

NORTHWEST  
**REGIONAL**  
**MEDICAL FORUM**

October 15, 2016 • Seattle

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October 15, 2016 • Seattle, WA

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## WELCOME TO THE 2016 MOWAT-WILSON SYNDROME FOUNDATION NORTHWEST REGIONAL MEDICAL FORUM

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The Board of Directors of the Mowat-Wilson Syndrome Foundation, and its volunteers, welcome all of you to the MWS Northwest Medical Forum!

We would like to thank Dr. Margaret Adam for all of her help in organizing this event!

In this packet you will find helpful information for the meeting.

1. Conference Agenda
2. Contact List for the Forum
3. Speaker Bios
4. Child Care Information
5. Child Care Release – to be signed
6. Child “Individual Needs” Form
7. Photography Release Form - to be signed
8. Sponsor Appreciation Page

Sincerely,

The Mowat-Wilson Syndrome Foundation Board Members

## AGENDA

**Saturday, October 15, 2016**  
**Radisson Seattle Airport**  
**18118 International Boulevard, Seattle WA 89188**

### Friday, October 14, 2016

Time	Topic	Location
6:00 pm	<b>Welcome Party – Pizza Dinner</b> For families arriving on Friday...	San Juan Ballroom

### Saturday, October 15, 2016

Time	Topic	Location
9:00	<b>Welcome</b> <i>Deborah Curry, MWS Foundation Vice President</i>	San Juan Ballroom
9:15	<b>Mowat-Wilson Syndrome: Overview and Updates</b> <i>Associate Professor, Meredith Wilson</i>	San Juan Ballroom
10:15	<b>Break</b>	
10:30	<b>Development of pluripotent stem cell models for Mowat-Wilson Syndrome</b> <i>Dr. Jay Vivian, Ph.D.</i>	San Juan Ballroom
11:15	<b>Update on the Italian MWS Study</b> <i>William Dobyns, M.D.</i>	San Juan Ballroom
12:00	<b>Lunch</b> <i>Sponsored by: The Eggers Family Trust</i>	San Juan Ballroom
1:30	<b>Seizure Diagnosis and Management in MWS</b> <i>Edward J. Novotny, M.D.</i>	San Juan Ballroom
2:30	<b>Bowel Management in patients with Mowat-Wilson Syndrome</b> <i>Lusine Ambartsumyan, M.D.</i>	San Juan Ballroom
3:15	<b>Break</b>	
3:30	<b>Augmentative Alternative Communication (AAC)</b> <i>Marci Ravelli, M.S. CCC/SLP</i>	San Juan Ballroom
4:30	<b>Adjourn</b>	San Juan Ballroom

## EVENT LOCATIONS

**Welcome Party** – San Juan Ballroom

**Saturday Forum** – San Juan Ballroom

**Saturday MWS Child Care** –

Shaw Room – MWS Children 5 and under

Orcas Room – MWS Children 6 - 13

Lopez Room – MWS Children 14 up

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## CONTACTS

In case of an emergency during the conference, please feel free to reach out to any of the board members listed below.

**Susan Triunfo**, President, Tyrone, GA  
[susant@mowat-wilson.org](mailto:susant@mowat-wilson.org)  
Mobile: 678-576-4398

**Deborah Curry**, Vice President, Las Vegas, NV  
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**Karen Baer**, Secretary, Houston, TX  
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**David Curry**, Director, Las Vegas, NV  
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**Jessica Rakshys**, Director, Commack, NY  
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Mobile: 516-435-5139

**Sandie Flannery**, Director, Effort, PA  
[sandief@mowat-wilson.org](mailto:sandief@mowat-wilson.org)  
Mobile: 570-350-9652

## CHILD CARE INFORMATION

MWS child care will be provided in three rooms. Ages 5 and under will be in the Shaw Room, ages 6 - 13 in the Orcas Room and ages 14 up in the Lopez Room. The care givers will be medical staff from Seattle Children's Hospital, Preschool teachers and family members of MWS individuals. There will also be some older typical kids to help with activities. Activities will be similar to classroom activities.

Since our children's needs are so varied, there will not be any snacks or drinks provided in this room. There will be a break in the morning session and one in the afternoon. During this time, parents can go in and give their child a snack. Care givers will handle diaper changes/toileting if you wish, or you can be notified by phone if you prefer to do it yourself.

### IF YOUR CHILD HAS A FOOD ALLERGY, PLEASE LET US KNOW!

Each child will have a name tag which will include the name of the parents and a phone number in case of emergency. During the conference, please have your phone on vibrate so you will know if you are needed.

***Please be sure to complete the "Child Care Release" on the next page.***

Parents,

Please complete the following questionnaire regarding your child in order to better assist the classroom helpers that will be caring for your children.

Thank you,  
Childcare Helpers

	Circle One	Additional/Helpful Information
Allergies	YES NO	
Ambulatory	YES NO	
Seizures	YES NO	
Toilet Trained	YES NO	
Mode of Communication	N/A	
Likes/Dislikes	N/A	
Miscellaneous	N/A	

Additional Information:

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## BIOGRAPHIES

### **Dr. Margaret Adam**

Margaret L.P. Adam, MD, is an attending physician at Seattle Childrens Hospital and an associate professor in the Department of Pediatrics at the University of Washington, School of Medicine. Dr. Adam's clinical interests include the diagnosis and management of infants, children, and adolescents with a variety of genetic conditions.

Dr. Adam has a special interest in Mowat-Wilson syndrome, for which she has conducted clinical research in the past. She is currently involved as the primary geneticist in the Multidisciplinary Disorders of Sex Development (DSD) clinic at Seattle Children's Hospital and she recently joined the Multidisciplinary 22q11 Deletion syndrome clinic. She also provides genetic consultations for pregnant women through the Prenatal Diagnostic Center at the University of Washington. Her current clinical research projects include working as a dysmorphologist on the Autoimmune Diseases in Pregnancy Project with the Organization of Teratology Information Specialists (OTIS) and on the Collaboration to Establish the Prevalence of Fetal Alcohol Spectrum Disorders in association with the University of California, San Diego.

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### **Mowat-Wilson Syndrome: Overview and Updates** **Associate Professor Meredith Wilson**

Meredith Wilson is a consultant medical geneticist based in the Department of Clinical Genetics at Children's Hospital Westmead, one of the two major paediatric hospitals in Sydney, New South Wales, Australia. After medical school in Adelaide, Meredith initially trained as a pediatrician in Sydney, then did further training in clinical genetics in Sydney and at the Murdoch Institute in Melbourne. She has been a consultant clinical geneticist since 1991 at the Children's Hospital at Westmead and also at the Westmead adult hospital nearby. Meredith was Head of the Children's Hospital Department of Clinical Genetics from 1998-2008 and head of the Westmead Hospital Adult Genetics service until 2014, and was appointed as a Clinical Associate Professor in the University of Sydney in 2009. Meredith's work involves many areas of clinical genetics, and includes consultations with families and patients of all ages, in prenatal, neonatal, paediatric and adult genetics areas. She has contributed to a number of scientific papers in regard to different genetic syndromes.

In the late 1990's when David Mowat was also working at the Westmead Children's Hospital, they together identified a group of children with very similar features, which they subsequently published in the Journal of Medical Genetics in 1998 as a "new" syndrome. That condition later became known as Mowat-Wilson syndrome, in acknowledgement of the fact that David and Meredith were the first to recognize and describe this important genetic syndrome.

## **Development of pluripotent stem cell models for Mowat-Wilson Syndrome**

### **Jay Vivian, Ph.D.**

Dr. Vivian is a Research Associate Professor in the Department of Pathology and Laboratory Medicine at the University of Kansas Medical Center. He also serves as the Scientific Director of the Transgenic and Gene Targeting Institutional Facility at KUMC since 2009. Dr. Vivian's ongoing research interests include the use of mouse and pluripotent stem cells as models for gene function and human disease. Dr. Vivian brings over 23 years' experience in pluripotent stem cell models. A major effort in his lab includes the isolation and engineering of patient specific human induced pluripotent stem cells for studies of human disease, with a focus on pediatric neurological and neurodegenerative disorders. He works closely with clinicians in these studies, including Dr. Merlin Butler. Dr. Vivian received his PhD in Genes and Development from the University of Texas M.D. Anderson Cancer Center, and postdoctoral studies at the University of North Carolina.

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## **Augmentative and Alternative Communication**

### **Marci Ravelli, M.S. CCC/SLP**

Marci Revelli is a speech language pathologist who specializes in the area of Augmentative Alternative Communication. With over 25 years of experience, Ms. Revelli has worked in hospitals, private and public schools as well as in private practice in Seattle and in the Boston area. She currently runs the Augmentative Alternative Communication program at Seattle Children's Hospital and provides AAC assessment, training and treatment for children of all ages. Ms. Revelli also runs Augmentative Alternative Communication Services (AACS), a program that provides school training, consultation and in-home AAC training and treatment. She has presented locally and nationally with a special interest in the use of mobile technology, curriculum modifications and inclusion programming for children with AAC needs.

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## **Seizure Diagnosis and Management in MWS**

### **Edward Novotny, M.D.**

Dr. Edward Novotny is a Professor of Neurology and Pediatrics at the University of Washington, School of Medicine and Seattle Children's Hospital. He completed his medical degree at Saint Louis University, his residency in Pediatrics at the University of California, Davis, and his clinical Neurology training at Stanford University. He went on to complete post-doctoral training in Epilepsy/Clinical Neurophysiology and MRI/MRS Biophysics at both Stanford University and Yale University.

Dr. Novotny specializes in the study of clinical neurophysiology, clinical epilepsy and advanced neuroimaging. He not only cares for patients with seizure disorders, but he also has participated in numerous research projects to help identify the underlying causes of epilepsy and best practices for treatment. This work has included clinical trials for newer treatments for children with seizures disorders.



## **Bowel Management in patients with Mowat-Wilson Syndrome**

### **Lusine Ambartsumyan, M.D.**

Dr. Ambartsumyan is currently an Assistant Professor of Pediatrics at University of Washington and the Director of the Gastrointestinal Motility Program at Seattle Children's Hospital. Her time is spent caring for children with gastrointestinal motility disorders in the inpatient and outpatient settings, outreach and regional clinics, and in the multidisciplinary clinics in conjunction with Divisions of Urology and Surgery at Seattle Children's Hospital. Dr. Ambartsumyan's clinical interests include mechanisms of fecal continence and defecation disorders in children with anorectal malformations, spinal cord dysraphisms, intractable constipation/fecal incontinence, and post surgical Hirschsprung Disease. She also specializes in the diagnosis and management of patients with gastroparesis, pseudo-obstruction, achalasia, and aerodigestive disorders. Dr. Ambartsumyan's research focuses on the pathophysiology and management of defecation disorders in children. Her primary research involves the study of mechanisms of functional and organic fecal incontinence (anorectal malformations, Hirschsprung disease), spinal cord modulation of anorectal function (neurogenic bowel), and the physiological relationship between the anorectum and the bladder in children with and without spinal cord malformations. She is currently a principal investigator in multiple clinical research studies specifically exploring the anorectal function and predictors of successful bowel management in patients with functional and organic fecal incontinence. Dr. Ambartsumyan has active teacher and mentorship roles for medical students, residents, fellows, junior faculty and ancillary staff. National visibility is evidenced by her role in NASPGHAN Neurogastroenterology & Motility Committee and speaking engagements in multiple national venues including UMDF Mitochondrial Medicine 2016 Symposium. International visibility is evidenced by her visiting professorship and speaking engagements at the Pediatric Urology and Surgery Symposium at Arabkir Women's and Children's Hospital in Yerevan Armenia, July 2014.

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## **Update on the Italian MWS Study**

### **William B. Dobyns, M. D.**

William B. Dobyns, MD, has made major contributions to the field of human genetics, particularly the nature and genetic causes of developmental disorders including mental retardation, autism and brain malformations. As both a medical geneticist and pediatric neurologist, Dr. Dobyns offers a rare combination of expertise. He examines patients with all types of genetic diseases, emphasizing children with complex developmental problems.

He also offers genetic counseling for families of these children. Dr. Dobyns is a recognized expert on many complex developmental disorders of the brain including mental retardation, autism, birth defects of the cerebellum such as Dandy-Walker malformation, and birth defects of the cerebral hemispheres such as microcephaly and megalencephaly (small and large brain size), lissencephaly or "smooth brain" disorder, and polymicrogyria (pebbled brain surface). He has made significant contributions to the understanding, classification and genetic cause of many different developmental disorders.

## CHILD CARE RELEASE AND CONSENT FOR MEDICAL/EMERGENCY TREATMENT

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Child's Name

Date of Birth

The undersigned parent or legal guardian hereby consents to the participation of the above-named child in the child care program.

The undersigned parent or legal guardian hereby expressly grants consent to the Mowat-Wilson Syndrome Foundation ("MWSF") and its volunteers to provide or authorize medical care for participating children, including emergency care.

The undersigned hereby further authorizes emergency transportation if necessary.

If there is no emergency, MWSF will first use reasonable efforts to contact the parent or guardian before administering or authorizing any treatment.

Finally, while MWSF and its volunteers endeavor to ensure the safety and well-being of all children participating in child care, accidents may still occur. The undersigned parent or legal guardian assumes all risk of injury or harm to the participating children, and agrees to fully release, indemnify, and hold harmless MWSF and its volunteers from any liability or claim arising from the participation of the above-named children in child care.

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Signature of Parent/Guardian

Date

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Printed Name of Parent/Guardian

Emergency Phone Number

- I will handle diaper changes/toileting.
- I agree to let caregivers handle diaper changes/toileting.

## PHOTO/VIDEO RELEASE

*The Mowat-Wilson Syndrome Foundation ("MWSF") has arranged for video recording and live internet broadcast of the conference to make it available to those unable to attend in person. MWSF may make the video available for purchase or distribution. Video and photographs will also be taken at the Friday night welcome party and throughout the weekend. Please know that, as a conference attendee, you and your children may appear in the video recording or live broadcast. We ask that all conference attendees sign the following release.*

I hereby irrevocably grant to the Mowat-Wilson Syndrome Foundation ("MWSF") the absolute right and permission to live stream, publish, distribute, and otherwise use conference video and photographs that include(s) my image and/or voice.

I understand video will be live streamed to the internet. I understand video and photographs may be published to the MWSF website, Facebook, and other media. I understand that I will not receive any compensation for, or rights to, these works.

I release and discharge MWSF, the videographer, and their agents from any and all claims arising out of, or in connection with, use of conference video and photographs. This release shall be binding upon me and my heirs, legal representatives, and assigns.

### Adult Attendees

I am a legally competent adult. I have read and understood the above, and I agree.

Adult Attendee Name	Signature	Date
Adult Attendee Name	Signature	Date
Adult Attendee Name	Signature	Date
Adult Attendee Name	Signature	Date

### Child Attendees

I am the parent/guardian of the following children (please list all children attending conference):

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As parent/guardian of the children listed above, I agree to the foregoing release on their behalf.

Parent/Guardian Name	Parent/Guardian Signature	Date
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# SPONSORS

LUNCH SPONSOR – The Eggers Family Trust

PROGRAM SPONSOR –



TOYS FOR CHILD CARE –

