

## PRELIMINARY PROGRAMME

### November 29

#### 14.00 Welcome

Living with A-T: Introduction from an Italian Family

Chair: Richard Gatti, US

#### 14.20 Keynote Lecture I

*Pleiotropic roles of ATM in cell biology and future research directions*

Yossi Shiloh, Israel

### 1-SESSION: CANCER

Chair: Jan Loeffen, The Netherlands

#### 15.05 Clinical

*Retro- and prospective tumour survey in A-T*

Nicolas Davies, UK

#### Basic Science: Chromothripsis

#### 15.20 *Chromothripsis in DNA repair disorders*

Zeynep Tumer, Denmark

#### 15.35 *ALL in A-T: Link between ATM mutations and chromothripsis*

Aur lie Ernst, Canada

#### 15.50 Group Discussion of Clinic Issues

- protocols for management of common cancers
- guidelines for tumor surveillance (especially for solid tumors)
- biorepository for sample storage consistent saving of biopsy tissue
- the molecular basis of cancer in A-T

#### 16.20 Break

### 2-SESSION: RESPIRATORY ROUND TABLES

Chair: Jayesh Bhatt, UK

#### *Round Table I: Respiratory physiotherapy and other treatment modalities*

#### 16.40 When to initiate airway clearance in someone with minimal symptoms and best modality

Nicky McNarry, UK

#### 16.50 The role of hypertonic saline in treatment

Deborah Snijders, Italy

#### 17.00 When and in whom to commence prophylactic antibiotics

Stefan Zielen, Germany

#### 17.10 Discussion

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**Round Table II: Functional evaluation of the lung**

**17.20 Scores for lung evaluation in A-T**

Ifat Sarouk, Israel

**17.30 Hallmarks of lung disease in A-T versus other PIDs**

Francesca Santamaria, Italy

**17.40 Link between infections and lung disease in A-T**

Abrey Yeo, Australia

**17.50 Discussion**

**Round Table III: Magnetic Resonance Imaging (MRI) of the lung**

**18.00 Update from the UK**

Andrew Prayle, UK

**18.10 Ultra Short Echo Time MRI for automatic analysis of pediatric A-T pulmonary progression and treatment response**

Anthony Paproki, Australia

**18.20 Discussion**

**18:30 Break for evening activities**

**November 30**

Chair: Ridha Barbouche, Tunisia

**8.30 Keynote Lecture II**

***The porcine model for A-T***

David Pearce, US

**3-SESSION: BASIC NEUROSCIENCE**

Chair: Yossi Shiloh, Israel

**9.00 Overview of the neurological features of AT**

Tom Crawford, US

**9.15 What is known about the cerebellum and ataxia?**

Henry Paulson, USA

**9.30 Physiologically-induced DNA double-strand break repair is defective in Purkinje cells in a mouse model of ataxia-telangiectasia**

Martin Lavin, Australia

**9.45 What can be learned from DNA repair deficiencies in the mouse brain?**

Peter McKinnon, US

**10.00 Induced Pluripotent Stem cells and organoids: experimental models to approach neurodegeneration**

Domenico Delia, Italy

**10.15 Group Discussion**

- What does this mean for the clinic?

**10.45 Break**



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**4-SESSION: CLINICAL NEUROLOGY**

Chair: Andrea Nissenkorn, Israel

**11.15 CATNAP imaging study: an update**  
Robert Dineen, UK

**11.30 Functional Scale for A-T: an update**  
Maureen Lefton-Greif, US

**11.45 Interactive virtual reality system to maximize motor learning profiles**  
Andrea Turolla, Italy

**12.00 Wearable body sensors to quantify neurodegeneration**  
Richard Festenstein, UK

**12.15 Computer learning for A-T**  
Anoopum Gupta, US

**12.30 Group Discussion**

**13.00 Lunch and Poster viewing**

**5-SESSION: THERAPEUTIC CHALLENGES FOR OLDER PATIENTS**

Chair: Malcolm Taylor, UK

**CLINICAL OVERVIEWS**

**14.00 Endocrine and lipid alterations in A-T**  
Stefan Zielen, Germany

**14.10 Skin and reproductive abnormalities in A-T**  
Howard Lederman, US

**14.20 Liver disease in A-T**  
Batya Weiss, Israel

**14.30 Heart disease in A-T**  
Francesco Violi, Italy

**14.40 RAMP (Response of Individuals with A-T to Metformin & Pioglitazone):**  
Laura McCreight, UK

**BASIC SCIENCE: METABOLISM**

**14.50 The role of ATM in cellular metabolism: an update**  
Karl Herrup, HongKong

**15.00 ATM and cell metabolomics**  
Vincenzo Costanzo, Italy

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**15.10 Group Discussion**

- *Diabetes*
- *Safety of statins in lowering lipids*
- *Use of estrogen for osteoporosis*

**6-SHORT SESSION: IMMUNOLOGY**

Chair: Howard Lederman, USA

**15.30 Inflammation in A-T**

Sharon McGrath-Morrow, US

**15.45 Understanding the disease from the variability of the immune defects**

Claudio Pignata, Italy

**16.00 Preemptive applied cell therapy for A-T**

Ralf Schubert, Germany

**16.15 Group Discussion**

**16:35 Coffee break**

**7-SESSION: CLINICAL TRIALS FOR A-T**

Chair: Terry Sanger, US

**16.50 ATTeST (EryDex System): an update**

Luca Benatti, EryDel Representative, Italy

**17.00 Pros and cons of intra-erythrocyte dexamethasone administration: a long term follow-up**

Roberto Micheli, Italy

**17.10 Oral betamethasone formulation: an update**

George Kottayil, Grace Therapeutics Representative, US

**17.20 The potential of deep brain stimulation for A-T**

Terry Sanger, US

**17.35 Non-viral vector gene therapy approach for A-T**

James Dixon, UK

**17.50 Anti-sense oligonucleotides therapy to target splice site mutations in the ATM gene**

Oded Regev, US

**18.00 Group Discussion**

**8-SHORT SESSION: COGNITION AND PSYCHOLOGICAL / SOCIAL ISSUES IN A-T**

Chair: William Davis, UK – Ennio Del Giudice, Italy

**18.15 Cognition and A-T: an update**

Franziska Hoche, US

**18.30 Key psychosocial and relational issues**

Ennio del Giudice, Italy



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**18.45 Live well with A-T: addressing the psychological and emotional impact of A-T on quality of life**

Anne Murray, UK

**19.00 Group Discussion**

**19:20 Break for evening activities**

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**9- SHORT SESSION: FACTORS AFFECTING DISEASE SEVERITY**

Chair: Martin Levin, Australia

**9.00 Gene-gene interactions involving ATM in shaping the phenotype**

Patrick Concannon, US

**9.15 How NGS can contribute to identify genetic factors that modify disease severity**

Vincenzo Nigro, Italy

**9.30 Identifying potential mechanisms to suppress A-T pathologies**

Rimma Belotserkovskaya, UK

**9.45 Group Discussion**

- Phenotypic variability within families

**10.05 Coffee break**

**10-SESSION: UNUSUAL OR DIFFICULT CASES AND DISCUSSION**

Chair: Luciana Chessa, Italy

**10.25 Six unusual or complex cases discussion**

**11-SESSION: RESEARCH PROJECTS FROM YOUNG INVESTIGATORS: box lunch and presentations**

Chair: Domenico Delia, Italy

**11:30 Seven 8 min proposals**

**13.00 CLOSING REMARKS**

Cynthia Rothblum-Oviatt, US

William Davis, UK

Claudio Pignata, Italy

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**Information**

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**Venue**

Congress Center of the University of Naples Federico II  
(Via Partenope, 36 - Naples, Italy)

*Congress Center of the University Federico II, an historical building located in the center of Naples on the seafront promenade, just in front of the stunning Castel dell'Ovo, Borgo Marinari and with a view of Capri island.*

**Abstract submission**

We invite to submit abstracts on the symposium topics for oral and poster presentations. Abstracts must be submitted online through the conference website ([www.centercongressi.com/ATconference2018](http://www.centercongressi.com/ATconference2018)).

**AT Conference Topics:**

- ✓ CANCER
- ✓ BASIC NEUROSCIENCE
- ✓ CLINICAL NEUROLOGY
- ✓ THERAPEUTIC CHALLENGES FOR OLDER PATIENTS
- ✓ IMMUNOLOGY
- ✓ CLINICAL TRIALS FOR A-T
- ✓ COGNITION AND PSYCHOLOGICAL/SOCIAL ISSUES IN A-T
- ✓ FACTORS AFFECTING DISEASE SEVERITY
- ✓ UNUSUAL OR DIFFICULT CASES AND DISCUSSION
- ✓ RESEARCH PROJECT FROM YOUNG INVESTIGATORS

**Registration and accommodation booking**

Registration and reservations (including social events and hotel) should be made online at [www.centercongressi.com/ATconference2018](http://www.centercongressi.com/ATconference2018).

Please consider that November may be very busy with tourists in Naples, so early booking is advisable.

**Important dates**

Abstract submission opens: May 31, 2018

Deadline for abstract submission: September 17, 2018

Notification of abstract acceptance: September 25, 2018

Deadline for early bird registration: September 30, 2018

*We look forward to seeing you at*

***Ataxia Telangiectasia Clinical Research Conference!***

Organizing Secretariat



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