

**8th ALSTRÖM SYNDROME INTERNATIONAL FAMILY CONFERENCE
AND
SCIENTIFIC SYMPOSIUM
MAY 12 – 16, 2016
PLYMOUTH, MASSACHUSETTS USA**

WEDNESDAY, MAY 11 PRE-CONFERENCE ACTIVITIES

- 09:00 – 11:00 ASI DEPARTMENT HEAD MEETING [**Halifax**]
13:30 – 15:00 PRE-CON MEETING WITH HOTEL 1620 MANAGERS [**Halifax**]
18:00 – 20:00 DINNER ON YOUR OWN [**On-site or Nearby Possibilities**]
20:00 – 23:00 A-TEAM, PARENTS AND PROFESSIONALS NETWORKING AND RELAXING [**Mayflower, Pub, Carver, and Plympton**]

THURSDAY, MAY 12 PRE-CONFERENCE ACTIVITIES/REGISTRATION/ORIENTATION

The following spaces will be available pretty much fulltime throughout the conference!

- ~ **Room 402** – Conference small meeting room for those in need.
- ~ **Amphitheater** – Family Friendly Movies **in the evening**
- ~ **Board Room** – Let’s Talk Tech! Vendors, displays, hands-on opportunities, and more.
- ~ **Room 302** – Tot Tending (ages 1-3)
- ~ **Carver** – Day Care and Activity Center for Younger Children (ages 4-6)
- ~ **Plympton** – A-Team Lounge and Informal Discussion
- ~ **Pool** (SUPERVISION required!!)
- ~ **Manomet** - Memory and Reflection Room – A Quiet Space
- ~ **The Carl Henry (Mayflower)** – A place to gather in the evenings and relax!

- 06:30 – 09:30 BREAKFAST [**Hotel 1620 Harbor Grille Restaurant or Nearby Area Restaurants**]
- 07:30 – 08:30 ASI DEPARTMENT HEAD MEETING [**402**]
- 09:00 – 22:00 CONFERENCE REGISTRATION [**Main Lobby Area**]
- 15:15 – 16:15 **Snapshot of What to Expect** (for new families) [**Halifax**] **Robin Marshall, Shelly Marshall**
- 16:00 – 17:00 Alström Angels Board Meeting [402]
- 17:30 – 19:00 Welcome Reception [**Mayflower/Carl Henry**]
- 19:00 – 21:00 DINNER-ON-OWN [**HARBOR GRILLE OR AREA RESTAURANTS**]
[**SCIENTISTS/ASI BOARD, SSC ON PILGRIM BELLE**]
- 20:00 – 23:00 A-TEAM, PARENTS AND PROFESSIONALS NETWORKING AND RELAXING [**Mayflower/Carl Henry**]

08:30-18:00

SCIENTIFIC SESSIONS
SYMPOSIUM FOR PHYSICIANS AND SCIENTISTS [Amphitheater]
LIVE FEED AVAILABLE TO ALL NEXT DOOR [Board Room]

Generously sponsored by Alstrom Syndrome Canada

- 08:30 - 08:45 WELCOME, CONFERENCE OVERVIEW, AND ANNOUNCEMENTS *Jan Marshall, ASI Board and Chair, Scientific Advisory Board, The Jackson Laboratory, Bar Harbor, ME;*
- 08:45 - 09:30 HISTORICAL PERSPECTIVE: UNRAVELLING THE MOLECULAR BASIS OF ALSTROM SYNDROME *Gayle B Collin, MS, The Jackson Laboratory, Bar Harbor, ME*
- 09:30 - 10:15 THE NIH CILIOPATHY STUDY *Meral Gunay-Aygun, MD, National Human Genome Research Institute, NIH, Bethesda, MD*
- 10:15 - 10:30 COFFEE BREAK
- 10:30 - 11:15 FIBROSIS IN ALSTROM SYNDROME: CLINICAL AND EXPERIMENTAL STUDIES. *Vera Bettini, MD, University of Padua, San Bartolomeo Hospital, La Spezia, Italy*
- 11:15 – 12:00 PBI-4050, PRECLINICAL AND CLINICAL RESULTS – *John Moran, MD, Chief Medical Officer ProMetic Life Sciences Inc., Laval, Quebec, Canada*
- 12:00 – 13:00 **LUNCH and ANNOUNCEMENTS [Harbor Grille]**
- 13:00 – 13:45 ADVANCED LIPID PHENOTYPES IN ALSTROM SYNDROME, *Robert Shamburek, MD, National Heart, Lung and Blood Institute, NIH, Bethesda, MD*
- 13:45 – 14:30 ADIPOCYTE DYSFUNCTION AND ITS CONSEQUENCE IN ALSTRÖM SYNDROME *Tarekegn Hiwot, MD, Queen Elizabeth Hospital, University of Birmingham, Birmingham, UK*
- 14:30 – 15:15 ADIPOSE TISSUE-RELATED ENDOCRINE ASPECTS AND THEIR SYSTEMIC IMPACTS *Vincent Marion, PhD, INSERM, Strasbourg, France*
- 15:15 – 15:30 COFFEE BREAK
- 15:30 – 16:15 SERIAL CHANGE IN CARDIOVASCULAR FUNCTION IN ALMS *Richard Steeds, MD, Queen Elizabeth Hospital, University of Birmingham, Birmingham UK*
- 16:15 – 17:00 VISUALIZING ALSTROM SYNDROME *Alexander Berger, The Jackson Laboratory, Bar Harbor, ME*
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- 18:00 – 20:00 COCKTAILS/DINNER on the Pilgrim Belle (ASI Board, Scientists Stakeholders Steering Committee)

FRIDAY, MAY 13 DAY 1

As noted above, supervised recreational space has been provided for the children. In addition, there will be carefully chaperoned and age- appropriate activities on Friday, Saturday, and Sunday, including off-campus trips to area attractions. The following spaces will be available pretty much fulltime throughout the conference!

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- 06:30 – 08:00 BREAKFAST BUFFET [**Mayflower**]
- 06:30 – 10:00 REGISTRATION FOR LATE COMERS [**Lobby Area**]
- 08:00 – 08:30** CHILDREN DEPART TO ACTIVITIES [**Back Door Reception Area**]
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- 08:30 – 08:45 WELCOME AND INTRODUCTIONS [**Plymouth/Kingston**] *Robin Marshall, Executive Director; Phyllis Aschenbrenner, ASI Chair Board of Directors*
- 08:45 – 9:00 TWENTY YEARS!! *Jan Marshall, ASI Board and Chair, Scientific Advisory Board, The Jackson Laboratory, Bar Harbor, ME*
- 09:00 - 9:30 INTRODUCTION TO GENETICS FOR ALSTROM SYNDROME: *Clair Francomano, MD, Director Adult Genetics,, Harvey Institute of Human Genetics, Greater Baltimore Medical Center, Baltimore, MD; Antonie Kline, MD, Director Pediatric Genetics, Harvey Institute of Human Genetics, Greater Baltimore Medical Center, Baltimore, MD*
- 09:30 – 10:00 VISION LOSS in ALSTROM SYNDROME: OPTIONS AND HOPES: *Alex V. Levin, MD, Wills Eye Hospital, Philadelphia, PA*
- 10:00 – 10:15** **COFFEE BREAK**
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- 10:15 – 11:00 CARDIAC FUNCTION PANEL DISCUSSION
- CARDIAC PHENOTYPES IN ALSTRÖM SYNDROME - OVERVIEW: *Cathy Carey, MD, Torbay Hospital, Torquay UK*
 - CARDIAC FUNCTION IN ALSTRÖM SYNDROME: *Richard Steeds, MD, Queen Elizabeth Hospital, Birmingham, UK*
 - PEDIATRIC CARDIOMYOPATHY IN ALSTROM SYNDROME: *Andrew Warren, MD, IWK Medical Center, Halifax, NS Canada*
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- 11:00 – 11:30 FAMILY STUDIES ON HYPERLIPIDEMIA IN ALSTRÖM SYNDROME: *Robert Shamburek, MD, National Heart, Lung and Blood Institute, NIH, Bethesda, MD*
- 11:30 – 12:00 NIH STUDY – CLINICAL AND MOLECULAR INVESTIGATIONS INTO CILIOPATHIES: LIVER DISEASE IN ALSTROM SYNDROME: *Meral Gunay-Aygun, MD, National Human Genome Research Institute, NIH, Bethesda, MD*
- 12:00- 13:00 **LUNCH and ANNOUNCEMENTS [Mayflower] A Team Special Lunch [Pilgrim Belle]**
- 13:00 – 13:30 PULMONARY MANIFESTATIONS IN 38 PATIENTS WITH ALSTRÖM SYNDROME, *Caroline Boerwinkle, Baylor College of Medicine, Houston, TX*
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- 13:30 – 14:00 ASPECTS OF LEARNING IN DEAFBLINDNESS - OPPORTUNITIES AND LIMITATIONS FOR PERSONS WITH ALSTRÖM SYNDROME: *Berit Rönnåsen, PhD, Örebro University Hospital, The Swedish Institute for Disability Research, Örebro, Sweden*
- 14:00 – 14:30 DEAFBLINDNESS, THEORY of MIND, COGNITIVE FUNCTIONING and SOCIAL NETWORKS in ALSTRÖM SYNDROME: *Hans-Erik Frölander, PhD, The National Agency for Special Needs Education and Schools, Örebro, Sweden*
- 14:30 – 14:45 COFFEE BREAK
- 14:45 – 15:15 ARC: PUT YOURSELF IN THE RESEARCH DRIVING SEAT : *Matthew Smith, BS, Health Information Technology Manager, Genetic Alliance; Principal Investigator, Joubert Syndrome Registry, Joubert Syndrome & Related Disorders Foundation (JSRDF)*
- 15:15 – 15:45 TECHNOLOGY FOR LIFE: *Marco Gregnanin, CEO, ITEX, Rome, Italy*
- 15:45 – 16:30 TRANSPLANT BASICS: WHAT YOU NEED TO KNOW ABOUT KIDNEY TRANSPLANTATION: *Michelle Josephson, MD, Medical Director Kidney Transplant, University of Chicago, Chicago, IL; Patient Perspectives: Adam Kozarewicz (A-Team) Chicago, IL; David Faircourt (A-Team), Oceano, CA*
- 16:30 – 17:00 ENGAGEMENT PROCESS OVERVIEW – What lies in store for Saturday *Anne Nordstrom, PhD*
- 18:00 – 20:00 DINNER **[Ballroom]** *LIFE in the TRENCHES*: Some personal journeys....
- 20:00 – 23:00 HOT TOPICS *Topics of interest for informal discussions*
- [Boardroom]** LET'S TALK TECH **Conversation, displays, hands-on opportunities.**
Marco Gregnanin - ITEX, Rome, Italy; Ellen Hall, Joann Becker, and Martha Simon - Perkins School for the Blind, Watertown, MA; Jerry Berrier and Joann Becker - iCan Connect/Perkins School for the Blind; Catherine Bly, Easter Seals Massachusetts; Enhanced Vision; Regina Presley, Cochlear Americas.
- [Plympton]** A TEAM LOUNGE: TRANSITIONING TO ADULTHOOD – *Open discussion*
- [Lobby Business Center]** ARC REGISTRATION - *Matt Smith, Health Information Technology Manager, Genetic Alliance;*
- 20:00 – 23:00 RELAXING IN THE CARL-HENRY **[Mayflower]**
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SATURDAY, MAY 14 DAY 2

- 06:00 – 08:30 BREAKFAST BUFFET [Mayflower]
- 08:00 Children 6 – 17 depart for Activities
- 08:30 - 09:00 OPTIMIZING NUTRITION FOR OPTIMAL HEALTH:
Clair Francomano, MD, Director, Harvey Institute of Human Genetics, Greater Baltimore Medical Center, Baltimore, MD
- 09:00 – 09:45 WHEN HEARING AIDS ARE NOT ENOUGH: *Regina Presley, AuD, Senior Cochlear Implant Audiologist, Greater Baltimore Medical Center, Baltimore, MD*
PATIENT PERSPECTIVES: *Lilka Sander, Bromhof, South Africa; Shannon Caldwell, London, KY*
- 09:45 – 10:15 SHORT COURSE OF ENDOCRINOLOGY & METABOLISM IN ALSTROM SYNDROME: WHAT YOU SHOULD KNOW AND WHAT YOU COULD DO: *Pietro Maffei, MD, PhD, University of Padua, Padua, Italy*
- 10:15 – 10:45 INTRODUCTION to the DAY: *Anne Nordstrom, PhD, Walpole, NH*
- 10:45 – 11:00 COFFEE BREAK and DISPERSE to Designated Topic Areas (Plymouth, Kingston, Duxbury, Reception Area, and Halifax)
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- 11:00 – 13:00 ROUND ONE: STAKEHOLDER ENGAGEMENT
Discuss and identify meaningful elements critical to effective research and progress
(Topics Chosen by Stakeholder Steering Committee (SSC) December, 2015)

Topics for May 2016 ASI Conference

RESEARCH TOPICS

Uncommon Symptoms in Alström Syndrome

Documenting the impact and prevalence of less common symptoms e.g. fatigue, nosebleeds, anemia, pulmonary, stomach pain, muscle cramps and other unexplained pain and understanding when it is important to get treatment for unusual symptoms that arise.

Heart

Understanding the progression, impact and potential therapies for heart issues in Alström Syndrome

Metabolic and Liver Functions in Alström Syndrome

Identifying options to manage diet and obesity in AS
Exploring successful strategies for Diabetes control in AS
Understanding the progression, impact and potential therapies for liver disease in AS
Exploring the causes of and effective methods for control of high triglycerides and cholesterol in AS

Kidney

Understanding the progression, impact and potential therapies for kidney issues in AS

Vision in Alström Syndrome

Understanding the progression, impact and potential therapies for vision issues in Alström Syndrome

Hearing in Alström Syndrome

Understanding the progression, impact and potential therapies for hearing issues in Alström Syndrome

SUPPORT TOPICS

Living with Alström Syndrome

Managing the challenges and experiences of adult independent living with AS
Coping with depression, anxiety and phobias in AS
Coping with the economic impact of living with AS

Caring for Alström Syndrome

Successfully parenting children with Alström Syndrome
Managing the emotional and social impact for caregivers
Coping with depression, anxiety and phobias in AS
Coping with the economic impact of living with AS
Dealing with behavioral issues along the autism spectrum

EDUCATION TOPIC

Learning with Alström Syndrome

Discovering which technology and equipment is appropriate for each age level and how early to begin learning adaptive mobility and life skills
Helping teachers understand the challenges faced by students with AS
Choosing the best education option and optimizing schooling for the dual sensory impaired child

13:00 – 14:00 **LUNCH [Mayflower]**

14:00 – 16:00 **ROUND TWO: STAKEHOLDER ENGAGEMENT**
Sort and prioritize meaningful elements critical to research and progress

RESEARCH TOPICS

Uncommon Symptoms in Alström Syndrome
Heart dysfunction in Alström Syndrome
Metabolic, Lipid, and Liver Functions in Alström Syndrome
Kidney function in Alström Syndrome
Vision in Alström Syndrome
Hearing in Alström Syndrome

SUPPORT TOPICS

Living with Alström Syndrome
Caring for Individuals with Alström Syndrome

EDUCATION TOPIC

Learning with Alström Syndrome

16:00 – 16:30 **SUMMARY of the DAY and INSTRUCTIONS for VOTING on RESEARCH PRIORITIES [Plymouth/Kingston]: Anne Nordstrom, PhD**

16:30 – 23:00 **VOTING on RESEARCH PRIORITIES BEGINS [Former Registration Area]**

FIRST LOOK AND OPEN BIDDING FOR SILENT AUCTION ITEMS! [Harbor Grille]
COCKTAILS AND CASH BAR [Reception Area]

ARC REGISTRATION – [Lobby Business Area]- Matt Smith , Health Information Technology Manager, Genetic Alliance

18:30 – 23:00 **DINNER [Ballroom] with DJ, Dancing, 20th Anniversary Award,**
Keynote Address ~ Shannon Caldwell, A-Team Leader

AUCTION BIDDING WARS [Harbor Grille]. Auction closes at 21:00.

SUNDAY, MAY 15 DAY 3

- 07:00 – 09:00 BREAKFAST BUFFET [**Mayflower**]
- 09:00 – 11:30 CHELSEY’S OLYMPICS [**Nelson Park**] For children ages 4 -12. *Shelly Marshall, President Alström Olympic Committee, Burleson, TX*
- 09:00 – 11:30 JUST for SIBLINGS – For brothers and sisters aged 13 and up. [**Amphitheater**] *Cassie and Lynn Johnston, Lubbock, TX*
- 09:00 – 10:30 **CONCURRENT SESSIONS**
[**Duxbury**] A-TEAM ONLY: THE QUEST FOR INDEPENDENCE, AND MORE: *Shannon Caldwell, Chair, London, KY*
[**Kingston**] EDUCATION PANEL DISCUSSION; *Gina Denbow, Chair, Campobello, New Brunswick*
[**Plymouth**] CURBSIDE CONSULTATIONS: AN OPPORTUNITY FOR ONE-ON-ONE DISCUSSIONS
Clinical Specialists
- 10:30 – 12:00 **CONCURRENT SESSIONS**
[**Kingston/Duxbury**] ASK THE A-TEAM: AN UNPARALLELED OPPORTUNITY FOR PARENTS, RESEARCHERS, AND DOCTORS TO SEEK ENLIGHTENMENT. Jamie Seeger, moderator.
[**Plymouth**] CURBSIDE CONSULTATIONS CONTINUE...
[**Board Room**] MEDICAL SELF-MANAGEMENT AND LET’S TALK TECH CONTINUES...
- 12:00 – 13:30 **LUNCH on own in friendly Plymouth**

ASI Board and Stakeholders Steering Committee Meeting [**Room 402**]
- 14:00 – 16:00 Whale Watch Boat Trip
- 16:30 – 19:30 Fabulous Fantastic Full-out Fun at Family-Friendly Edaville USA [**Edaville USA**]
- 20:00 – 23:00 Networking and Relaxing in the Carl Henry [**Mayflower**]

MONDAY, MAY 16 DAY 4

06:30 – 08:30 BUFFET BREAKFAST [**Mayflower**]

9:00 – 10:00 PRELIMINARY RESULTS from VOTING [**Mayflower**] *Anne Nordstrom, PhD*

10:00 – 11:00 **General Meeting for Families: Meeting Your Needs/Meeting ASI's** [**Mayflower**]

11:00 – 12:00 CHECK OUT and FAREWELLS



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