

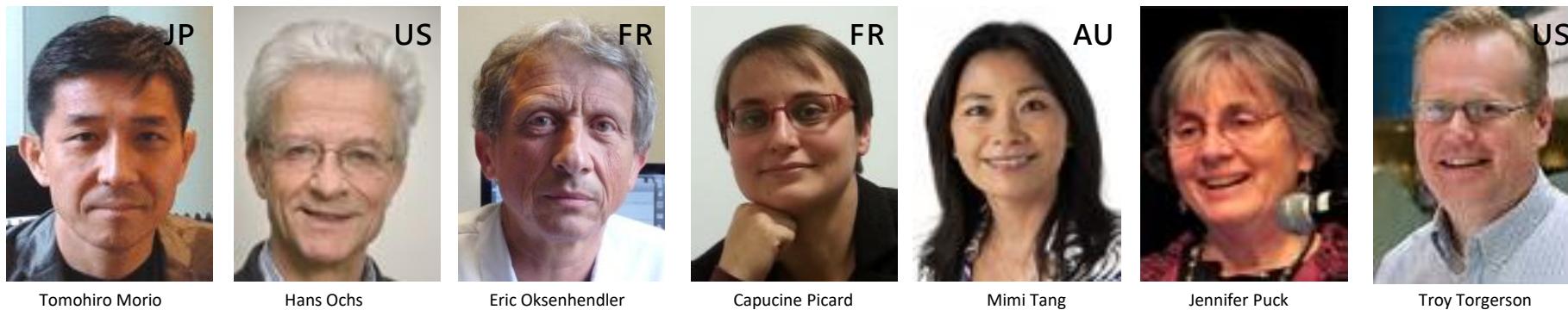
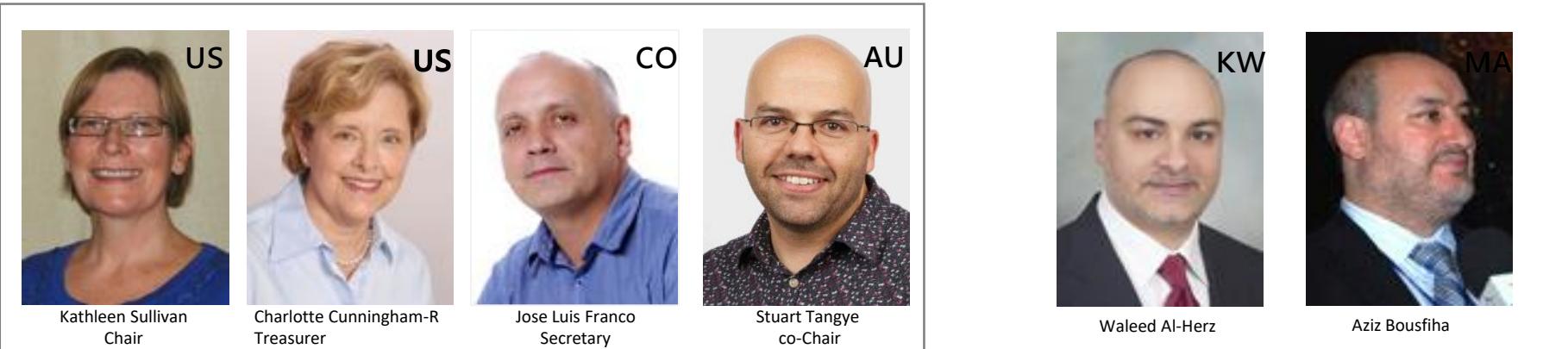
IUIS

Inborn Errors of Immunity Committee (IEI)

Primary Immunodeficiency



Inborn Errors of Immunity: The Expert Committee on Primary Immunodeficiency



Primary
Immuno-
deficiency
Expert
(PID)

*To advance knowledge and expertise on
primary immunodeficiencies worldwide
We:*

- *Provide an up-to-date classification of all primary immunodeficiency diseases (PIDs)*
- *Assist with the identification, diagnosis and management of patients with these uncommon conditions*
- *Support diagnostic and therapeutic guidelines developed by national societies and others, to assist healthcare providers*
- *Promote awareness, diagnosis and treatment of PIDs in all regions of the world*
- *Produce ad hoc reports on any aspect of PIDs and to assist in the welfare of patients with these conditions*

Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015

Capucine Picard^{1,2} · Waleed Al-Herz^{3,4} · Aziz Bousfiha⁵ · Jean-Laurent Casanova^{1,6,7,8,9} ·
Talal Chatila¹⁰ · Mary Ellen Conley⁶ · Charlotte Cunningham-Rundles¹¹ ·
Amos Etzioni¹² · Steven M. Holland¹³ · Christoph Klein¹⁴ · Shigeaki Nonoyama¹⁵ ·
Hans D. Ochs¹⁶ · Eric Oksenhendler^{17,18} · Jennifer M. Puck¹⁹ · Kathleen E. Sullivan²⁰ ·
Mimi L K. Tang^{21,22,23} · Jose Luis Franco²⁴ · H. Bobby Gaspar²⁵

The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies

Aziz Bousfiha¹ · Leïla Jeddane¹ · Waleed Al-Herz^{2,3} · Fatima Ailal¹ ·
Jean-Laurent Casanova^{4,5,6,7,8} · Talal Chatila⁹ · Mary Ellen Conley⁴ ·
Charlotte Cunningham-Rundles¹⁰ · Amos Etzioni¹¹ · Jose Luis Franco¹² ·
H. Bobby Gaspar¹³ · Steven M. Holland¹⁴ · Christoph Klein¹⁵ ·
Shigeaki Nonoyama¹⁶ · Hans D. Ochs¹⁷ · Eric Oksenhendler^{18,19} ·
Capucine Picard^{5,20} · Jennifer M. Puck²¹ · Kathleen E. Sullivan²² · Mimi L. K. Tang^{23,24,25}

*The biannual meeting was held February 22-23 in London (UK)
Work products include:*

- *Updated PID Classifications Tables*
- *Online database of PIDs discussed and developed with an app for cell phones*
- *New member elected: Tomohiro Morio (Japan)*
- *Revised PID Classification highlighting 353 Inborn Errors of Immunity (in press: Journal of Clinical Immunology)*
- *Revised Phenotypic Approach for IUIS PID Classification and Diagnosis: Guidelines for Clinicians at the Bedside (In press: Journal of Clinical Immunology)*
- *OTHER: New Co-chair elected: Stuart Tangye (Australia)*

Conferences promoting the PI-EC of the IUIS World-wide



Christoph Klein:

- 8th International Symposium (joint meeting EHA and Japanese Society of Hematology), Miyazaki, Japan. May, 2017.
- Kathleen Sullivan:
- Georgia Society of Allergists, Atlanta, GA. May, 2017.
- WAO Symposium and XI Congreso Asociación Colombiana de Asma, Alergia e Inmunología, Cartagena, Septiembre, 2017.

Waleed Al-Herz:

- 3rd international congress of immunology, asthma, and allergy. Tehran, Iran. Feb 2017.

Stuart Tangye:

- Western Sydney University, Macarthur Series in Immunology. March 2017.
- Royal College of Pathologists of Australia, 23rd Immunopathology Course, May 2017.

Jean Laurent Casanova:

- Tisdale Lecturer, Department of Medicine, University of Vermont (M Poynter), Burlington, VT, USA, January, 2017.
- Talking Science Lecture, The Rockefeller University, New York, NY, USA. January, 2017
- International Congress of Immunology, Asthma and Allergy (M Moin & M Ayazi), Tehran, Iran, February, 2017.
- Frontiers in Science (A Schwartz), Tempe, AZ, USA, March, 2017.
- 26th Annual Henry Kunkel Society Meeting, The Rockefeller University, New York, NY, USA. March, 2017
- 4th International Conference on Primary Immunodeficiency Diseases (S Gupta, JL Casanova, L Hammarstrom), Bengaluru, India, March 2017
- Beutler Institute Lectures, Beutler Institute, Xiamen, China. March, 2017.
- AAI-Steinman Award for Human Immunology Research Lecture (AH Sharpe), AAI Immunology 2017, Washington, DC, USA. May, 2017.

Charlotte Cunningham Rundles:

- 2017 TAAIS Annual Meeting, San Antonio Tx USA. April, 2017.
- 2017 PID Annual Conference, CIS meeting Seattle, WA. April, 2017.

Jose Luis Franco:

- VIII Congreso de la Sociedad Peruana de Inmunología y Centro Nacional de Alergias, Asma e Inmunología. Lima, Peru. September, 2016.
- VI Curso Internacional de Inmunología Pediátrica , Cali, Colombia. November., 2016.
- VII Simposio Nacional de Inmunodeficiencias Primarias, Bogotá, Colombia. Abril, 2017
- Hospital Pablo Tobon Uribe, Round Cases. Medellín, Colombia, March, 2017
- World Allergy Organization (WAO) Symposium and XI Congreso Asociación Colombiana de Asma, Alergia e Inmunología (ACAAI), Cartagena, Septiembre, 2017.

FUNDACIÓN FIP COLOMBIA 10 AÑOS



A brief history of Primary Immunodeficiencies

Una Breve historia de las Inmunodeficiencias Primarias

Hans D. Ochs, MD*; Amos Etzioni, MD*

Primary Immunodeficiencies (PIDs) is a relatively new field, but it has its roots in the late 19th and early 20th century Europe. The new idea of taking an experimental approach to biology was fundamental to establishing cellular pathology, the microbial basis of infectious diseases, adaptive and innate immunity, active and passive immunization, and to understand the composition of proteins and DNA, and the Mendelian laws of inheritance [1]. These efforts were recognized when the scientific establishment awarded the Nobel prize in Physiology and Medicine to Doctor Emil von Behring in 1901, and to Doctors Elie Metchnikoff and Paul Ehrlich in 1908. While Behring introduced passive immunization against diphtheria and tetanus, Ehrlich proposed the side chain theory, which postulates that "toxins" (now classified as antigens) were recognized and fixed by cell surface molecules (side chains, equivalent to the B cell receptor), protruding from the cell wall; Ehrlich suggested that toxins and side chains combine like a lock and key, stimulating the cell to produce more specific side chains (antibodies). Metchnikoff recognized that cellular components (phagocytes) contribute equally to the defense against microorganisms [2].



*Hans Dieter Ochs (izq.) es médico pediatra e inmunólogo profesor de pediatría de la División de Inmunología en el Departamento de Pediatría de la Facultad de Medicina (Universidad de Washington) y Co-director del Centro de diagnóstico inmunológico del Seattle's Children Research Institute (Seattle, Estados Unidos).

*Amos Etzioni (der.) es médico pediatra y profesor de Pediatría e Inmunología en la Facultad de Medicina Technion en la Universidad de Haifa y director del Children's Hospital en el Rambam Health Campus (Haifa, Israel). Son los editores del libro : "Primary Immunodeficiency Disorders: A Historic and Scientific Perspective".

Ehrlich propuso la teoría de la cadena lateral, la cual postula que las "toxinas" (actualmente conocidas como antígenos) fueron reconocidas y fijadas por moléculas de la superficie celular (cadenas laterales, equivalentes al receptor de linfocitos B), sobresaliendo de la pared celular. Ehrlich sugirió que las toxinas y las cadenas laterales se combinan como una cerradura y una llave, estimulando a la célula a producir cadenas laterales más específicas (anticuerpos). Por otra parte, Metchnikoff reco-

Las Inmunodeficiencias Primarias (IDPs) son un campo relativamente nuevo, sin embargo tienen sus raíces en la Europa fines del siglo XIX y principios del siglo XX. La nueva idea de adoptar un enfoque experimental en la biología fue fundamental para establecer la patología celular, las bases microbianas de las enfermedades infecciosas, la inmunidad innata y adaptativa, la inmunización activa y pasiva, y comprender la composición de las proteínas y el ADN y las leyes mendelianas de la herencia [1]. Estos esfuerzos fueron reconocidos cuando la comunidad científica otorgó el premio Nobel en Fisiología y Medicina al Doctor Emil von Behring en 1901, y a los Doctores Elie Metchnikoff y Paul Ehrlich en 1908. Mientras von Behring introdujo la inmunización pasiva contra la difteria y el tétanos, Ehrlich sugirió que las toxinas y las cadenas laterales se combinan como una cerradura y una llave, estimulando a la célula a producir cadenas laterales más específicas (anticuerpos). Por otra parte, Metchnikoff reco-

addition to susceptibility to infections [18, 19]. In some of these entities, immune dysregulation and autoimmunity are the predominant findings and may appear before infections. Some syndromes of immune dysregulation are caused by malfunction in response to specific immune suppression, as has been illustrated by gain of function (GOF) mutations in STAT1 and STAT3. As the PIDs field has progressed and new sophisticated techniques became available, syndromes with infections limited to a narrow spectrum of microorganisms were recognized, including patients with susceptibility to atypical mycobacteria, such as BCG [20], individuals susceptible to EBV [21], to invasive infections with *S. pneumoniae* and *S. aureus* during childhood [22], or to *Candida* infections.

Exploring the historic events leading to the identification of patients with PIDs, and to the recognition of the molecular basis of these disorders, followed by the discovery of effective therapies is an exciting, but simultaneously humbling experience. Immunology, as we learn and teach it in the 21st century, can be traced to a small group of investigators who developed a hypothesis-driven and evidence-based understanding of immunology. The passion to identify microbes, design strategies for the control of infections, to understand immune mechanisms, to decipher and to read the human genome has, in the final analysis, been driven by the commitment of those participating in this multi-generational story to conquer disease and improve the life of human beings. The reward for these efforts is the enthusiasm of our PID patients, who have formed their own organizations, support PIDs-focused meetings and help provide funding to ensure that the momentum driving progress in these diseases is maintained.

en IDPs avanzaron y se desarrollaron nuevas técnicas, comenzaron a reconocerse pacientes afectados por síndromes con infecciones limitadas a un espectro estrecho de microorganismos, incluidos la susceptibilidad a micobacterias atípicas como la *Mycobacterium bovis* BCG [20], infecciones por virus de Epstein-Barr [VEB], infecciones bacterianas invasivas por *S. pneumoniae* y *S. aureus* en la infancia [22], o por hongos como la *Candida*.

Explorar los eventos históricos que llevaron a la identificación de pacientes con IDPs y al reconocimiento de las bases moleculares de estas enfermedades, seguido por el descubrimiento de terapias eficaces ha sido una experiencia emocionante e igualmente sobrecogedora. La inmunología, tal como la aprendemos y la enseñamos en el siglo XXI, puede ser trazada a un pequeño grupo de investigadores que desarrollaron una comprensión de la inmunología basada en la hipótesis y la evidencia. La pasión por identificar microbios, diseñar estrategias para el control de las infecciones, comprender los mecanismos inmunológicos y descifrar y leer el genoma humano han sido en última instancia, impulsados por el compromiso de los participantes en esta historia multigeneracional de entender y superar estas enfermedades y así mejorar la vida de los seres humanos. La recompensa a estos esfuerzos es el entusiasmo de nuestros pacientes con IDPs quienes han formado sus propias organizaciones, apoyan reuniones centradas en IDPs y ayudan a proveer los fondos para asegurar que el momento que impulsa el progreso del estudio de estas enfermedades se mantenga.

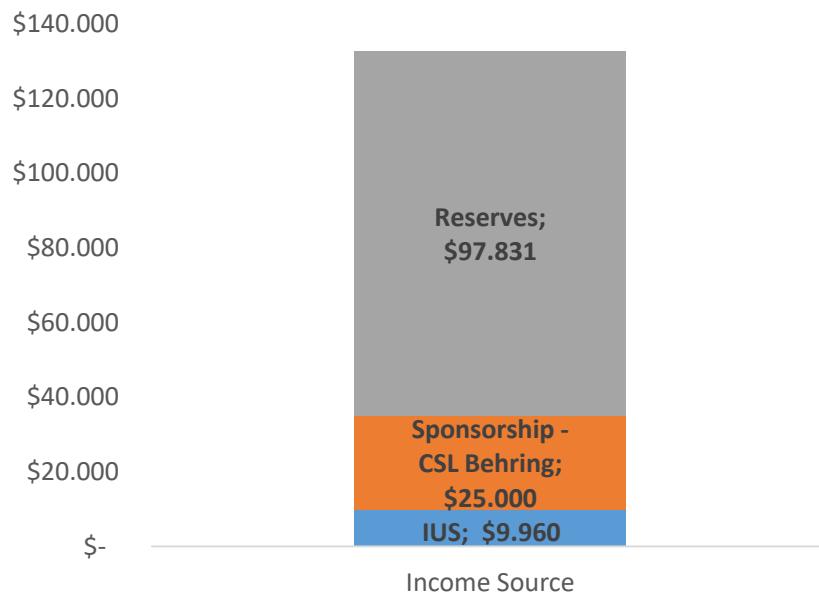


Comité de Expertos en Inmunodeficiencias Primarias de la Unión Internacional de Sociedades Inmunológicas (IUIS), Londres 2017. De izquierda a derecha arriba: Jean Laurent Casanova, Yanick Crow, Bobby Gaspar, Christoph Klein, Talal Amine Chatila, Aziz Boushifia, Amos Etzioni, Mimi Tang, Eric Oksenhendler, Hans Dieter Ochs, Waleed Al-Herz. Abajo: Stuart Tangye, Jennifer Puck, José Luis Franco, Charlotte Cunningham-Rundles, Kathleen Sullivan, Capucine Picard y Troy Troquerson (Steven Holland y Shigeaki Nonoyama no presentes en la foto).

PID 2016-2018 Budgets



	FUNDING SOURCE (\$)						
	Income Source	Annual PID Meeting	Administration Fees	HPO Workshop Attendance	Barcelona Meeting	TOTAL spent to date	TOTAL remaining
IUIS	\$ 9'960		9960			9960	0
Sponsorship - CSL Behring	\$ 25'000	18353	4355	816	1476	25000	0
Reserves	\$ 97'831	10245				10245	87586
TOTAL	132791.08		14315	816	1476	45205	87586



Challenges:

- *Funding: Successful partial funding for the online database. Look for more funding.*
- *Post a searchable list online at the IUIS website.*
- *Develop a cellphone app for PID.*
- *Extend our publications to other types (editorials).*

Opportunities 2018:

Move to digital content to update the list of PID more frequently.

Implement strategies to interact with other IUIS committees to seek for synergistic opportunities.

PID CLASSIFICATION APP

