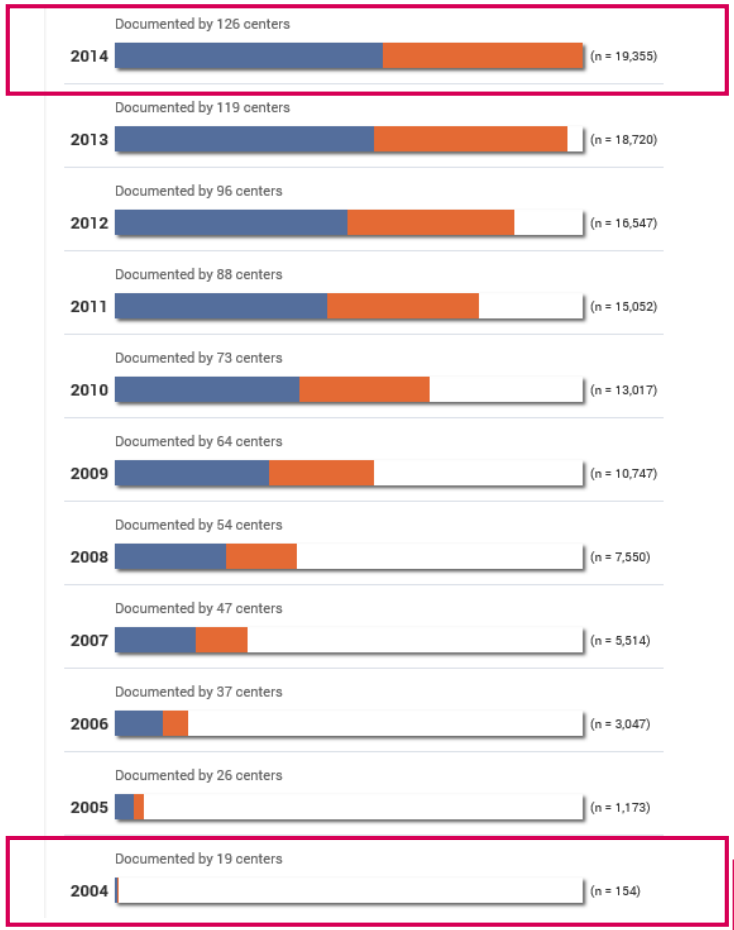
PREVALENCE IN EUROPE



Documented prevalence of PID in Europe, 2014.
ESID Database Statistics

NUMBER OF PATIENTS WITH PID IN EUROPE

Registered by the ESID, June 2014



Primary Immune Deficiencies (PID)

CLINICAL FEATURES

Patients with PID are more susceptible to:

INFECTIONS

Recurrent and severe. If not properly treated, can be fatal or lead to serious sequelae.

AUTOIMMUNE DISORDERS

Anemia, thrombocytopenia

Arthritis

Enteropathy

CANCER

Mainly leukemia and lymphoma

ALLERGY

Respiratory

Food

Skin

CHRONIC ILLNESS, CHRONIC PATIENTS

With all that this implies for patients and their families, physically, psychologically and socially

Martín-Nalda A, Soler-Palacín P, Caracseghi F, Hernández M, Martínez-Gallo M, Figueres-Nadal C. ***Immunodeficiències primàries: cal pensar-hi.***
Pediatr catalana 2011; 71: 23-29

Primary Immune Deficiencies (PID)

THE MOST SEVERE FORM

The most severe form is severe combined immune deficiency (SCID), which became widely known through an affected patient, the "bubble boy".

- These children cannot be exposed to our environment without the risk of dying from any infection.
- Stem cell transplantation is required to cure these patients.
- They must remain isolated until they find a compatible donor.
- 90% survival when transplantation is performed before the 3rd month of life compared to 60% after.
- 20 cases the past 10 years in Catalonia.

"Bubble boy" David Vetter lived in a protective environment designed by NASA engineers. He died of complications after receiving a bone marrow transplant in 1984, at the age of 12. / **BAYLOR COLLEGE OF MEDICINE PHOTO ARCHIVES**

100% die before one year of age without stem cell transplantation.

A prompt diagnosis can save their life. Importance of **newborn screening**.

Primary Immune Deficiencies (PID)

TREATMENT

PALLIATIVE

- Immunoglobulin replacement: IVIG, SCIG
- Anti-infective prophylaxis
- Vaccination

CURATIVE

- Hematopoietic stem cell transplantation (bone marrow transplantation)
- Gene therapy

Additional statistics:		2014
How many alive patients** overall are on Ig replacement?	(n=6,476)	50.36%
... alive patients** on IVIG	(n=4,239)	32.96%
... alive patients** on SCIG	(n=2,181)	16.96%
... alive patients** on IMIG	(n=22)	0.17%
Total number of alive patients**:	(n=12,860)	100%

**Alive patients with no information on therapy are excluded from this evaluation.

Ig replacement statistics, June 2014. ESID Database Statistics

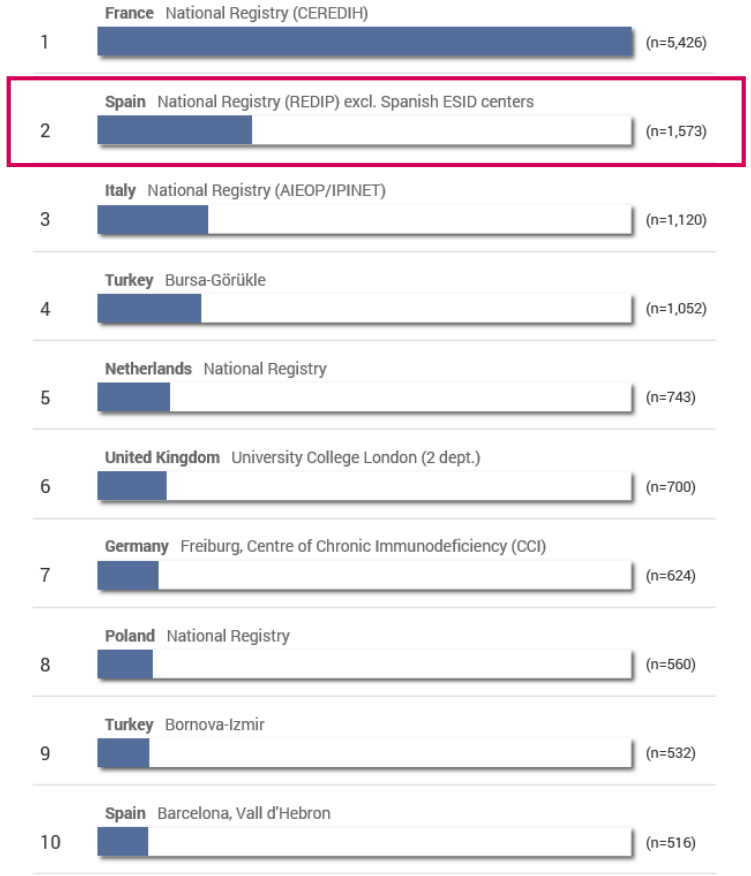
Immunoglobulin (Ig) has an important role in many of these diseases.

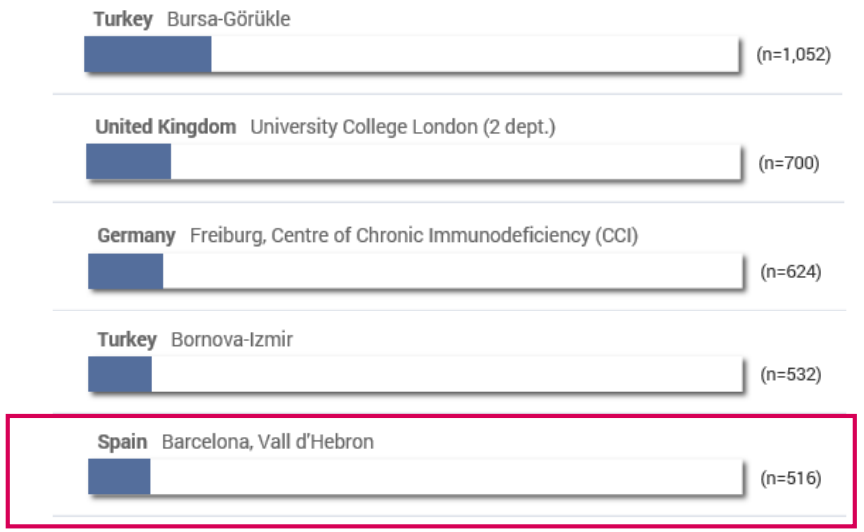
+ NEED FOR MULTIDISCIPLINARY FOLLOW UP IN A REFERENCE CENTER

Primary Immune Deficiencies (PID)

REFERENCE CENTERS

Reference and fellowship centers for PID in Europe according to the ESID





University Hospital Vall d'Hebron occupies the 5th position among centers that register directly to the ESID Database.

Ranking of documenting centers, June 2014. ESID Database Statistics

Primary Immune Deficiencies (PID)

REFERENCE CENTERS

Reference and fellowship centers for PID in Europe according to the ESID

Centers for fellowship in PID, June 2014. ESID website

- Freiburg, Germany - Centre for Chronic Immunodeficiency
- Munich, Germany – University Children's Hospital
- Paris, France – Necker Medical School
- London, United Kingdom - Great Ormond Street Hospital
- Newcastle, United Kingdom - Newcastle Upon Tyne
- Florence, Italy – Anna Meyer Children's Hospital
- **Barcelona, Spain – Unitat de Patologia Infecciosa i Immunodeficiències de Pediatria (UPIIP), Hospital Universitari Vall d'Hebron** *(Pediatric Infectious Diseases and Immunodeficiencies Unit)*

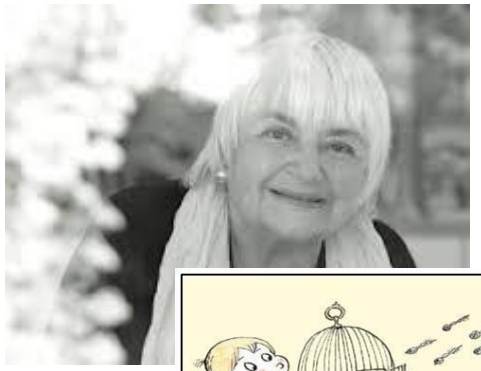
upiip
Unitat de Patologia Infecciosa i Immunodeficiències de Pediatria.
Hospital Universitari Vall d'Hebron. Barcelona.

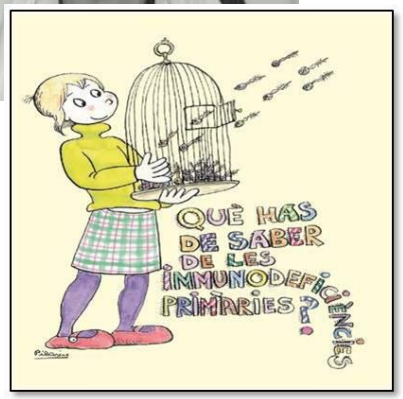
PID FOUNDATION Barcelona

Primary Immune Deficiencies (PID)

AWARENESS

- **Little information** reaches the general population and non-specialist healthcare professionals.
- **Training and informative campaigns** are required.
- **Training courses** for primary care pediatricians and other health professionals are needed **to improve early diagnosis of PID.**
- **Public awareness should be raised.**





LA VANGUARDIA

Les immunodeficiències primàries

Les famílies i els especialistes demanen la detecció precoç d'immunodeficiències primàries en els nounats

El combat dels indefensos

Sis anys amb una medul·la nova

Immunodeficiències greus es curen amb trasplantaments primerenca de medul·la òssia

Són poc freqüents i passen inadvertides com a infeccions repetides

Diagnosticar-les aviat és determinant en el pronòstic del malalt

BARCELONA PID FOUNDATION

IMMUNOLOGY & INFECTION

PID FOUNDATION Barcelona

Primary Immune Deficiencies (PID)

AWARENESS

TV

Radio

Press

sábado 31 de mayo de 2014

Ventana del paciente "Foro Albert Jovell"

Hasta un 90% de supervivencia con diagnóstico de IDCG

CARLOTA VILLAR
Vicepresidenta ACADIP

Hoy en día en Cataluña ya son 22 las enfermedades metabólicas que se incluyen en el cribado neonatal o en la prueba del talón del recién nacido. Entre las patologías que se detectan, por desgracia, no se incluye la detección de una Inmunodeficiencia Combinada Grave (IDCG). Los que la sufren son conocidos como "niños burbuja". Estos padecen múltiples infecciones y, además, sin un trasplante de médula ósea mueren el 100% antes de cumplir el año de vida. El diagnóstico precoz mediante la detección de los TRECS (productos de desecho de la glándula timo) en la sangre del talón permitiría poder derivar a estos niños a un trasplante. Asimismo, si esto se llevara a cabo antes de los tres meses de vida se conseguiría una supervivencia del 90%.

Actualmente se conocen más de 250 IDP.

societat

"Tens un munt de dubtes i molta por"

Les famílies amb fills amb immunodeficiències primàries demanen avenços en el diagnòstic precoç

Reportatge

Nova unitat per fer diagnòstics a infants

BARCELONA PID FOUNDATION
IMMUNOLOGY & INFECTION

PID FOUNDATION Barcelona

Primary Immune Deficiencies (PID)

INVOLVEMENT OF SOCIETY

PATIENT AND FAMILIY ASSOCIATIONS:

ACADIP
ASSOCIACIO CATALANA DE DEFICITS IMMUNITARIS PRIMARIS

AEFAT
ASOCIACION ESPAÑOLA FAMILIA ATAXIA-TELANGIECTASIA

AEDIP
Asociación Española de Déficits Inmunitarios Primarios

22q

PID FOUNDATIONS:

- International PID foundations: IDF, Care for Rare Foundation, Jeffrey Modell Foundation, etc...

Immune Deficiency Foundation

jmcn Jeffrey Modell Centers Network

Care-for-Rare Foundation
Stiftung für Kinder mit seltenen Erkrankungen

- **Barcelona PID Foundation**: the only foundation in Catalonia and Spain fully dedicated to PID.

PID FOUNDATION Barcelona

PID FOUNDATION Barcelona

COMMUNITY INVOLVEMENT

WELL-KNOWN PEOPLE THAT GIVE SUPPORT TO PID PATIENTS:

Kilian Jornet – mountain runner

Pilarín Bayès - illustrator

Pep Bou - artist

COMMUNITY INVOLVEMENT

WELL-KNOWN PEOPLE THAT GIVE SUPPORT TO PID PATIENTS:

Àlex Rins – motorcycle racer

RCD. Espanyol – football players

Jordi Viladoms - motorcycle racer

...

BARCELONA PID FOUNDATION

WHO ARE WE?

BARCELONA PID FOUNDATION (Immunology & Infection) is a non-profit organization founded in 2014 by family members of PID patients and a group of health professionals dedicated to treating patients with PID and associated infectious complications.

The scope of the Barcelona PID Foundation is primarily Catalonia. However, it can work in the rest of Spain and internationally.

BARCELONA PID FOUNDATION

WHO WE ARE?

BOARD OF TRUSTEES – Founding board consisting of PID experts and relatives of patients

CONCEPCIÓ FIGUERAS NADAL – *Chair*

Degree in Medicine & Surgery (UAB, MD)

Pediatrician

Doctor of Medicine (UAB, PhD)

Senior Consultant in Pediatric Infectious Diseases (Unitat de Patologia Infecciosa i Immunodeficiències de Pediatria, Hospital Universitari Vall d'Hebron)

Associate professor (Universitat Autònoma de Barcelona)

Head of the Research Group Infecció en el Nen Immunodeprimit (VHIR)

ANDREA MARTÍN NALDA- *Secretary*

Degree in Medicine & Surgery (U. Zaragoza, MD)

Pediatrician

Consultant in Pediatric Infectious Diseases and PID (Unitat de Patologia Infecciosa i Immunodeficiències de Pediatria, Hospital Universitari Vall d'Hebron)

Member of the Research Group Infecció en el Nen Immunodeprimit (VHIR)

CARLOTA VILLAR AMER - *Vocal*

Degree in Dentistry (UIC)

Master in Orthodontics and Dentofacial Orthopedics

Associate professor (Universitat Internacional de Catalunya)

Mother of a child with PID

PERE SOLER-PALACÍN – *Vice Chair*

Degree in Medicine & Surgery (UAB, MD)

Pediatrician

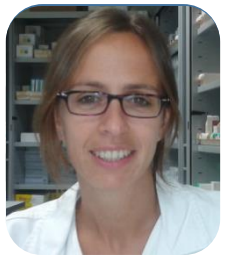
Doctor of Medicine (UAB, PhD)

Consultant in Pediatric Infectious Diseases and PID (Unitat de Patologia Infecciosa i Immunodeficiències de Pediatria, Hospital Universitari Vall d'Hebron)

Master in Pediatric Infectious Diseases (UAB, MsC)

Associate professor (Universitat Autònoma de Barcelona)

Member of the Research Group Infecció en el Nen Immunodeprimit (VHIR)



AURORA FERNÁNDEZ POLO- *Treasurer*

Degree in Pharmacy

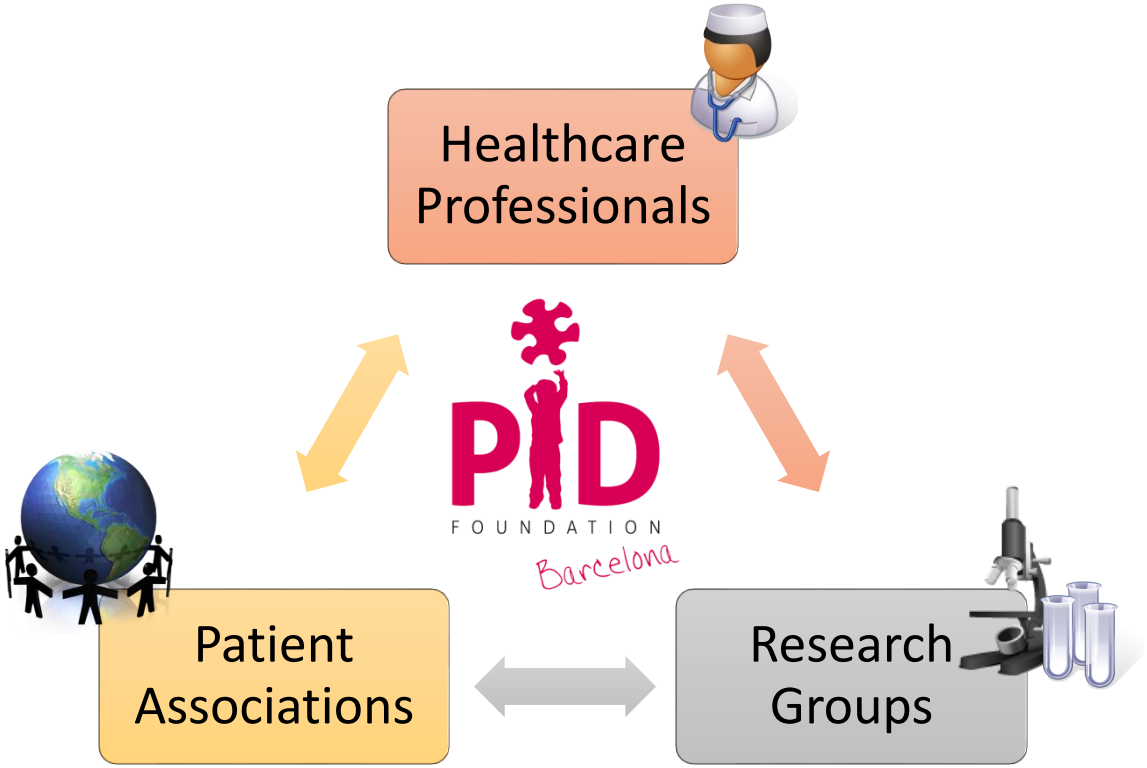
Specialist in Hospital Pharmacy

Master in Rare Diseases (Universitat de València, MsC)

Pharmacist (Àrea de Materno-infantil del Servei de Farmàcia, Hospital Universitari Vall d'Hebron)

Member of the Research Group Infecció en el Nen Immunodeprimit (VHIR)

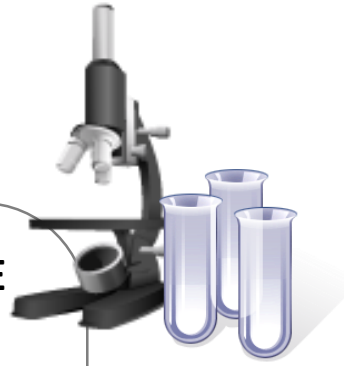
THE ROLE OF BARCELONA PID FOUNDATION



Enable and facilitate interdisciplinary links between healthcare professionals, research groups, and patient and family associations

MISSION

Our aims:



1

PROMOTE KNOWLEDGE AND RESEARCH IN PID

To promote research and dissemination of knowledge about PID and the infectious complications that may arise in these patients

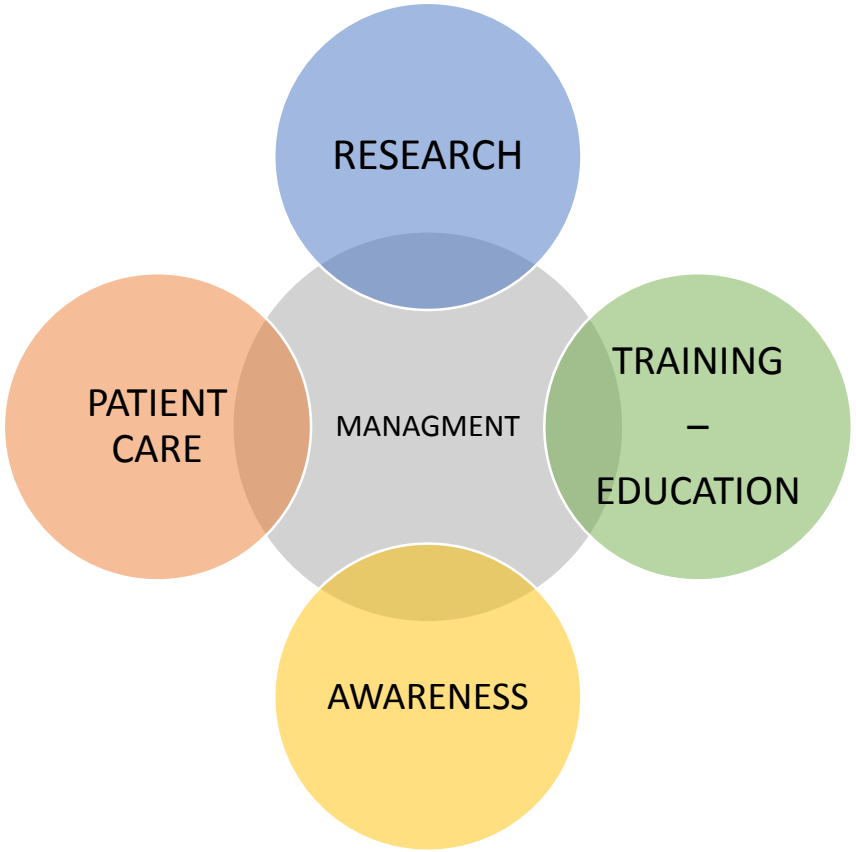
2

LEND SUPPORT

To support comprehensive health care of patients with PID and improve their quality of life through informative, educational, and clinical advice and social assistance campaigns

BARCELONA PID FOUNDATION

ACTIVITIES



RESEARCH

- Support research to improve scientific knowledge about PID
- Fight for prompt diagnosis of the more severe forms of PID. Promote screening programs in newborns

TRAINING - EDUCATION

- Train doctors, researchers, and students in PID and infectious diseases.
- Offer clinical advice and health education programs in PID and its infectious complications for non-specialized professionals nationally and internationally.

AWARENESS

- Design advertising strategies to raise awareness of PID and its infectious complications.

PATIENT CARE

- Provide support and expert medical recommendations to PID patients and patient associations

BARCELONA PID FOUNDATION

PROJECTS 2015 - 2016

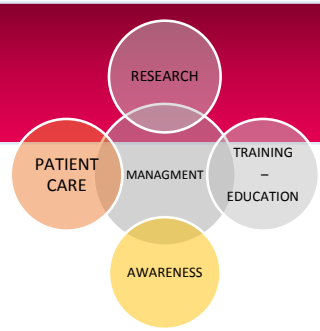
RESEARCH

- Patient registry in national and international databases
- Research focused on improving the diagnosis and treatment of PID and its infectious complications
- Early diagnosis. Newborn screening for the more severe forms of PID

TRAINING - EDUCATION

- Provide travel grants to young researchers who attend meetings and training courses of the ESID *(European Society for Immunodeficiency)* and other national or international meetings
- Carry out health education programs to raise awareness about PID

BARCELONA PID FOUNDATION



PROJECTS 2015 - 2016

AWARENESS

- Adapt the IPOPI (*International Patient Organization for Primary Immunodeficiencies*) information leaflets on PID
- Organize and promote social and sporting events to raise awareness of PID

PATIENT CARE

- Develop educational support programs for patients with PID and their families
- Provide specialized medical guidance to pediatric patients with PID, their families and patient associations for PID.
- Provide social aid to relatives of patients with PID (eg, food, transportation)
- Improve and optimize healthcare areas used to treat pediatric patients with PID

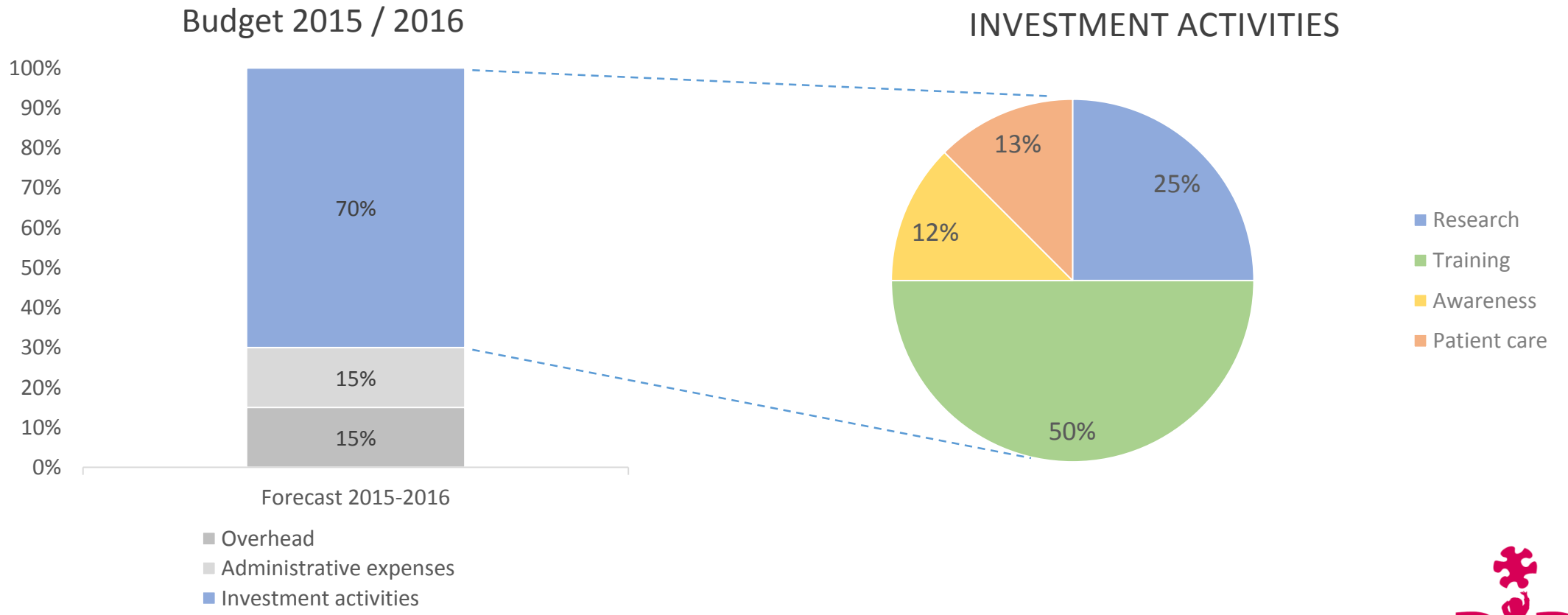
In collaboration with the ACADIP

ACADIP
ASSOCIACIO CATALANA DE DEFICITS IMMUNITARIS PRIMARIS

PID FOUNDATION Barcelona

BUDGET

YEARLY BUDGET



PARTNERS AND AGREEMENTS

AGREEMENTS

- Hospitals and research institutes
- Patient associations

PARTNERS

- Private entities
- Pharmaceutical companies
- Government
- Other partners

BARCELONA PID FOUNDATION

IMMUNOLOGY & INFECTION

Thanks a lot!

Fundació Barcelona per a les Immunodeficiències Primàries