

Let's talk about AT*

*Ataxia Telangiectasia



Agenda

25-27 June

 **QUINCE**
THERAPEUTICS



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 ASSOCIAZIONE
NAZIONALE
ATASSIA
TELANGIECTASIA

Wednesday 25.06.2025 | 13:15 – 18:00

MORNING ARRIVAL AND REGISTRATION

13:15	Welcome from Rachel Poupard, Chair of the AT Society
13:30 – 15:00	<u>Diagnostic challenges and unusual/difficult cases of AT</u> Chairs: Asbjorg Pedersen (Oslo University Hospital, Norway) and Maartje Blom (Leiden University Medical Centre, The Netherlands)
13:30	Invited speaker – Michel Willemsen (Radboud University Medical Centre, The Netherlands): The spectrum of neurological involvement in AT
13:50	Mirjam Van Der Burg (Leiden University Medical Centre, The Netherlands): The triangle of immunological challenges of early identification of AT through SCID newborn screening
14:00	Invited speaker – Xiao Peng (Children's Hospital at Montefiore, USA): The AT genetic landscape and considerations for genomic diagnosis
14:20	Liz McDermott (Nottingham University Hospital NHS Trust, UK): Immune deficiency in children diagnosed in infancy and early childhood – recent experience from the UK National AT Clinic
14:25	Nienke Van Os (Radboud University Medical Centre, The Netherlands): Serum alpha fetoprotein in AT: new lessons about an old biomarker
14:30	General session discussion
15:00	Coffee/tea break
15:30 – 17:00	<u>Respiratory medicine and AT</u> Chairs: Martin Lavin (University of Queensland, Australia) and Rosella Abeti (University College London, UK)
15:30	Invited speaker (online) – Sharon McGrath-Morrow (Children's Hospital of Philadelphia, The University of Pennsylvania, USA): Lung function and inflammatory markers in AT
15:50	Invited speaker – Yossi Shiloh (Tel Aviv University School of Medicine, Israel): Respiratory symptoms in AT: exploring the connection between genome instability and cellular senescence
16:10	Corrine Gaston (Royal Papworth Hospital, UK): Piloting Ultrasound Evaluation of Swallowing (USES) as a monitoring tool in individuals with AT
16:20	Manuela Lo Bianco (University of Catania, Italy): Exploring ultrasound measurements for diaphragmatic function in AT
16:30	General session discussion




17:00 - 18:00	<p><u>Insights of clinical relevance</u></p> <p>Chairs: Malcolm Taylor (Institute of Cancer and Genomic Sciences, University of Birmingham, UK) and Giuliana Giardino (University of Naples 'Federico II', Italy)</p>
17:00	Ari Barzilia (Tel Aviv University, Israel): The role of a single metabolite in impaired cerebellar integrity and functionality in ATM deficient mice
17:10	Ramsay Bowden (University of Cambridge and Royal Papworth Hospital, UK): Insights from mutational signature analysis of tumours in AT patients and ATM carriers
17:20	Michele Menotta (University of Urbino, Italy): MiniATM Family Expression in ATTeST: Molecular Insights into Unusual ATM Variants in AT Cells
17:30	Margherita Doria (Babino Gesù' Children's Hospital, Italy): Dysfunctional NK cells add to adaptive immunity defects in AT
17:40	General session discussion
18:00	<u>Networking and poster session 1</u>
19:00	Working group meetings 1: Clinical trial readiness (Room 17), Newborn screening (Room 8)
19:30	<p>Working group meetings 2: Clinical trial readiness continued (Room 17), Improving clinical management (Room 8)</p> <p>For more information about the networking groups, please scan the QR code</p>
	
20:00	Dinner

8:30 – 10:30	<u>Mechanisms causing the disorder AT</u> Chairs: Yossi Shiloh (Tel Aviv University School of Medicine, Israel) and Ramsay Bowden (University of Cambridge and Royal Papworth Hospital UK)
8:30	Invited speaker - Martin Lavin (University of Queensland, Australia): Defective energy metabolism contributes to mitochondrial dysfunction in AT
8:45	Invited speaker - Peter McKinnon (St Jude's Children's Research Hospital, USA): Function of ATM in the Nervous System
9:00	Invited speaker - Tanya Paull (University of Texas at Austin, USA): Transcriptional stress and gene expression changes in AT
9:15	Invited speaker - Kim Chow (Chinese University of Hong Kong): Alpha-Ketoglutarate alleviates mitochondrial glutamine dependency arising from insulin resistance-induced metabolic inflexibility in AT
9:30	Invited speaker - Marco Foiani (IFOM, Milan, Italy): Mechanical vulnerability of Purkinje cells leads to nuclear envelope invaginations, chromatin hypermethylation and mechano-dysfunction in AT patients
9:45	Invited Speaker - Sam Nayler (QIMR Berghofer Medical Research Institute, Queensland, Australia): Cerebella organoids for understanding disease pathogenesis in AT; energy metabolism, neuro-immune crosstalk and therapeutics
10:00	General discussion
10:30 – 11:00	Coffee/tea break
11:00 – 12:30	Further clinical issues affecting adults with AT Chairs: Claudio Pignata (Federico II University, Italy) and Emily Petley (University of Nottingham, UK)
11:00	Invited speaker - Vincenzo Costanzo (IFOM-ETS, Milan, Italy): Understanding and correcting metabolic dysfunctions in ATM-deficient cells
11:20	Invited speaker - Howard Lederman (John Hopkin's University School of Medicine, USA): New problems in older patients: Surprises as we get better at treating immunodeficiency, lung disease and lymphoid malignancies
11:40	William Whitehouse (University of Nottingham, UK): Endocrine and metabolic events in children with AT treated long-term with erythrocyte-delivered Encapsulated Dexamethasone Sodium Phosphate (eDSP)



11:50	<p><u>Critical advances in AT scientific research (Part I):</u></p> <p>Margherita Rossi (IRCCS San Raffaele Institute, Milan, Italy): Harnessing ATM protein trans-splicing to establish an efficient gene-based therapy for AT</p>
12:00	<p>Giuliana Giardino (University of Nales 'Federico II', Italy): Assessing the efficiency of split-inteins mediated protein trans-splicing in correcting the gene defect of AT</p>
12:10	<p>General discussion</p>
12:30 – 13:30	<p>Lunch</p>
13:30 – 14:50	<p><u>Cancer in AT</u></p> <p>Chairs: Tatjana Stankovic (Institute of Cancer and Genomic Sciences, University of Birmingham, UK) and Sandeep Potluri (Birmingham Children's Hospital, UK)</p>
13:30	<p>Invited speaker – Richa Sharma (Cleveland Clinic, USA): Cancer prevalence, outcomes, and treatment-related toxicities in AT patients</p>
13:50	<p>Invited speaker – Sara Elitzur (St Jude Children's Research Hospital, USA): The clinical approach to patients with AT and hematological malignancies: treatment and surveillance</p>
14:10	<p>Renata Neeves (University of Nottingham, UK): Feasibility of whole-body MRI for cancer screening in children and young people with AT: a mixed methods cross-sectional study</p>
14:20	<p>General discussion</p>
14:50 – 15.20	<p>Coffee/tea break</p>
15:20 – 17.00	<p><u>Critical advances in AT scientific research (Part II)</u></p> <p>Chairs: David Coman (Queensland Children's Hospital, Australia) and Bruno Salomone Gonzales (University of Bristol, UK)</p>
15:20	<p>Invited speaker (online) – Tim Yu (Harvard Medical School, USA), Claudio DeGusmao (Harvard Medical School, USA) and Valerie Natale (Forgotten Diseases Research Foundation USA): An investigational, mutation-specific ASO therapy for AT</p>

Thursday 26.06.2025 | 08:30 – 17:00

15:50	Invited speaker – Matthis Synofzik (University of Tübingen, Germany): Bringing ASO therapies for AT patients into clinics at scale: a bench-to-bedside platform for personalised ASO development
16:10	James Dixon (University of Nottingham, UK): Brain-penetrating ATM gene therapy for transient or stable phenotype-rescue of AT patient cells
16:20	Masatoshi Takagi (Institute of Science, Japan): Development of Adenovirus ATM expression vector capable of genome integration
16:30	General discussion
17:00 – 18.00	<u>Networking and poster session 2</u>
18:00 – 18:30	Working group meeting: Psychological support (Room 8)
18:30 – 19:00	Working group meetings: Cancer management (Room 17), Related disorders e.g.: AOA1 (Room 8)
	<p>For more information about the networking groups, please scan the QR code</p> 
20:00	Dinner



9:00 – 10:00	<u>Core outcomes in AT trials - Consensus meeting</u> William Whitehouse (University of Nottingham, UK)
10:00 – 11:30	<u>Clinical Neurology.</u> Chairs: Barbara Pietrucha (Children's Memorial Health Institute, Poland) and May Yung Tiet (Cambridge Medical School, UK)
10:00	Invited speaker (online) – Tom Crawford (John Hopkins Hospital, USA): Development and value of clinical measures of neurologic function
10:20	Franziska Hoche (Harvard Medical School, USA): The neurovascular biology of AT
10:40	Emily Petley (University of Nottingham, UK): The natural history of AT N-HAT in the UK population
10:50	Trudy Burgers and Asbjorg Stray-Pedersen (Oslo University Hospital, Norway): Development and utility of a combined neuro-motor test battery in AT
11:00	General session discussion
11:30 – 12:00	Coffee/tea break
12:00 – 13:30	<u>Priorities to progress clinical trials</u> Chairs: Howard Lederman (John Hopkin's University School of Medicine, USA) and Nienke van Os (Radboud University Medical Centre, The Netherlands)
12:00	Invited speaker – Divya Kulkarni (Massachusetts General Hospital/Harvard Medical School, USA): Disease progression in AT: At-home wearables study
12:20	Invited speaker – Maureen Lefton-Greif (John Hopkin's University School of Medicine, USA): Towards clinical trial readiness: the AT functional scale of what matters most to patients
12:40	Rosella Abeti (University College London, UK): Exploring omaveloxolone efficacy in AT
12:50	Rita Horvath (University of Cambridge, UK): Increasing Trial Readiness in AT (TREAT-AT) - an approach to developing ASO treatment for adults carrying the 'UK mutation'
13:00	General session discussion
13:30 – 14:30	Lunch
14:30 – 15:15	<u>Mental health and cognitive function</u> Chair: Jennifer Thornton (A-T Children's Project, USA)

14:30	Speakers: Anne Murray (AT Society, UK) and Jane Flint (AT Society/ Nottingham University Hospital): Beyond the AT diagnosis: supporting mental well-being, with insights from the AT Society
15:05	Sergio Rinella (University of Catania, Italy): Lighting on the Cerebellar Cognitive-Affective Syndrome (CCAS) in children with AT
15:15 – 16:45	<u>Treatment of AT including planned and ongoing clinical trials</u> Chairs: Michel Willemsen (Radboud University Medical Centre, NL) and Nivedita Thakur (McGovern Medical School at UTHealth, USA)
15:15	Invited speaker – David Coman (Queensland Children’s Hospital, Australia): Triheptanoin trial
15:30	Invited speaker – Maureen Roden (Quince Therapeutics): Update on eDSP (encapsulated Dexamethasone Sodium Phosphate)
15:45	Invited speaker – Hilde Nilsen (University of Oslo, Norway): Nicotinamide Riboside trial
16:00	Invited speaker – Taylor Fields (Intrabio) and Fran Platt (University of Oxford, UK): Pivotal Trial of N-acetyl-L-leucine (IB1001) for AT
16:15	General session discussion
16:45 – 17:15	Final remarks and end of clinical meeting
17:30 – 18:45	This session is aimed at AT families; delegates of the clinical meeting are invited to attend <u>General management of AT</u> (Room 15) Chairs: Clare Bolton (Royal Papworth Hospital, UK) and Anna Wilsden (Nottingham University Hospital, UK)
17:30	Kim Chow (The Chinese University of Hong Kong) and Sarah Durnan (Nottingham University Hospitals NHS Trust & University of Nottingham, UK): Management of diet, energy levels and metabolism in AT
18:05	Lisa Bunn (University of Plymouth, UK): Making physiotherapy work for you
18:30	Yossi Shiloh (Tel Aviv University School of Medicine, Israel): What do we know about the health effects of being a carrier of an AT mutation?
19:15	Dinner
<div> <div>SCAN ME</div>  </div> <p>Further clinical research discussions will take place on Saturday 28th June 8:30 - 1:00pm at the AT Society Family Weekend. This session is for AT families but conference delegates are welcome to attend. Scan QR code for more details.</p> <p>https://atsociety.org.uk/family-weekend-2025/</p>	