



GOSH Immune Dysregulation Day

In the Era of Personalised Medicine

Thursday 23 January 2025, 9:00-17:30 GMT

9:00-9:30	Registration and refreshments for those attending in person (Ryan Connolly Room, Zayed Centre for Research, 20c Guilford Street, London, WC1N 1DZ) Those joining online to log in at 9:28	
9:30-10:00	AIRE clinical indication and treatment approach	Alexandra Kreins , Wellcome Career Fellow, University College London
10:00-10:30	Immune dysregulation in profound combined immunodeficiency	Carsten Speckmann , Consultant in Paediatric Immunology and Hematopoietic Stem Cell Transplantation, University of Freiburg, Germany
10:30-11:15	STAT1/2/3 GOF mutation paediatric and adult experience	Maaïke Kusters , Consultant in Paediatric Immunology, GOSH and Siobhan Burns , Professor in Translational Immunology, University College London
11:15-11:30	Discussion	
11:30-12:00	Break	
12:00-12:30	ALPS and related disorders of immune dysregulation	Koneti Rao , Consultant in Paediatric Haematology, National Institutes of Allergy and Infectious Diseases (NIAID), National Institutes of Health (NIH), Bethesda, USA
12:30-13:00	APDS1/2: what we know and how we treat?	Gulbu Uzel , Consultant in Paediatric Immunology, National Institutes of Allergy and Infectious Disease (NIAID) and National Institutes of Health (NIH), Bethesda, USA
13:00-13:30	Highlights on Laboratory investigations and interpretations for immune dysregulation	Kimberly Gilmour , Director of Laboratory Medicine, GOSH
13:30-14:30	Lunch break	
14:30-15:00	Patient representation(s)	
15:00-15:30	Distinguishing features of different forms of autoimmune lymphoproliferative inborn errors of immunity: data from ESID registry	Mariaelena Maccari , Consultant in Paediatric Immunology and Hematopoietic Stem Cell Transplantation (HSCT), University of Freiburg, Germany
15:30-16:30	Summary of data presented in IEWP 2024 on HSCT in the context of targeted therapy	Carsten Speckmann (as above) and Reem Elfeky , Consultant in Paediatric Immunology, GOSH
16:30-17:00	Nursing role in management of patients with immune dysregulation: challenges and opportunities	Tanja Stankova , Immunology Nurse Specialist, GOSH
17:00-17:30	Closing remarks	
17:30	Finish	

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