**Symposium Plan**

3rd 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 Schwarzberg** - 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"
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

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.

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.
4th 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"

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