

# **A-TCRN** | **A-T Clinical Research Network**

## **PROGRAMME**

### **ATAXIA-TELANGIECTASIA CLINICAL REASERCH CONFERENCE**

**WARSAW, POLAND**

**6-8 October, 2016**

Thursday 6 October

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**12:00-13:00 Lunch**

**13:00 WELCOME**

Barbara Pietrucha and William Davis

**13:15 Introductory From Family Living with A-T**

Jerzy Nowak, the Polish Razem Zdążymy Foundation for A-T Patients

**13:30 SESSION 1. NEUROLOGY**

**Chair:** Andrea Nissenkorn, Israel

**CLINICAL**

**13:35 Natural History of A-T Study**

William Whitehouse/Salma Ali, UK

**13:50 Static/Progressive Impairment and Dysfunction in A-T**

Tom Crawford, USA

**14:05 Initial results from the Childhood A-T Neuroimaging Assessment Project (CATNAP)**

Robert Dineen, UK

**BASIC SCIENCE: RESEARCH TOWARD THE CLINIC**

**14:20 Impact of ATM Deficiency on Neural Stem Cells**

Penny Jeggo, UK

**14:30 DNA Damage Responses and Transcriptional Regulation in iPSC-Derived Neurons from A-T Patients**

Domenico Delia, Italy

**14:40 The Role of ATM in Brain Homeostasis and Functionality**

Ari Barzilai, Israel

**14:50 PANEL DISCUSSION**

Chair as Moderator

**15:30-15:50 Break**

**15:50 SESSION 2. IMMUNOLOGY**

**Chair:** Asbjorg Stray-Pedersen, Norway

**CLINICAL**

**15:55 Ataxia Telangiectasia: Immunodeficiency and Survival**  
Corry Weemaes, The Netherlands

**16:10 Antibody Deficiency in Ataxia-Telangiectasia**  
Isabella Quinti, Italy

**16:25 The Differences of T and B Cell Maturation in A-T Patients On/Without Immunoglobulin Supplementation**  
Barbara Piatosa/Barbara Pietrucha, Poland

**16:40 Inflammatory Cytokines in A-T**  
Howard Lederman, USA

**16:55 DNA Double-Strand Break Repair – Common Mechanism with Multiple Proteins Involved: Similarities and Differences in Clinical and Immunological Phenotype of Patients with Mutations in *Artemis*, *ATM*, *NBN*, and *BLM***  
Barbara Piątosza, Poland

**17:10 PANEL DISCUSSION**  
Chair as Moderator

**17:50-18:35 KEYNOTE PRESENTATION**  
**Keynote Speaker: Richard Gatti, US**  
**Developing SMRT Drugs for Treating Nonsense Mutations: A New Model, Single Drugs for Groups of Diseases**

**19:30 Dinner**

Friday 7 October

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**8:30 SESSION 3. NEXT-GEN A-T RESEARCH**

Chair: Brad Margus, USA

**8:35 Ataxia Telangiectasia and Genomic Science**

Malcolm Taylor, UK

**8:50 New Born Screening with TRECS**

Asbjorg Stray-Pedersen, Norway

**9:05 Applying Big Data Approaches to A-T**

Brad Margus, USA

**9:20 PANEL DISCUSSION**

Chair as Moderator

**10:00-10:20 Break**

**10:20 SESSION 4. METABOLIC AND MULTI-SYSTEMIC PROBLEMS**

Chair: Howard Lederman, USA

**CLINICAL**

**10:25 A-T in Older Patients**

Howard Lederman, USA

**10:40 Premature Aging in Patients with Ataxia Telangiectasia:  
Is It Possible to Find New Surrogate Markers of Disease Progression?**

Stefan Zielen, Germany

**10:55 Metabolic syndrome and liver involvement in ataxia-telangiectasia**

Andrea Nissenkorn, Israel

**11:10 PANEL DISCUSSION**

Chair as Moderator

**12:00-13:30 Lunch**

**13:30 SESSION 5. RESPIRATORY**

Chair: Jayesh Bhatt, UK

**CLINICAL**

**13:35 Multidisciplinary Respiratory Management of A-T: An Update**

Jayesh Bhatt, UK

**13:50 Swallowing Incoordination and Management**

Maureen Lefton-Greif, US

**14:05 Lung Disease Assessment in Ataxia-Telangiectasia with Magnetic Resonance Imaging**

Goffredo Serra, Italy

**14:20 IMAGIN-AT – Advanced Imaging and Physiology Respiratory Endpoints for Clinical Trials in Ataxia-Telangiectasia**

Andrew Prayle, UK

**14:35 Role of Oxidative Stress in Pulmonary Disease in Ataxia-Telangiectasia**

Martin Lavin, AU

**BASIC SCIENCE: RESEARCH TOWARD THE CLINIC**

**14:50 Pulmonary Inflammation and Fibrosis in the Atm-Deficient Mouse Model**

Ralf Schubert, Germany

**15:00 PANEL DISCUSSION**

Chair as Moderator

**15:45-16:20 Break**

**16:20 SESSION 6. EMERGING TREATMENTS**

Chair: Richard Gatti, USA

**CLINICAL**

**16:25-17:10 ERYDEX STUDY UPDATE**

**Speaker:** Antonio Ferrari, Italy

**A Multi-center, Randomized, Double-blind, Placebo controlled Trial to Evaluate the Effects of Intra-Erythrocyte Dexamethasone Sodium Phosphate on Neurological Symptoms in Patients with Ataxia Telangiectasia (ATTeST)**

**17:10 RBCs as Circulating Carriers for the Release of Dexamethasone in the Treatment of AT**

Mauro Magnani

**17:25 New Aspects on the Steroid Treatment in Patients Affected with Ataxia-Teleangiectasia**

Claudio Pignata, Italy

**17:40 Treatment with Intra-Erythrocyte Dexamethasone Sodium Phosphate in AT-patients**

Roberto Micheli, Italy

**17:55 PANEL DISCUSSION**

Chair as Moderator

**18:35 Break**

**19:30 Dinner and Entertainment**

Saturday 8 October

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**8:30 SESSION 7. ONCOLOGY**

**Chair:** Krzysztof Kałwak, Poland

**CLINICAL**

**8:35-9:05 CLINICAL ONCOLOGY KEYNOTE**

**Keynote Speaker:** John (Torrey) Sandlund, USA

**Cancer Risk in AT Patients, Cancer Treatment Approaches, Guidelines and Future Directions**

**9:05 New Developments and Initiatives in Treatment of Hematological Malignancies in Patients with Ataxia Telangiectasia**

Jan Loeffen, The Netherlands

**9:20 Three Cases of Allogenic Stem Cell Transplantation in A-T – Single Centre Experience**

Marek Ussowicz, Poland

**9:35 Allogeneic Haematopoietic Stem Cell Transplantation in Patients with DNA Repair Defects Other Than Ataxia-Teleangiectasia Syndrome**

Krzysztof Kałwak, Poland

**9:50 ATM Heterozygote Risks in Cancer**

Thilo Dörk, Germany

**10:05 ATM and The Risk for Contralateral Breast Cancer**

Pat Concannon, USA

**BASIC SCIENCE: RESEARCH TOWARD THE CLINIC**

**10:20 ATM Deficient Phenotype in Tumour Cells – Possibilities for Targeted Treatment**

Tanya Stankovic, UK

**10:30 PANEL DISCUSSION**

Chair as Moderator

**11:10-11:30 Break**

**11:30 SESSION 8. BASIC SCIENCE AND PRESENTATION FROM POSTERS (10 min each)**

**Chair:** Tej Pandita, USA

**11:35 Longevity in Classical and Variant Ataxia-Telangiectasia: A Genotype-Phenotype Association?**

Nienke J.H. van Os, The Netherlands

**11:45 Pluripotent Stem Cells and DNA Damage Response to Ionizing Radiation**

Tej Pandita, USA

**11:55 An Integrated ATR, ATM and mTOR-Mechanical Network Controlling Nuclear Plasticity**

Marco Foiani, Italy

**12:05 The Relationship Between Patient Perception and Swallow Function in Patients with Ataxia-Telangiectasia: A Pilot Study**

Corinne Mossey-Gaston, UK

**12:15 Variant Ataxia Telangiectasia: Clinical and Genetic Features in 37 Adult Patients Attending the UK National Service.**

Katherine Schon, UK

**12:25 Antioxidant/Oxidant Status in Patients with Ataxia-Telangiectasia**

Mateusz Maciejczyk, Poland

**12:35 Risk of Atherosclerosis and Diabetes in Patients with Ataxia Telangiectasia.**

Beatriz T Costa-Carvalho, Brasil

**12:45 The Effect of Home Respiratory Physiotherapy in A-T Patients**

Ifat Sarouk, Israel

**12:55-13:45 Lunch**

**13:45 SESSION 9. A DISCUSSION OF UNUSUAL CASES**

Chair: Luciana Chessa, Italy

**13:50 Classification of A-T Patients by Genotype and Phenotype: How Many Unusual Cases?**

Luciana Chessa, Italy

**14:05 PRESENTATION OF CASES AND DISCUSSION (10 min for each case)**

Chair as Moderator

**14:15 Cutaneous granulomas in patients with Nijmegen Breakage Syndrome (NBS) and Ataxia-telangiectasia (AT) – diagnostic and treatment challenge**

Edyta Heropolitańska-Pliszka, Poland

**14:25 Coexistence of A-T and Allgrove syndrome in 6- year- old boy.**

Małgorzata Skomska-Pawliszak, Poland

**TBD**

**16:00 SESSION 10. THE WAY FORWARD**

Cynthia Rothblum-Oviatt, USA

Barbara Pietrucha, Poland

William Davis, UK

**16:30 Adjourn**