6th IPOPI PID FORUM:
Principles of Care for Primary Immunodeficiencies

Managing PID diagnosis and care in all countries (P6)

Susana Lopes da Silva, MD

Centro de Imunodeficiências Primárias, Lisboa

ACCESS TO PID CARE WORLD-WIDE

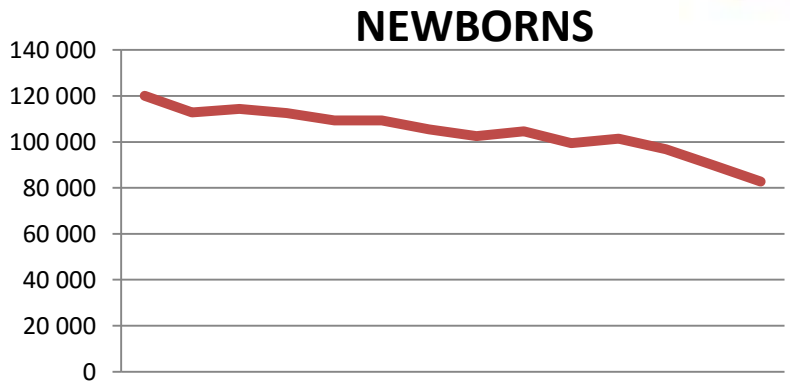
ACCESS TO PID DIAGNOSIS

ACCESS TO PID TREATMENTS

PORTUGAL

10.51 MILLION INHABITANTS

DECREASING BIRTH RATE



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PIDs IN PORTUGAL

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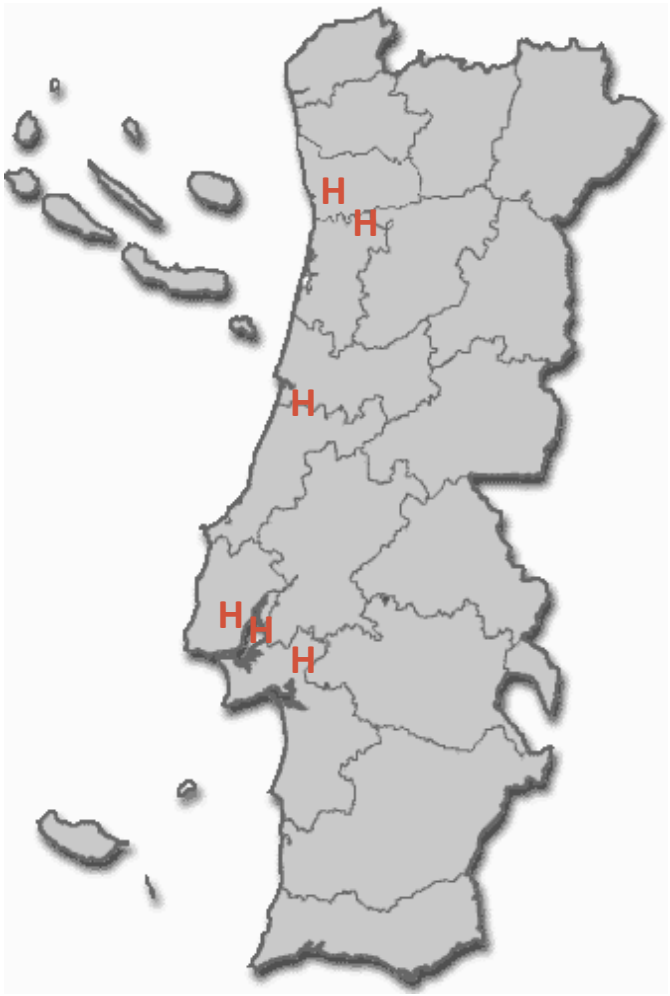
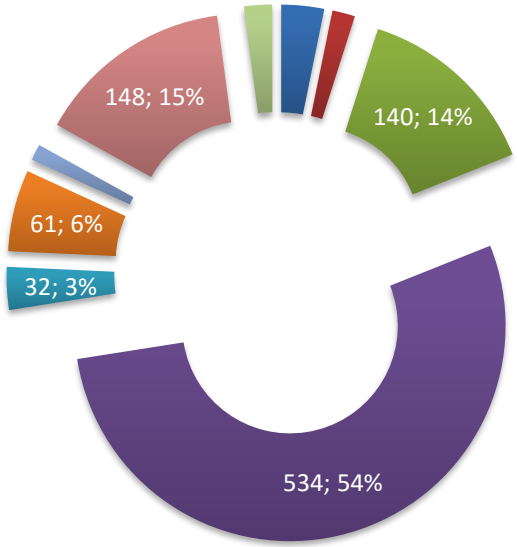




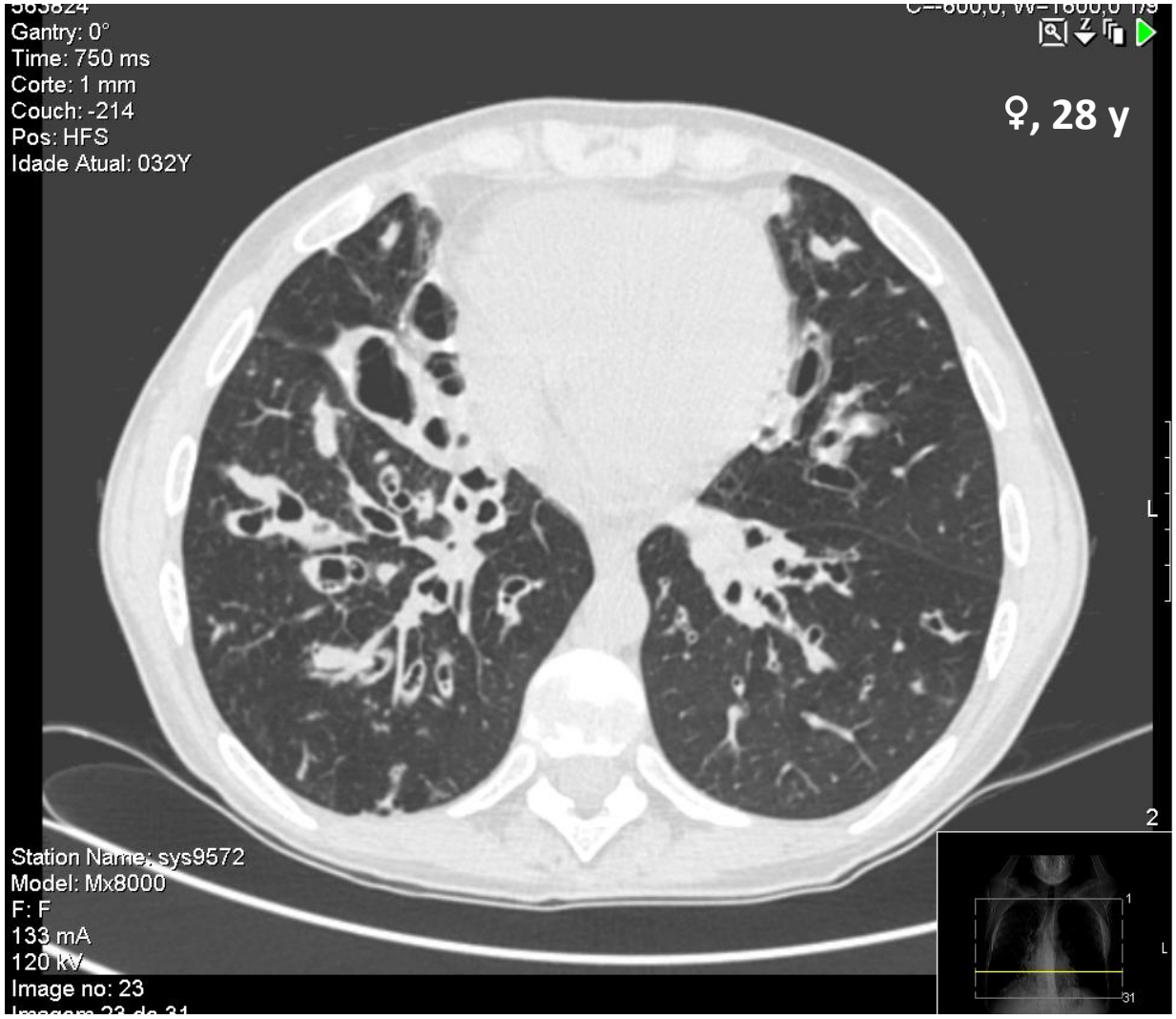
N=997

- SCID
- CID
- COMBINED SYNDROMIC
- ANTIBODIES
- IMMUNEDYSREGULATION
- PHAGOCYTE
- INNATE
- AUTO-INFLAMMATORY
- COMPLEMENT

REGISTRY

- SINCE 1998: NATIONAL REGISTRY- REPORID
- ESID REGISTRY
- JEFFREY MODELL FOUNDATION

PIDs IN PORTUGAL



CVID
Bronchiectasis

LATE DIAGNOSIS
INADEQUATE TREATMENT

PIDs IN PORTUGAL

No newborn screening & **Universal BCG at birth**

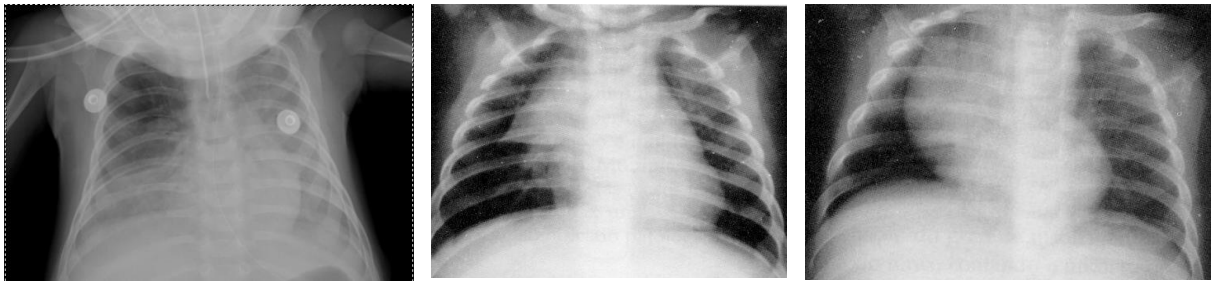
Human Nude Phenotype

FOXN1 deficiency

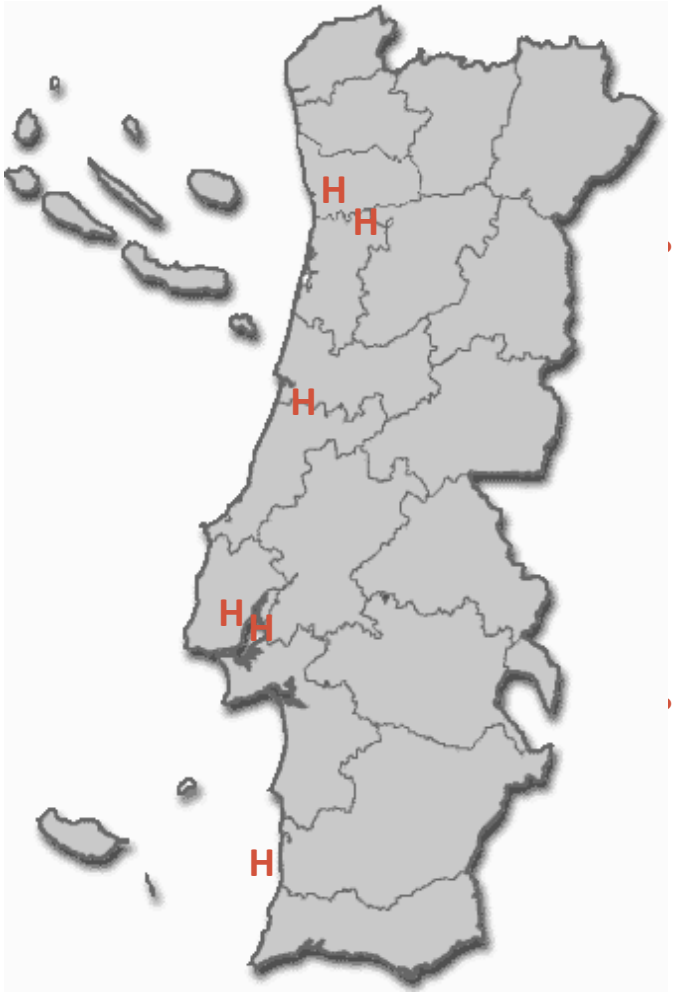
Thymus Transplantation

X-SCID (γC defect)

HSC Transplantation



PIDs IN PORTUGAL



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IMUNOGLOBULIN REPLACEMENT THERAPY

- IVIG- 79 PATIENTS
- SCIG- 73 PATIENTS
- 36% children (N=56)

HEMATOPOIETIC STEM CELL TRANSPLANTATION

- First HSCT in Portugal: 1994 (LAD1)
- 39 patients underwent HSCT for PID since 1994
- 10/39 deaths (26%)
- 1 haploidentical HSCT

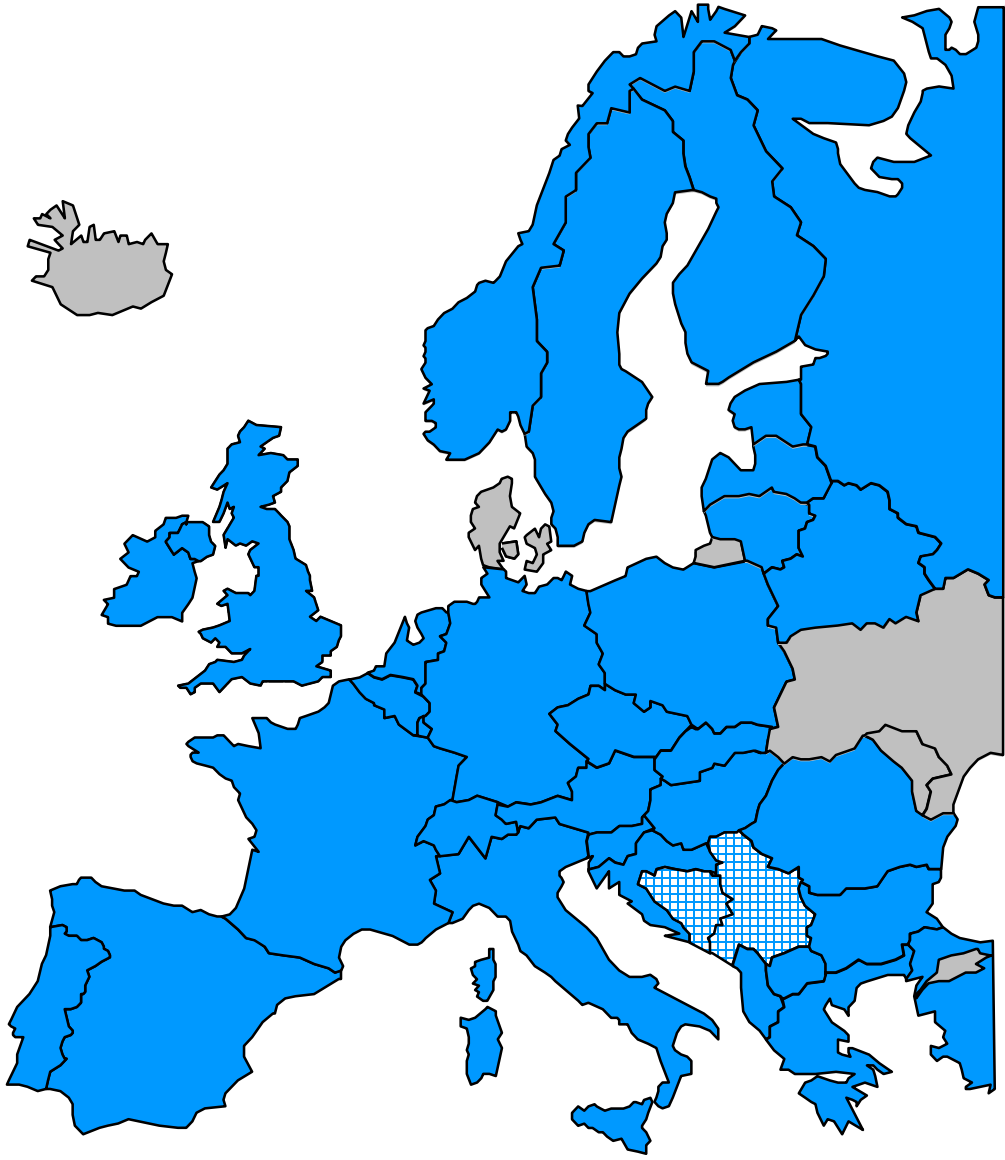
PIDs IN PORTUGAL

EUROPE 2014

IVIG 2014

- full availibility
- limitation
- no availibility
- no response

A. Sediva et al. ESID 2014



PIDs IN PORTUGAL

EUROPE 2014

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- full availibility
- limitation
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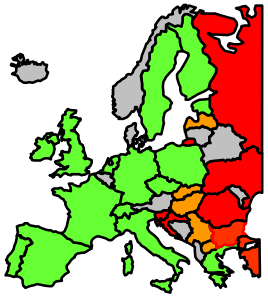
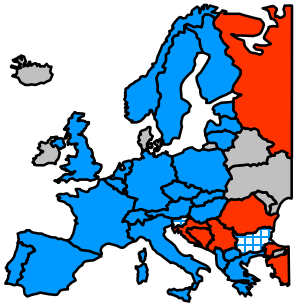
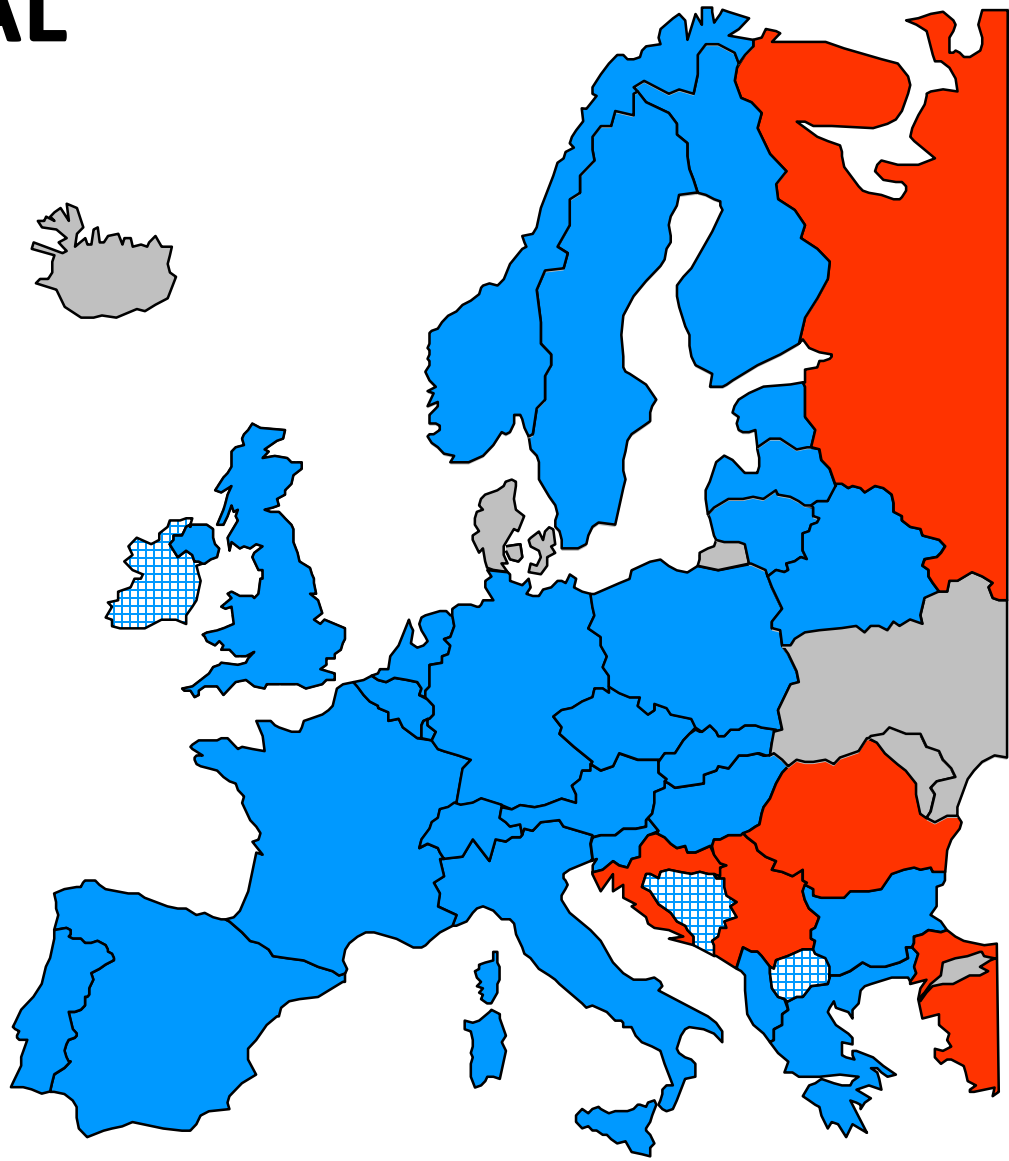
SCIG 2011



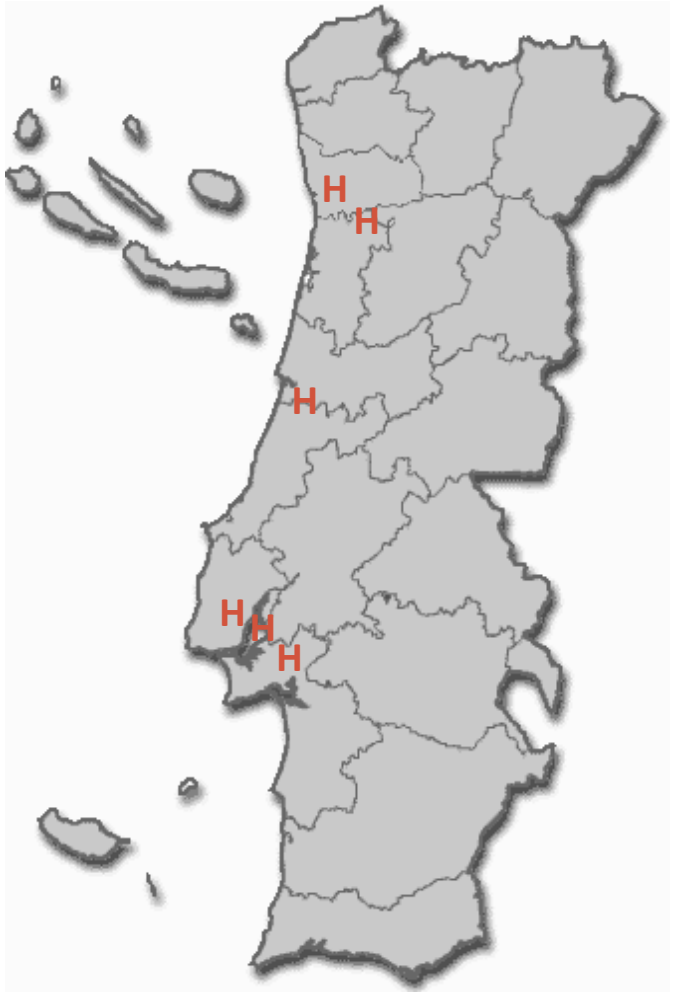
SCIG 2013





A. Sediva et al. ESID 2014

PIDs IN PORTUGAL



MEDICAL STAFF

FELLOWSHIPS IN WELL-RECOGNIZED PID CENTRES

- LONDON
- OXFORD
- NEWCASTLE
- PARIS
- BARCELONA
- SÃO PAULO

PARTICIPATION IN INTERNATIONAL MULTICENTRIC PROJECTS

- EU CALL ON PUBLIC HEALTH (E. GAMBINERI)
- European Network for Immune Deficiencies (S. NEJENTSEV)

Centro de Imunodeficiências Primárias, Lisboa

MM Lisboa Instituto de Medicina Molecular

Faculdade de Medicina Universidade de Lisboa

Hospital de Santa Maria

Centro de Imunodeficiências Primárias

FMUL/IMM/CHLN

Mission

- Diagnosis and Management of PID Patients
- Teaching, Training and Dissemination of Knowledge
- Research
- Interaction with Society

Formally signed protocol by Faculdade de Medicina de Lisboa (FMUL), Instituto de Medicina Molecular (IMM) and Hospital de Santa Maria (HSM), 2007, involving the following departments:

Clínica Universitária de Pediatria (Director: Prof M. do Céu Machado)
Serviço de Imunoalergologia (Director: Prof M Pereira Barbosa)
Clínica Universitária de Medicina II (Director: Prof Rui M Victorino)
Serviço de Genética Clínica (Director: Dra Isabel Cordeiro)
Serviço de Hematologia (Director: Prof J Alves do Carmo)
Unidade de Imunologia Clínica IMM (Director: Prof Ana E. Sousa)
Laboratório de Imunologia Clínica FMUL (Director: Prof Rui M Victorino)

PIDs IN PORTUGAL

INCREASING AWARENESS

PIDs IN PORTUGAL

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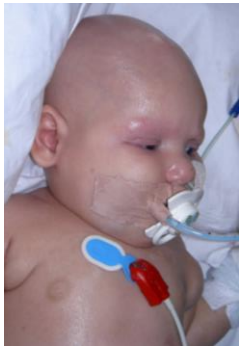
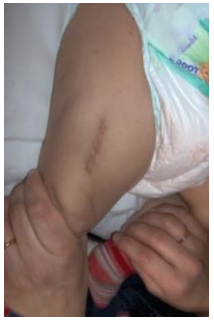


blood

2011117: 688-696
doi:10.1182/blood-2010-06-292490 originally published online October 26, 2010

First use of thymus transplantation therapy for FOXN1 deficiency (nude/SCID): a report of 2 cases

M. Louise Markert, José G. Marques, Bénédicte Neven, Blythe H. Devlin, Elizabeth A. McCarthy, Iv K. Chinn, Adriana S. Albuquerque, Susana L. Silva, Claudio Pignata, Geneviève de Saint Basile, R M. Victorino, Capucine Picard, Marianne Debre, Nizar Mahlaoui, Alain Fischer and Ana E. Sousa



blood

Prepublished online September 5, 2014;
doi:10.1182/blood-2014-08-591370

Mutations in *TRNT1*, encoding the CCA-adding enzyme, cause congenital sideroblastic anemia with B cell immunodeficiency, periodic fevers and developmental delay (SIFD)

Pranesh K. Chakraborty, Klaus Schmitz-Abe, Erin K. Kennedy, Hapsatou Mamady, Turaya Naas, Danielle Durie, Dean R. Campagna, Ashley Lau, Anoop K. Sendamarai, Daniel H. Wiseman, Alison May Stephen Jolles, Philip Connor, Colin Powell, Matthew M. Heeney, Patricia-Jane Giardina, Robert J. Klaassen, Caroline Kannengiesser, Isabelle Thuret, Alexis A. Thompson, Laura Marques, Stephen Hughes, Denise K. Bonney, Sylvia S. Bottomley, Robert F. Wynn, Ronald M. Laxer, Caterina P. Minniti, John Moppett, Victoria Bordon, Michael Geraghty, Paul B.M. Joyce, Kyriacos Markianos, Adam M. Rudner, Martin Holcik and Mark D. Fleming

Pyogenic Bacterial Infections in Humans with MyD88 Deficiency

Horst von Bernuth,[1,2] Capucine Picard,[1,2,3] Zhongbo Jin,[4,5] Rungnapa Pankla,[4,6] Hui Xiao,[7] Cheng-Lung Ku,[1,2] Maya Chrabieh,[1,2] Imen Ben Mustapha,[1,2,8] Pegah Ghandil,[1,2] Yildiz Camcioglu,[9] Júlia Vasconcelos,[10] Nicolas Sirvent,[11] Margarida Guedes,[10] Artur Bonito Vitor,[12] María José Herrero-Mata,[13] Juan Ignacio Aróstegui,[14] Carlos Rodrigo, Laia Alsina,[16] Estibaliz Ruiz-Ortiz,[13] Manel Juan,[14] Claudia Fortuny,[16] Jordi Yagüe,[14] Jordi Antón ,[16] Mariona Pascal,[14] Huey-Hsuan Chang,[17] Lucile Janniere,[1,2] Yoann Rose,[1,2] Ben-Zion Garty,[18] Helen Chapel,[19] Andrew Issekutz,[20] László Maródi,[21] Carlos Rodriguez-Gallego,[22] Jacques Banchereau,[4] Laurent Abel,[1,2] Xiaoxia Li,[7] Damien Chaussabel,[4] Anne Puel,[1,2] Jean-Laurent Casanova[1,2,23*]

Human Molecular Genetics, 2011, Vol. 20, No. 8 *1509–1523*
doi:10.1093/hmg/ddr029
Advance Access published on January 25, 2011

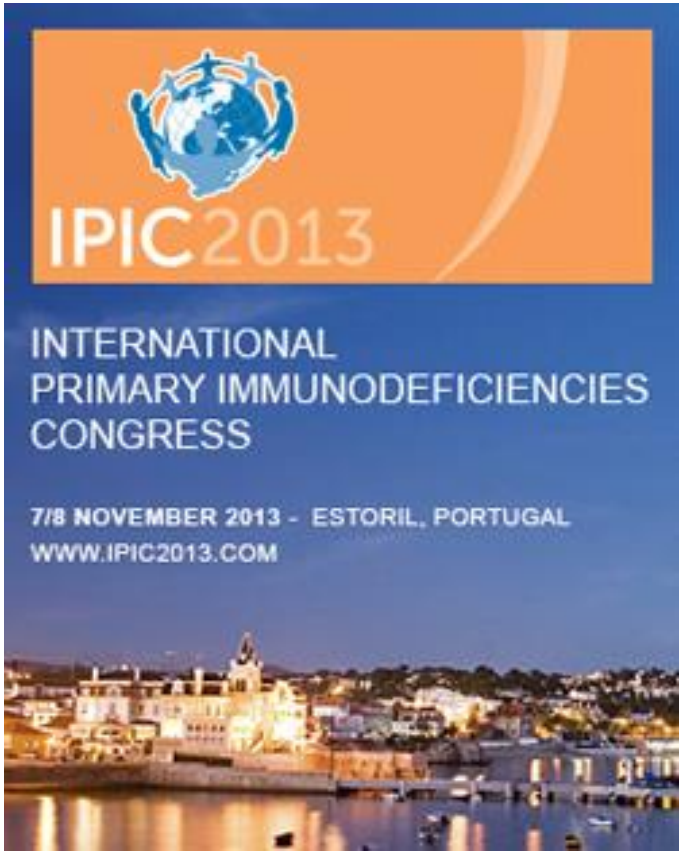
Hemophagocytic lymphohistiocytosis in two patients with underlying IFN-γ receptor-deficiency

Bianca Tesi[1,2,*]MD, Elena Sieni[3,*]MD, Conceição Neves[4], Francesca Romano[5],Valentina Cetica[3]PhD, Ana Isabel Cordeiro[4]MD, Samuel Chiang[6]MSc, Heinrich Schlums[6]MSc, Luisa Galli[7]MD, Stefano Avenali[8]MD, Annalisa Tondo[3]MD, Clementina Canessa[5]MD, Jan-Inge Henter[1]MD,PhD, Magnus Nordenskjöld[2]MD,PhD, Amy P. Hsu[9], Steven M. Holland[9]MD, Joao F. Neves[4,*]MD, Chiara Azzari[5,*]MD,PhD, Yenan T. Bryceson[6,*]PhD

Partial recessive IFN-γR1 deficiency: genetic, immunological and clinical features of 14 patients from 11 kindreds

Ithaisa Sologuren[1,†], Stéphanie Boisson-Dupuis[4,5,†], Jose Pestano[6,‡], Quentin Benoit Vincent[4,‡], Leandro Fernández-Pérez[7,‡], Ariane Chapgier[4], María Cárdenes[1,9], Jacqueline Feinberg[4], M. Isabel García-Laorden[1], Capucine Picard[4,10], Esther Santiago[1], Xiaofei Kong[5], Lucile Jannière[4], Elena Colino[12], Estefanía Herrera-Ramos[1], Adela Francés[13], Carmen Navarrete[16], Stéphane Blanche[11], Emilia Faria[17], Paweł Remiszewski[19], Ana Cordeiro[18], Alexandra Freeman[20], Steven Holland[20], Katia Abarca[21], Mónica Valerón-Lemaur[14], José Gonçalo-Marques[22], Luisa Silveira[23], José Manuel García-Castellano[2,15], José Caminero[3], José Luis Pérez-Arellano[8,13], Jacinta Bustamante[4], Laurent Abel[4], Jean-Laurent Casanova[4,5,11,¶] and Carlos Rodríguez-Gallego[1,8,9,*,¶]

PIDs IN PORTUGAL



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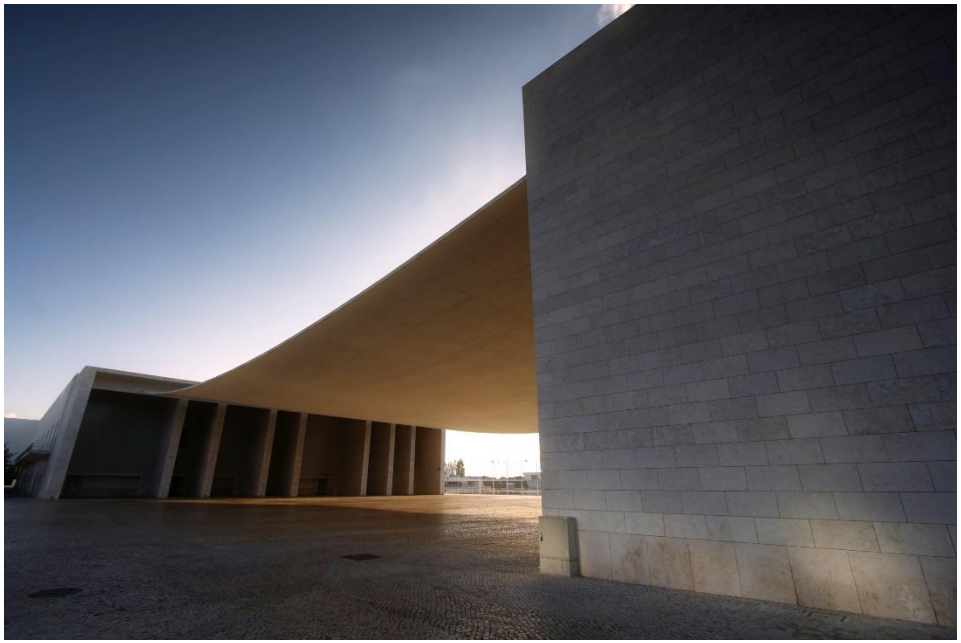
ESID LISBOA 2018



PIDs IN PORTUGAL

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ESID LISBOA 2018

EXPECTATIONS

- Increase of Public, Political and Health Care Workers arousal for PID
- Reinforcement for the implementation of:
  - SCID Newborn screening
  - Establishment of a SINGLE HSCT Centre for PID
  - Investment on both clinical and basic investigation

6th IPOPI PID FORUM:
Principles of Care for Primary Immunodeficiencies

Managing PID diagnosis and care in all countries

Susana Lopes da Silva, MD

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