

## A Symposium for Researchers and Clinicians on

**XLP** X-Linked  
Lymphoproliferative  
Disease



**WAS** Wiskott Aldrich Syndrome  
and X-Linked  
Thrombocytopenia

**3<sup>rd</sup>-4<sup>th</sup> November 2014 - LONDON**

[www.xlp-was2014.org](http://www.xlp-was2014.org)

### Symposium Plan

#### 3<sup>rd</sup> of November 2014

##### 08:45 – 09:15 Gather together and On-site Registration

##### 09:15 – 09:30 Introduction

David Hartley, The XLP Research Trust and Professor Bobby Gaspar, UCL Institute of Child Health, UK  
Sumathi Iyengar- Amir Kedar, WAS organizations and Professor Adrian Trasher, UCL Institute of Child Health, UK

##### 09:30 – 10:00 - Plenary Session A (XLP – WAS)

**Prof. Luigi Notarangelo** – Harvard Medical School, Boston, USA.

**"The immune dysregulation of WAS: novel insights"**

##### 10:00 – 10:30 - Plenary Session B (XLP – WAS)

**Prof. Stuart Tangye** – Garvan Institute of Medical Research, NSW, Australia.

**"XLP: 1974 to the present day"**

##### 15 minute talks with 5 minutes discussion after each talk

##### 10:40 – 11:40 -First WAS Session

**Chair: Adrian Thrasher**

**10:40–11:00 Prof. Hans Ochs** – Seattle Children's, USA.

"Reversion of WAS gene mutation: Does it affect immune function, and clinical phenotype?"

**11:00–11:20 Prof. Silvia Giliani** – University of Brescia, Italy

"Low platelet counts in female carriers of WAS mutations showing high level of X-inactivation of the non-mutated chromosome in hematopoietic precursors and myeloid-derived leukocytes"

**11:20–11:40 Dr. Pamela Lee** – Queen Mary Hospital, Hong Kong.

"Improving awareness and genetic diagnosis of Wiskott-Aldrich syndrome in Asia: A multi-centered collaborative study on 90 patients from the Asian Primary Immunodeficiency (APID) Network"

##### 11:40 – 12:10 - Break

##### 12:10 – 13:30 - Second WAS Session

**Chair: Siobhan Burns**

**12:10–12:30 Dr. Pamela Lee** – Queen Mary Hospital, Hong Kong.

"Wiskott-Aldrich Syndrome Protein Targets Enteropathogens to Autophagy and Limits Inflammasome Activation"

**12:30–12:50 Dr. Junfeng Wu** – Children's Hospital of Chongqing Medical University, China.

"TCR diversity is selectively skewed in T-cell populations of Wiskott-Aldrich syndrome patients"

**12:50–13:10 Prof. Silvia Giliani** – University of Brescia, Italy.

"Influence of WAS gene alterations on late stages of human B cell differentiation"

**13:10–13:30 Dr. Brian Davis** – University of Texas, USA.

"Genetic correction of induced pluripotent stem cells from a Wiskott-Aldrich Syndrome patient normalizes the immune defects"

##### 10:40 – 11:40 - First XLP Session

**Chair: Bobby Gaspar**

**10:40–11:00 Dr Hirokazu Kanegane** – University of Toyama, Japan.

"Whole exome sequencing reveals atypical phenotype of X-linked lymphoproliferative syndrome"

**11:00–11:20 Prof Stuart Tangye** – Garvan Institute of Medical Research, NSW, Australia.

"Primary immunodeficiencies as models to delineate requirements for anti-EBV immunity"

**11:20–11:40 Prof Colin Duckett** – University of Michigan Medical School, USA.

"Why do defects in XIAP, a protein with many cellular roles, lead to XLP2, and how can this guide our treatment of the disease"

##### 12:10 – 13:30 - Second XLP Session

**Chair: Claire Booth**

**12:10–12:30 Dr Andrew Snow** – University of the Health Sciences, Bethesda, MD, USA.

"SAP facilitates recruitment and activation of LCK at NTB-A receptors during restimulation-induced cell death"

**12:30–12:50 Dr Pamela Schwarzborg** – National Human Genome Research Institute, NIH, USA

"Defective Lymphocyte Interactions in XLP and other Primary Immunodeficiencies"

**12:50–13:10 Dr Philipp Jost** – Technical University of Munich, Germany.

"XIAP restricts TNF- and RIP3-dependent cell death and inflammasome activation"

**13:10–13:30 Prof. Andrea Graziani** – University of Piemonte Orientale, Novara, Italy

"Inhibition of Diacylglycerol kinase alpha rescues TCR-induced diacylglycerol signaling and restimulation induced cell death in XLP lymphocytes"

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**13:30 – 14:30 - Lunch**

**14:30 – 16:10 - Third WAS Session**

**Chair: Austen Worth**

**14:30 – 14:50 Dr. Mira Barda Saad** – Bar Ilan University, Israel.

"The negative regulation of the Wiskott-Aldrich syndrome protein"

**14:50 – 15:10 Prof. Ramesh Narayanaswamy** - Harvard Medical School, Boston, USA.

"Assembly Binding of the WASP/N-WASP interacting protein WIP to actin regulates focal adhesion and adhesion"

**15:10 – 15:30 Dr. Steve Thomas** -University of Birmingham, UK.

"Platelet actin nodules are podosome-like structures dependent on Wiskott-Aldrich Syndrome protein and Arp2/3 complex"

**15:30 – 15:50 Dr. Lucia Sereni** - San Raffaele Scientific Institute, Milan, Italy.

"Evaluation of platelet defect in a new mouse model lacking WASp in the megakaryocytic lineage"

**15:50 – 16:10 Prof. Fabio Candotti** - Division of immunology and Allergy, CHUV, Switzerland.

"New Findings On Platelets And Red Blood Cells (Rbc) In Wiskott-Aldrich Syndrome (WAS)"

**16:15 – 16:45 - Fourth WAS Session**

**Chairs: Sumathi Iyengar, Amir Kedar**

**The WAS Patient perspective**

**16:15 – 16:25 Mrs. Helen Mahony**

**16:25 – 16:35 Mr. Mike Wheeler**

**16:35 – 16:45 Mrs. Rebecca Rolfe**

**16:45 – 17:30 - Plenary Session C (XLP – WAS)**

**Chair Adrian Thrasher**

**Prof. Mike Dustin** – Kennedy Institute of Rheumatology, University of Oxford, UK.

**"The immunological synapse"**

**17:30 – 18:30 – Welcome reception**

*An informal get-together to renew acquaintances and meet new colleagues will be held at the Meeting Venue. All registered participants are invited to join.*

**14:30 – 15:50 - Third XLP Session**

**Chair: TBA**

**14:30 – 14:50 Dr. Sujal Ghosh** - University Hospital Düsseldorf, Germany.

"ITK deficiency - clinical and immunological findings in nine patients"

**14:50 – 15:10 Dr Kim Nichols** - Children's Hospital of Philadelphia, USA.

"Janus kinase inhibition as a novel treatment for hemophagocytic Lymphohistiocytosis"

**15:10 – 15:30 Prof. Rebecca Marsh** - Cincinnati Children's Hospital, USA.

"Reduced Intensity Conditioning Regimens Containing Alemtuzumab, Fludarabine, and Melphalan Are Safe for Patients with XIAP Deficiency"

**15:30 – 15:50 Dr. Troy Messick** – The Wistar Institute, USA.

"Development of small molecule inhibitors of EBV for the treatment of XLP-associated diseases"

**16.15 – 16.45 – Fourth XLP Session**

**Chair David Hartley**

***The XLP Research Trust – moving forward supporting the XLP research community.***

**Open discussion of future plans and direction for the charity.**



## **4<sup>th</sup> of November 2014 (Joined clinical session WAS – XLP)**

### **08:30- 09:00 – Second day registration**

### **10 minute talks with 5 minute discussion after each talk**

#### **09:00- 10:00- HSCT session**

**Chair: Bobby Gaspar**

- Advances in HSCT – newer chemotherapeutic agents, prevention and treatment of GvHD, haplo-identical transplant, Reduced Intensity Conditioning, mixed chimerism
- Long term outcome of HSCT

**09:00 – 09:15 Dr. Paul Veys** - Great Ormond Street Hospital for Children, London, UK  
"Recent advances in stem cell transplantation for immunodeficiency diseases"

**09:15 – 09:30 Prof. Lisa Filipovich** - Cincinnati Children's Hospital, USA.  
TBA

**09:30 – 09:45 Dr. Philipp Agyeman** - Great North Children's Hospital, Newcastle upon Tyne, UK.  
"Successful outcome of HSCT for children with WAS using treosulfan and fludarabine – UK experience"

**09:45 – 10:00 Dr. Michael Albert** - Dr. von Hauner Childrens Hospital, Munich, Germany.  
"Results of allogeneic stem cell transplantation from matched donors after myeloablative busulfan-based conditioning"

#### **10:00 - 11:00 - Clinical manifestation and outcome**

**Chair: TBA**

- Long term outcome for patients, QOL
- New treatments

**10:00 – 10:15 Dr. Kim Nichols** - Children's Hospital of Philadelphia, USA.  
"Treatment of epstein-barr virus-induced hemophagocytic lymphohistiocytosis with rituximab containing chemo-immunotherapeutic regimens"

**10:15 – 10:30 Prof. Anna Shcherbina** - Federal Research and Clinical Center, Moscow, Russia.  
"Treatment of thrombocytopenia in Wiskott-Aldrich Syndrome"

**10:30 – 10:45 Dr. Nizar Mahlaoui** - Centre de Référence National Déficits Immunitaires Héritaires (CEREDIH), Hôpital Necker-Enfants, Paris, France.  
TBA

**10:45 – 11:00 Dr. Michael Albert** - - Dr. von Hauner Childrens Hospital, Munich, Germany.  
"Effect of splenectomy, stem cell transplantation, gene therapy or no definite treatment on frequency of disease related complications and physician-perceived quality of life in WAS"

### **11:00– 11:30 - Break**



**11:30 – 13:30 - Gene Therapy**

**Chair: Adrian Thrasher**

- Advances in GT – Site specific gene corrections, Zinc finger nucleases, non-viral techniques
- Outcome of GT

**11:30 – 11:45 Dr. Maximilian Witzel** - The Gene Center of the University of Munich, Germany.

"Haematopoietic Stem Cell Gene Therapy for Wiskott-Aldrich Syndrome- The German trial"

**11:45 - 12:00 Dr. Alessandro Aiuti** - San Raffaele-Telethon Institute for Gene Therapy, Milan, Italy.

"Lentiviral gene therapy for WAS"

**12:00 - 12:15 Dr. Anne Galy** - Inserm Unit 951, Genethon, Evry, France.

Clinical Efficacy of Gene Therapy for Severe Wiskott-Aldrich Syndrome: Results of an international study.

**12:15 - 12:30 Dr. Sung Yun Pai** - Harvard Medical school, Boston, USA.

"Gene therapy for WAS: Results from Boston"

**12:30 – 12:45 Dr. Claire Booth** - Great Ormond Street Hospital for Children, London, UK.

"Gene therapy for XLP"

**12:45 – 13:00 Dr. Toni Cathomen** - Medical Center - University of Freiburg, Germany.

TBA

**13:00– 13:30 Open Discussion**

**13:30 – 14:00 - Summary**

Prof. Bobby Gaspar, Prof. Adrian Thrasher, UCL Institute of Child Health, London, UK