

Alport Syndrome Symposium for Physicians, Researchers, and Families

Saturday, August 28, 2010

Target Audience

- Clinician-scientists, basic researchers and pediatric nephrologists interested in a state-of-the-art discussion of the genetics and pathogenesis of Alport syndrome, including insights regarding potential treatments gained from studies of animal models and registry data and questions needing further investigation
- Investigators at the trainee and junior faculty level who are curious about unanswered questions and research opportunities in the area of basement membrane disease, including Alport syndrome
- Patients and families with Alport syndrome who want to hear the latest information about Alport syndrome, including ongoing and projected clinical research studies

Goals and Objectives

- To bring together an international group of Alport syndrome researchers with affected patients and families
- To provide clinician-scientists, basic researchers, pediatric nephrologists and families with the most recent information on the genetics, clinical features, pathogenesis, diagnosis and treatment of Alport syndrome and the direction of future research
- To attract junior investigators and trainees to Alport syndrome research
- To build collaborations among patients and families, clinicians, researchers and the medical community that will translate to increased recruitment for clinical trials
- To educate and empower patients and families by allowing interaction with many of the foremost clinician-scientists and basic investigators involved in Alport syndrome research

Program

Morning Session

Moderators: Billy G. Hudson, PhD, Vanderbilt University, Nashville, TN, USA and Clifford E. Kashtan, MD, University of Minnesota, Minneapolis, MN, USA

9:00 – 9:30 AM	Overview of Pathogenesis Jeffrey H. Miner, PhD <i>Washington University St. Louis, MO, USA</i>
9:30 – 10:00 AM	Current Diagnosis and Treatment Martin C. Gregory, MD, PhD <i>University of Utah Salt Lake City, UT, USA</i>
10:00 – 10:30 AM	Outcomes and Transplants Patrick Niaudet, MD <i>Hôpital Necker-Enfants Malades Paris, France</i>
10:30 – 10:45 AM	Break
10:45 – 11:15 AM	Pharmaceutical Treatment Christoph Licht, MD <i>University of Toronto Toronto, Canada</i>
11:15 – 11:45 AM	Biological Treatment Oliver Gross, PD, MD, FASN <i>University of Göttingen</i>

Göttingen, Germany

11:45 AM – 12:15 PM	Hearing Loss Dominic Cosgrove, PhD <i>Boystown National Research Hospital Omaha, NE, USA</i>
12:15 – 12:30 PM	Q&A of Morning Sessions
12:30 – 1:30 PM	Break

Afternoon Session

Moderators: Billy G. Hudson, PhD, Vanderbilt University, Nashville, TN, USA and Clifford E. Kashtan, MD, University of Minnesota, Minneapolis, MN, USA

1:30 – 2:00 PM	Women and Alport Syndrome Michelle Rheault, MD <i>University of Minnesota Minneapolis, MN, USA</i>
2:00 – 2:30 PM	Registries/Trials – Alport Syndrome Treatments and Outcomes Registry (ASTOR) Clifford E. Kashtan, MD <i>University of Minnesota Minneapolis, MN, USA</i>
2:30 – 3:00 PM	Research Process Overview Clifford E. Kashtan, MD <i>University of Minnesota Minneapolis, MN, USA</i>
3:00 – 3:30 PM	Children and Adolescents with Chronic Kidney Disease Bradley Warady, MD <i>Children's Mercy Hospital Kansas City, MO, USA</i>
3:30 – 3:45 PM	Break
3:45 – 5 :00 PM	Q&A of Afternoon Sessions Panel or Small Group Discussion

Presentation Descriptions

Presentation 1: Overview of Pathogenesis, Jeffrey H. Miner, PhD

Dr. Miner is one of the world's foremost authorities on the ontogeny of basement membranes and the roles of different basement membrane components in maintaining glomerular permselectivity. He will provide a comprehensive view of current knowledge regarding glomerular basement membrane structure and function, providing a context for the talks to follow.

At the end of the presentation, the attendee will be able to describe how the major glomerular basement membrane proteins contribute to normal glomerular barrier function.

Presentation 2: Current Diagnosis and Treatment, Martin C. Gregory, MD, PhD

Dr. Gregory, director of the Alport Syndrome Study at the University of Utah, has performed seminal work on the genetics of Alport syndrome. He will present up-to-date information on the molecular genetics of Alport syndrome, and provide the latest information on genotype-phenotype correlations.

At the end of the presentation, the attendee will be able to describe the genetics of Alport syndrome and the impact of different mutations on the rapidity of progression to end-stage renal disease and deafness.

Presentation 3: Outcomes and Transplants, *Patrick Niaudet, MD*

Dr. Niaudet is a world-renowned pediatric nephrologist, who will discuss aspects of the clinical progression of Alport disease manifestations and outcomes after renal transplantation, including current knowledge regarding post-transplant anti-glomerular basement nephritis in the renal allograft.

At the end of the presentation, the attendee will be able to describe the typical clinical course of Alport kidney, cochlear and ocular disease in affected males and females and the outcomes of renal transplantation.

Presentation 4: Pharmaceutical Treatment, *Christoph Licht, MD*

Dr. Licht will present published and unpublished data regarding the impact of pharmacologic therapies on Alport kidney disease in experimental animals and in human populations.

At the end of the presentation, the attendee will be able to identify pharmacologic approaches that have shown promise in experimental animals with Alport syndrome, and will understand the challenges involved in applying these observations to people with Alport syndrome.

Presentation 5: Biological Treatment, *Oliver Gross, PD, MD, FASN*

Dr. Gross, who is Executive Director of the European Alport Registry, has performed numerous studies of Alport syndrome therapies in experimental animals, including stem cell therapies. He will discuss the current status of stem cell and gene replacement therapies for the disease.

At the end of the presentation, the attendee will be able to describe the findings of studies of stem cell therapies in Alport syndrome, and the challenges associated with developing stem cell and gene replacement approaches for treatment of human Alport syndrome.

Presentation 6: Hearing Loss, *Dominic Cosgrove, PhD*

Dr. Cosgrove will provide a state-of-the-art presentation on the role of basement membranes in cochlear function and current theories regarding the pathogenesis of hearing loss in Alport syndrome.

At the end of the presentation, the attendee will be able to describe the alterations of cochlear architecture and function found in Alport subjects, and how these changes may be related to hearing loss.

Presentation 7: Women and Alport Syndrome, *Michelle Rheault, MD*

Dr. Rheault has characterized the first transgenic model of X-linked Alport syndrome. She will describe new findings regarding the role of X-chromosome inactivation in determining the outcome of X-linked Alport syndrome in heterozygous females, and discuss how kidney donation by heterozygous females should be approached.

At the end of the presentation, the attendee will be able to describe how X-chromosome inactivation influences the course and outcome of X-linked Alport syndrome in heterozygous females.

Presentation 8: Registries/Trials – Alport Syndrome Treatments and Outcomes Registry (ASTOR), *Clifford E. Kashtan, MD*

Dr. Kashtan is the Executive Director of the Alport Syndrome Treatments and Outcomes Registry (ASTOR). He will discuss the roles of registries in the United States and Europe in supporting clinical studies of Alport syndrome therapies and provide an update on new findings of registry studies. He will also present an overview of the clinical research process.

At the end of the presentation, the attendee will be able to discuss the latest findings of studies carried out by Alport syndrome registries.

Presentation 9: Research Process Overview, *Clifford E. Kashtan, MD*

Dr. Kashtan will present an overview of the clinical research process including a description of the activities associated with design, development and implementation of clinical research policies and procedures.

At the end of the presentation, the attendee will understand the complexities of designing and implementing clinical research studies of rare genetic disorders.

Presentation 10: Children and Adolescents with Chronic Kidney Disease, *Bradley Warady, MD*

Dr. Warady is one of the world's authorities on chronic renal failure and end-stage renal disease in children and adolescents, and on the challenges of insuring adherence with dietary prescriptions and medication regimens in adolescent patients. He will present current information on the causes and correction of dietary and medication non-adherence.

At the end of the presentation, the attendee will be able to discuss the causes and approaches to correction of dietary and medication non-adherence in adolescent patients.