

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

PROGRAMME

ATAXIA-TELANGIECTASIA CLINICAL REASERCH CONFERENCE

WARSAW, POLAND

6-8 October, 2016

12:00-13:00 Lunch

13:00 WELCOME

Barbara Pietrucha and William Davis

13.10 Welcome from Scientific Director of Children's Memorial Health Institute

Katarzyna Kotulska-Jóźwiak

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 Barzilaj, 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 Piątosza/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

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-Telangiectasia

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

8:30 SESSION 7. ONCOLOGY

Chair: Krzysztof Kalwak, 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 Kalwak, 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

12:45-13:45 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

14:35 Immunotherapy responsive neurological decline and spontaneous intracranial hypotension in a patient with classical Ataxia Telangectasia

Anke E Hensiek

14:45 Facial disfiguring cutaneous granuloma resistant to all available therapies

Kate Sinclair, AU

TBD

16:00 SESSION 10. THE WAY FORWARD

Cynthia Rothblum-Oviatt, USA

Barbara Pietrucha, Poland

William Davis, UK

16:30 Adjourn