

The **Poss**ABILITIES Project!



Celebrating the amazing uniqueness of our children

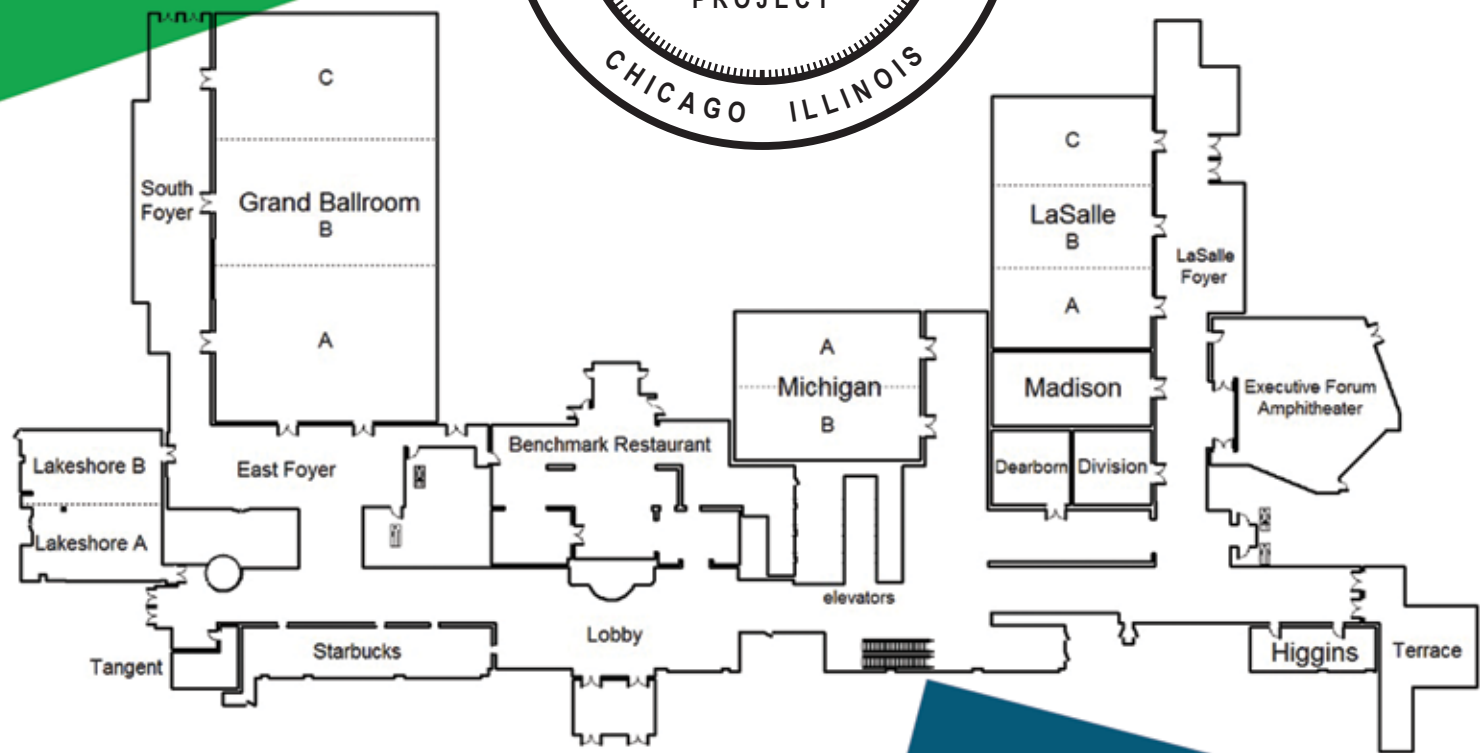


July 22-25, 2015



The Westin O'Hare





The Westin O'Hare
Meeting Rooms
Floor Plan

Welcome families and friends to the **2015 JSRDF** Conference – The Poss**ABILITIES** Project!

We are excited that you are here in the vibrant, amazing, and beautiful City of Chicago, and even more thrilled that we get to spend the next few days with you. Our extraordinary Conference Committee – **Jackie Olson, Jenni Swenson, and Denise Mack** – have worked tirelessly to create an educational, fun, informative, and special event. Thank you for all of the work you have put into this project!

We share a common journey together through the diagnosis of **Joubert Syndrome**. But, more than that, we have all become family. We share our successes, our challenges, our hopes, and our dreams. We celebrate the amazing uniqueness of our children. We grieve for those who have gone before us, and we honor their memory and their spirit always.

The biannual conference allows us to convene in one place together. Our valiant conference planning team spends innumerable hours working to ensure that the conference is an extraordinary and worthwhile event for all of us. But, it is not simply chosen because of a particular city or a particular hotel. We choose to be together because we feel safe. We know that we are among friends – and family. Here, there is no judgment. Just a comfortable, welcoming, and safe place to be.

Welcome home!

Here in Chicago **we want to focus on what our children can do**, and celebrate their **successes**, their **milestones**, their **courage** and their **perseverance**. We honor all our families for the love, dedication, passion, and drive that we see every day in the stories that you share around social media and beyond.

The Foundation, too, has been hard at work since Minneapolis 2013! The goals that current President Stephen Mack set into place have built on an already strong foundation and have led the Board to take the next steps to moving the Foundation forward. **Some highlights:**

- In 2014, the Foundation eliminated the required membership fee and has focused energy on fundraising. This translated into nearly \$30,000 in successful fundraising campaigns for the Foundation. We encourage everyone to continue that success in 2015 and beyond!
- The JS-LIFE and Mosaic projects catapulted the JSRDF to the national and international stages, highlighting the growing movement of patient-centered information sharing and research. JS-LIFE is THE model program showing how successful patient/client involvement is in driving research and information exchange. To date, you have answered more than 50,000 questions sharing your experiences and your knowledge! This project has earned the JSRDF both national and international recognition as a leading disease advocacy organization.
- We are working hard to upgrade and enhance the JSRDF website. Stay tuned for exciting things!
- In 2014 and the beginning of 2015, we held two town hall meetings – never done before! Nearly 150 people attended both sessions and we plan to continue those in the future.

President-elect Eric Swenson will begin his term as President by the time this conference ends. We are very excited for his leadership, vision and goals for the Foundation!

Welcome once again to Chicago! We are very happy to be here with you and hope you enjoy the **PossABILITIES** Project!

Stephen Mack
President

Eric Swenson
President-elect

Wednesday July 22

3:00-4:15 pm **Conference Orientation** – Provides an overview of conference events and introduction of “partner families” who help answer questions and navigate the weekend.

Executive Forum

3:00-4:15 pm **Teen/Young Adult Social for JSRD Individuals** – Meet new and reunite with old friends from our JS family. Moderated by a social worker, these sessions will help individuals with JS get to know each other. Suggested ages 14 – 25 with JS.

Madison

4:30-5:30 pm **Our Journeys with Joubert Syndrome, Scott MacLellan and Noldon Sparks** – Hear from Scott and Noldon, 2 remarkable young men with JSRD, about their milestones and challenges on their journey with JS. Followed by short Q and A with their parents.

Executive Forum

4:30-5:30 pm **Sibling Social - Led by Kim Walton & Melissa Fields**

Having a special needs child is a challenge for the entire family. This year we will have several activities for siblings of all ages! All siblings from kindergarten through middle school are invited to join us for our SIBLING SOCIAL to kick off these events! We will have games and activities designed to get to know each other a little better.

Madison

6:00-7:30 pm **Opening Dinner and Welcome to the PossABILITIES Project!** – Caesar Salad, Bruschetta, Baked Penne, Chicken Parmesan, Fettuccini Primavera and Medley of Roasted Vegetables.

For Children: Pizzas, Elbow and Bowtie Pasta, Baby Carrots and Breadsticks.

LaSalle Ballroom

About 7:45 pm **Music on the Patio** – Unwind after dinner with other conference attendees.

LaSalle Lobby and Patio

Thursday July 23

7:30-9:00 am **Buffet Breakfast** – Thursday Breakfast: Scrambled Eggs, Crisp Bacon, Breakfast Potatoes, Assorted Muffins and Breakfast Breads, Sliced Fruit, Assorted Juices, Milk and Coffee.

LaSalle Ballroom

8:30-8:55 am **Joubert Syndrome, a Historical Perspective, Eugen Boltshauser, MD**
Dr. Boltshauser will present an overview of Joubert Syndrome since its discovery in 1969. New diagnostic techniques and increasing clinical experience have greatly expanded our knowledge about this complex syndrome.

Executive Forum

9:00-9:25 am **Kidney Issues in Joubert Syndrome and Related Disorders, Jordan Symons, MD**
Children with Joubert Syndrome and related disorders may have kidney involvement as part of their overall medical condition. This presentation will present basic information on the kidneys and what they do, explain the differences in kidneys for children with Joubert Syndrome and related disorders, discuss the effects of the kidney-related problems, and explore what families can do to optimize kidney status.

Executive Forum

9:30-9:55 am **Liver and GI Issues in JSRD, Theo Heller, MD** – Dr. Heller will discuss the liver disease and some of the more common gastrointestinal manifestations found in JSRD.

Executive Forum

10:00-10:25 am **Joubert Syndrome and Ophthalmology Issues, Wadih Zein, MD** – Dr. Zein’s presentation will address the ocular (eye) manifestations of Joubert Syndrome with an attempt at correlation with molecular findings. In addition, recent advances in research for therapies of hereditary retinal degenerations will be discussed.

Executive Forum

11:00-11:20 am **UW Research Update: Genes, Phenotypes and Mechanisms - Dan Doherty, MD, PhD**
Dr. Doherty will summarize recent work from his laboratory including new genetic causes, clinical features, detailed gene-phenotype correlations and functional work on underlying mechanisms using patient cell lines.

Executive Forum

10:30-12:45 pm **Teen/Young Adult Topics for JSRD individuals and group lunch** – Get together again with JS teens and young adults and talk about your challenges and successes. Moderated by Scott MacLellan.

Madison

10:30-12:45 pm **Sibling Workshops (grades K-4th), Kim Walton & Melissa Fields**
During these sessions, we will answer some of the following questions: What do you love about your brother or sister? What do you dislike? Name some feeling words that come to mind when you think about your sibling. What have the two of you learned from one another? What would you change if you could? What could your parents do to make things easier for you?

Michigan B

11:30-11:55 pm **Neurological Explanation of Behavior in Hindbrain Disorders, Nina de Lacy, MD, MBA** – The hindbrain is perhaps more usefully thought of as an important member of many brain networks which nonetheless involve other brain regions to produce behaviors. Learn about the cognitions and behaviors the hindbrain is associated with. This will help parents understand WHY our JSRD loved ones may have negative behaviors.

Executive Forum

12:00-1:00 pm **Buffet Lunch** – Grilled Hamburgers, Cheeseburgers, Hot Dogs, Potato Salad, Chips and Dip, Corn, Baked Beans with Onion and Bacon, Sliced Watermelon, Sliced Cheddar and American Cheeses, Fresh Condiments, Lemonade, Iced Tea and Coffee.

LaSalle Ballroom

1:00-2:00 pm **JS-LIFE: Put Yourself in the Research Driving Seat, Matt Smith, Dana Knutzen**
Interested in learning more about how you can contribute directly to research to advance our understanding of Joubert Syndrome? Join your JS-LIFE team as they lead you through the JS-LIFE survey and explain how the information you enter directly impacts individuals with JS. Then stay to explore the values of Mosaic, where your shared experiences and knowledge can shape future research studies to improve the lives of those living with Joubert Syndrome. The JS-LIFE and Mosaic projects are research done by YOU for YOU. With your involvement, any research idea is possible. Come explore the opportunities!

Executive Forum

2:00-3:00 pm **Sibling Workshops (grades 5th-8th), Kim Walton & Melissa Fields** – During these sessions, we will answer some of the following questions: What do you love about your brother or sister? What do you dislike? Name some feeling words that come to mind when you think about your sibling. What have the two of you learned from one another? What would you change if you could? What could your parents do to make things easier for you?

Michigan B

2:05-2:30 pm **NIH Research Update, Meral Gunay-Aygun, MD** – Dr. Gunay’s talk will summarize the results of the 100 Joubert Syndrome patients prospectively evaluated at the National Institutes of Health (NIH) Clinical Center in Bethesda, Maryland. Evaluations included complete eye examinations, formal cognitive tests, ultrasonography and magnetic resonance imaging of kidneys, liver and spleen, and comprehensive blood and urine tests to evaluate kidney and liver function and growth.

Executive Forum

2:35-3:00 pm **Kidney Disease in Joubert Syndrome Latest Research Provides Hope for Future Treatments, John Sayer, MD** – In his talk, Dr. Sayer will detail studies on animals with Cep290 mutations and how this ciliopathy affects the kidneys. In studies using cells from mice and human kidneys with Cep290 mutations he will discuss the problems related to Cilia Dysfunction and how treatments can be used to rescue this defect. He will describe how this exciting work will hopefully pave the way towards human studies in Joubert Syndrome patients.

Executive Forum

3:30-3:55 pm **Respiratory and Sleep Issues in JS, Maida Chen, MD** – Dr. Chen will speak to clinical presentation and treatments of respiratory and sleep disorders related to Joubert Syndrome.

Executive Forum

4:00-5:00 pm **Panel Presentations of Updated Healthcare Recommendations** – Everyone should attend this one! The healthcare providers attending the conference will discuss updated recommendations for evaluation, monitoring, and treatment of individuals with Joubert Syndrome. Moderated by Dan Doherty, MD, PhD.

Executive Forum

4:00-6:00 pm **Whose Life is it, Anyway? – Barbara J. Thomas, MEd, CSE, JSOC, CSES.** Barb Thomas will provide our JSRD teens and young adults with the best information they need to make healthy and safe choices for themselves.

Madison

5:15-6:00 pm **Prenatal Diagnosis and Family Planning, Ian Glass, MD** – Dr. Glass will discuss the reproductive options currently available to couples and their family members to address prenatal diagnosis concerns including prenatal imaging and DNA testing specific for Joubert Syndrome.

Executive Forum

6:00 pm **Dinner on your Own** – Look in your conference bag for local dining vouchers – enjoy some tastes of Chicago with a portion of the sales going back to the Foundation.

7:00 pm **Dad’s Night Out** – Kings Lanes — 5505 Park Place, Rosemont, IL (not included in conference fees – pay on your own)

7:30 pm **Teen / Young Adult Movie Night for JSRD and Siblings** – Movie night! Look for movie choice on announcement board.

Executive Forum

Friday July 24

7:30-9:00 am **Buffet Breakfast** – Scrambled Eggs, Sausage, Chef's Choice of Breakfast Potatoes, Assorted Muffins and Breakfast Bread, Sliced Fruit, Assorted Juices, Milk and Coffee.
LaSalle Ballroom

8:30-12:00 pm **MD Consults** – These 10 minute consultations are designed to supplement your current healthcare team with specialists who may be more knowledgeable of JS. This is not a doctor's appointment, as the MDs are not licensed to practice in the state of IL nor do they have enough specific information about your child to give a prescribed treatment plan. You may however receive information about what is typical in JSRD (limit 2 appointments per family unless there are same day openings). Appointments can be scheduled at the registration desk.
Madison

8:30-12:00 pm **Joubert Syndrome Research Opportunities**
1:15-6:00 pm Consider participating in research studies our friends at The University of Washington Seattle and NIH are conducting. Look for info and instructions in your family welcome bag for information.
Second floor – Waveland and Fulton

8:30-9:30 am **The Power of Two: A Workshop Focusing on Couples with Children that have Joubert Syndrome & Related Disorders, Sabrina J. Pippin, MA** – This workshop will focus on improving the relationship between the parents who have children with Joubert Syndrome and related disorders. Additional stressors are present in a couple when there are health concerns in their children. Frequently, the needs of a child take precedence before a couple's needs, which can add imbalance and stress within a relationship. Exploration of a couple's needs and expectations will also be presented at this workshop. Role plays will be utilized to allow for comprehension of the material covered.
Executive Forum

9:45-10:45 am **Special Needs Estate & Future Planning, Brian and Benjamin Rubin** – The who, what, where, when, why and how of Special Needs Estate Planning including: types of and appropriate uses of Special Needs Trusts, gifting by grandparents & extended family members, the impact of divorce & child support on government benefit, the new ABLE Act, Guardianships & Alternatives, Providing guidance, education and information for the "Future Team".
Executive Forum
Or

Breaking it Down: An Open Forum on Genetics, Inheritance and the Clinical Features of JSRD, Dana Knutzen – After a full day of medical and scientific presentations, many of you may have lingering or unanswered questions about genetics, inheritance and the clinical features associated with JSRD. Maybe you didn't catch what was said, didn't understand a particular topic, or are just utterly confused and

overwhelmed. If this is you, be sure to attend this open forum where Dana, a genetic counselor, will attempt to break down your questions with easy to understand explanations.
LaSalle A/B

11:00-12:00 pm **Planning for Disabled Adults - What Do I Do When My Child Attains Legal Age? Tom McGuire**
Discussion of mechanics of guardianship proceedings for disabled adults including court process, management of assets, creation of estate planning documents, OBRA trusts, and other considerations.
Executive Forum

Or

Fight or Flight? Kim Walton – Stress physiology: Stress can take a huge toll on our physical and emotional health. Learn what happens in our bodies as we deal with the stresses of daily life.
LaSalle

12:00-1:00 pm **Buffet Lunch** – Hard and Soft Shell Tacos, Taco Seasoned Ground Beef, Olives, Onion, Tomato, Lettuce, Cheese, Salsa, Sour Cream, Tortilla Chips, Refried Beans, Spanish Rice, Lemonade, Iced Tea and Coffee.
LaSalle Ballroom

1:30-3:00 pm **Introduction to Alternative Therapies - Laura Houston & Rebecca Cain, The Therapy Tree** – This introduction to alternative therapies will cover the basic information on Essential Oils, Drop Therapy, Feldenkrais and Craniosacral Therapies.
LaSalle

1:30-2:30 pm **Men Facing Special Needs, James Harlan**
A discussion for men about how to manage the emotions and challenges about raising a special needs child, as well as the importance of seeking support.
Executive Forum

2:45-4:45 pm **S-E-X vs. Sexuality - Barbara J. Thomas MEd, CSE, JSOC** – What parents need to know about working and living with your teens and young adults. This session is for parents — not teens/young adults.
Executive Forum

3:15-4:00 pm **Yoga for Caregivers, Bernardo Isacovici**
A restorative yoga practice designed to relieve stress, assist in muscle stretches, and relaxation. This will be a gentle practice designed to restore, relax and breathe.
LaSalle

4:15-5:00 **Yoga for Affected Individuals, Bernardo Isacovici** – A yoga practice designed to help children with Joubert Syndrome to gently strengthen muscles and achieve general relaxation. This will be a fun session including play and involve breathing exercises appropriate for children of all ages.
LaSalle

5:15-6:00 pm **Celebration of Life Ceremony** – This reflective ceremony honors the memories of loved ones lost to JSRD. Everyone is welcome; however attend only if you feel comfortable. Coordinated by Kelly Altman (mom to Alex 2/17/99 -11/19/10).
Executive Forum

6:00 pm **Dinner on your Own**

Saturday July 25

7:30-9:00 am **Buffet Breakfast** – Biscuits and Sausage Gravy, Oatmeal, Danish, Fruit, Assorted Juices, Milk and Coffee.
LaSalle Ballroom

9:00-10:00 am **A Parents Toolbox to Tackle Bullying - Kortney Peagram, PhD** – This presentation is designed to inform parents about cyberbullying, bullying, aggression, and conflict. Facilitators work with parents on identifying these behaviors and coaching parents in developing positive strategies to use with their children and teens. Sessions include a facilitated group discussion to give parents an opportunity to talk to other parents about challenges in their home and share ideas. The session works to educate them about negative social behaviors and develop a parent's "toolbox" with strategies to fit their unique family needs. Our team provides parents with insights, ideas, strategies, and recommendations to use in their home.
LaSalle

Or

How Occupational Therapy Supports Functional Visual Development & Participation - Jennifer Rhein, OT – This discussion will focus on the role of occupational therapy both in general as well as specific to challenges faced by many individuals with JSRD. Special focus will be placed on how OT can help to support functional visual development and its impact on participation in daily activities. Practical strategies will be shared for families to use at home and questions are encouraged.
Executive Forum

10:00-11:00 am **Life Manual Workshop, Denise Mack** – A life manual is a booklet created to provide caregivers with instructions on the day to day care for your child. Denise will be discussing how a life manual can be beneficial for caregivers and will give attendees the tools they need to manage what is entailed in caring for their loved one. She will be providing templates to create customized life manuals. Attendees are encouraged to bring a device or laptop to this session.
Executive Forum

10:00-11:45 pm **Oxygen for Special Parents & Caregivers, Kim Walton** – We learned what it is, now let's learn how to handle it! Come relax and learn (and practice) some simple ways to minimize stress in your life and how these same techniques can be used with your spouse, family or KIDS! Put the oxygen mask on your own face first! Care of the child cannot exceed care of the parent or caregiver!
Madison

11:00-11:45 am **Who is My Child? Workshop, Denise Mack**
Denise will be sharing her experience with building relationships with IEP teams. This will be a hands on session where attendees will have a chance to highlight all of the traits that makes their loved one special. Templates will be provided to create a

custom information sheet to provide IEP team members as well as caregivers. Attendees are invited to bring a device or laptop to this session.
Executive Forum

12:00-1:00 pm **Lunch on Your Own**

1:00-3:00 pm **Parent Shares** – A time for folks facing similar struggles to get together and share ideas - no speaker.... just a casual time to share in a pre-set time and place.

Teen/Young Adult Parent Share
Scott MacLellan
Madison

Birth to 10 Parent Share
TBD

1:00-2:00 pm **Kid's Music Round Music & Movement Parent/ Child Class, Amanda Tulamalo**
This session is a hands on class for parents/ grandparents/caregivers to attend and participate with children. Learning basic music competence skills can tap into the brain in some amazing ways. Research proves that learning music can help children with language skills. For Amanda's own two children who are diagnosed with JS, she has witnessed how early childhood music learning helped with speech and language, fine motor skills, sensory processing, and movement. Learn how you can help your child learn basic music competence skills through movement and singing, while having fun and creating memories that will last a lifetime.
LaSalle Ballroom

2:00-3:00 pm **Music Party! - Amanda Tulumalo** – Grownups and children alike will sing, dance, and play instruments together. This is for everyone!
LaSalle Ballroom

3:00-4:00 pm **JSRDF General Meeting** – Consider getting involved! Your Board of Directors will share important information about the Foundation, our mission and our vision. You will also have a chance to join us if you are interested in volunteering for Board service!
Executive Forum

4:30-6:00 pm **Volunteer & Fundraising Fair - And Silent Auction Open** – Coordinated by Dana Knutzen – One FREE drink ticket for each full registration attendee that participates in the Volunteer Fair. Learn about big and small ways to get involved with the JSRDF.
Madison

6:00-10:00 pm **Closing Dinner and Dance**
Always a highlight of the conference. Celebrate with your new friends and dance as our DJ spins some tunes. Grilled Chicken and Penne Pasta, Roasted Beef Sirloin, or Vegetarian Option. All dinners come with Salad, Vegetable, Bread, Dessert, Coffee, Milk and Water. Kids option – Chicken Fingers, French Fries, Green Beans, Fruit Cup and Milk.
LaSalle Ballroom

Guest Medical Speaker **Bios**



Eugene Boltshauser, MD - Pediatric Neurology

Dr. Boltshauser is an emeritus professor (retired) of Child Neurology and is the former head of the Division of Child Neurology at Children's Hospital in Zürich, Switzerland. He has a longstanding interest in Joubert Syndrome and cerebellar malformations. His first paper on Joubert Syndrome dates back to 1977! He continues to publish articles on the clinical, neuropathological, imaging, and genetic aspects of Joubert Syndrome. Dr. Boltshauser first met Cheryl Duquette, one of the JSRDF's co-founders, in 1985 while on hospital rounds in Baltimore. It was at this time that the diagnosis of JS was given for her first child. Dr. Boltshauser has since attended several JSRDF conferences over the years—Montreal, Baltimore, New Orleans, Irving/Dallas, Cincinnati, Orlando, Minneapolis and now Chicago.

Maida Chen, MD - Pediatric Sleep Medicine

Dr. Chen is an Associate Professor of Pediatrics in the Division of Pediatric Pulmonary and Sleep Medicine, at the University of Washington and Seattle Children's Hospital. Dr. Chen has extensive clinical experience in pediatric sleep disorders, with a particular focus on patients with abnormalities in central respiratory control, including JS. Pediatric sleep medicine is a relatively new discipline, with characterization of many disorders with abnormal respiratory control, such as JS, still in early descriptive phases. She earned her MD at Northwestern University, trained in pediatrics at Rush-Presbyterian St. Luke's Medical Center, and completed her Pediatric Pulmonary Fellowship at Children's Hospital Los Angeles. She is currently board certified in both Pediatric Pulmonology and Sleep Medicine. She will give a talk on clinical and research advances in respiratory and sleep disorders related to Joubert Syndrome.

Nina de Lacy, MD, MBA - Neurobehavior

Nina de Lacy, MD MBA is a Fellow in Child and Adolescent Psychiatry at the University of Washington. She received her MD from UCSF, her MBA from Northwestern and her BA from Oxford University. At UCSF she first developed her longstanding interest in macroscale brain connectivity systems, thereafter doing her adult residency training at the UW in the Neuroscience Program. Dr. de Lacy is a systems neuroscientist who focuses on the genetic control of the development of brain connectivity via imaging genetics studies using techniques from advanced neuroimaging and computational genetics in specific neurogenetic disorders, autism and attention. Clinically, Dr de Lacy's interests are centered on neuropsychopharmacology in the developmental neuropsychiatric disorders. She cared for individuals with genetic syndromes and complex brain malformations in a number of specialty clinics at SCH and the UW and now manages psychiatry care in the Developmental Neuropsychiatry Program at SCH. She is also interested in neurophilosophy and spent a number of years training as a Lacanian psychoanalyst.

Dan Doherty, MD, PhD, Developmental-Behavioral Pediatrics

Dr. Doherty grew up in upstate New York and earned his MD/PhD at the University of California, San Francisco (UCSF), followed by Pediatric Residency and Developmental-Behavioral Pediatrics Fellowship at the University of Washington (UW) in Seattle. Now an associate professor at UW, he leads the UW Hindbrain Malformation Research Program and is Director of the Genetics Core for the UW Center on Human Development and Disability. Clinically, he cares for children with all types of disabilities and recently established the Seattle Children's Hospital Developmental Neuropsychiatry Program to provide integrated neurodevelopmental and psychiatric care for complex patients. He also provides prenatal counseling for women carrying fetuses with central nervous system imaging abnormalities. His research interests include identifying the genetic causes of hindbrain malformations such as Joubert Syndrome and studying these disorders in model systems such as zebrafish and cultured cells. Ultimately the research goals are to provide better information and treatments for patients with brain malformations as well as learn more about normal and abnormal brain development. When he's not at work, Dan spends time with his wife and two children, mostly at soccer games, music recitals, and plays.

Ian Glass, MBChB, MD - Genetics

Dr. Glass grew up and earned his MBChB medical degrees in New Zealand. He trained in pediatrics and genetics in the United Kingdom, earning his MD focusing on the genetic basis of inherited forms of mental retardation. He then relocated to the US for postdoctoral basic research and US genetics training at the University of California, San Francisco (UCSF) and Mount Sinai School of Medicine in NYC. Upon completion of his US training, he relocated to Brisbane, Australia to become a medical genetics staff specialist while also working on basic research. In 2000, Dr. Glass returned to the US to the University of Washington in Seattle where he became the clinical director for genetic services at Seattle Children's Hospital. In this post, he consults for genetic disorders for children, pregnant couples and adults with a host of genetic disorders and birth defects. Dr. Glass has a long standing interest in neurogenetic disorders and also became deeply immersed in the genetic bases of Joubert syndrome. His research goals are to improve the quality of information and health care provided for children and families with Joubert syndrome and related disorders. Ian lives in Seattle with his US born wife and two children and enjoys the beautiful Pacific Northwest.



Meral Gunay-Aygun, MD - Genetics

Dr. Gunay-Aygun is an Associate Professor of Pediatrics and Genetics at John Hopkins University School of Medicine, and an Attending Senior Staff at the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH). At NIH, Dr. Gunay-Aygun has been the principal investigator of a research study on ciliopathies, since 2003. To date, she has evaluated 280 ciliopathy patients under this protocol including 100 patients with Joubert Syndrome and related disorders.

Dr. Gunay-Aygun earned her medical degree in Turkey in 1987. In 1995, she came to the United States, and completed residency training in Pediatrics and Medical Genetics at Case Western Reserve University in Cleveland, OH. In 2002, she received a fellowship in Biochemical Genetics at NHGRI/NIH and joined their staff in 2004. She is a member of several professional medical organizations and is a scientific reviewer for several medical journals.

Theo Heller, MD - Gastroenterology

Dr. Heller is an Investigator in the Liver Diseases Branch of the National Institute of Diabetes, Digestive and Kidney Diseases in the National Institutes of Health. His interest centers on non-cirrhotic portal hypertension, or high blood pressure in the liver without cirrhosis and its complications. His research is primarily clinical with a translational component. He is particularly interested in and studies patients with Congenital Hepatic Fibrosis, Caroli's, and Nodular Regenerative Hyperplasia. The current focus of this research is how to define and measure progression in these syndromes, with an ultimate goal of developing better management strategies and therapies. He also has research interests in viral Hepatitis.

Dana Knutzen, MS, CGC - Genetic Counselor

Dana Knutzen is completing her second term on the board of directors for the Joubert Syndrome and Related Disorders Foundation (JSRDF). As a board member, she has worked to provide genetic education to the foundation's members while assisting with various research opportunities and volunteer engagement. Her work with the JSRDF originated during her prior employment with the University of Washington Hindbrain Malformation Research Program from 2003-2010.

For the past 10+ years, she has focused her clinical skills in the fields of prenatal and pediatric genetic counseling. Most recently, she has transitioned to the field of cancer genetics and has recently developed a cancer genetic counseling service for a local hospital system. She received her master's degree in genetic counseling from the University of Michigan in 2002. She is board certified through the American Board of Genetic Counseling and is a licensed genetic counselor in Washington State.

Dana has had the pleasure of attending previous JSRDF conferences from 2004 - 2013 and has had a long standing relationship with many members of the organization. She currently resides in Gig Harbor, Washington with her husband, Steve, and sons Caleb (8 yrs) and Juno (4 yrs).

John Sayer, MD, Nephrology

Dr. Sayer is a Clinical Lecturer at the Institute of Genetic Medicine, Newcastle University, UK and a Consultant Nephrologist. His research focus is to understand the molecular genetics underlying cystic kidney diseases, in particular ciliopathies. He has identified several new genetic causes of Nephronophthisis and Joubert Syndrome. Now using model systems, including cell culture, zebrafish, murine models and human urine derived renal epithelial cells, he is exploring disease mechanisms and therapeutic agents for Joubert Syndrome.

Jordan Symons, MD, Nephrology

Jordan Symons is Attending Nephrologist at Seattle Children's Hospital. He cares for children with all forms of Kidney Disease in the General Nephrology, Dialysis and Transplant Programs, including those with kidney-related complications of Joubert Syndrome. In addition to his clinical duties, Dr. Symons teaches at the University of Washington School of Medicine, where he is Professor of Pediatrics. Originally from New York City, Dr. Symons earned his Medical Degree in New York at Columbia University College of Physicians and Surgeons, completed Pediatric residency training at Children's Memorial Hospital in Chicago, and then took his Pediatric Nephrology training at Children's Hospital in Boston before moving to Seattle.

Wadih Zein, MD - Ophthalmic Genetics/Pediatric Ophthalmology

Dr. Zein, National Eye Institute Staff Clinician and Principal Investigator, studies hereditary diseases affecting the eye including Retinitis Pigmentosa and Cone-Rod Dystrophy. He has extensive experience with systemic diseases with ocular manifestations such as Bardet-Biedl syndrome, Alstrom syndrome, Joubert Syndrome (all classified as Ciliopathies). Dr. Zein's research focuses on disease natural history and Pathobiology in the hopes of gaining a greater understanding of these conditions. He has participated in the design and implementation of clinical trials for hereditary eye diseases. He also concentrates on the psychophysical and electrophysiological studies of ocular function. Dr. Zein serves as an ophthalmology Advisor and member of the Scientific Board of Patient Associations for Hereditary Diseases with Ocular Manifestations. Dr. Zein has authored and co-authored numerous scientific manuscripts and book chapters on topics related to Pediatric Ophthalmology and Ophthalmic Genetics. His interest in the study of the ocular manifestations of Joubert Syndrome and related disorders was solidified by his participation in the NHGRI/NIH study.



Presenter Bios

Brian N. Rubin

Attorney Brian Rubin, a practicing attorney since 1976, is the parent of three children, one of whom, Mitchell, has Autism. Brian's law practice, since 1982, when Mitchell was one year old, has been dedicated to serving the legal and future planning needs of his fellow families of children and adults with intellectual disabilities, developmental disabilities, and/or mental illness.

Brian has been elected as a Fellow of the American College of Trust and Estate Counsel (ACTEC), and has received the Martindale-Hubbell AV® Preeminent™ Rating. Brian is the President of The Arc of Illinois; has been a member of the Board of Directors of Clearbrook (serving more than 6,000 individuals with developmental disabilities) since 1989; is a member of the Board of Directors of Pact, Inc. (PAS/ISC Agent for DuPage County, also available to serve as Trustee of Special Needs Trusts, as well as available to serve as Guardian); is Past Chairman of the Board of Directors of the SLSF, the foundation supporting the Northwest Suburban Special Recreation Association (NWSRA); served as the founding Chairman of the Board of Trustees of the Foundation of The Special Education District of Lake County (SEDOL Foundation); served on the Board of Directors of Keshet (Jewish Parents of Children with Special Needs); served on the Board of Directors of Northpointe Resources; serves as an Officer and on the Executive Board of Directors of the Special Needs Alliance, the national, non-profit, association of experienced special needs planning attorneys (membership is by invitation only); served on the Special Needs Law Section Steering Committee, of the National Academy of Elder Law Attorneys; was a Charter Member of the Academy of Special Needs Planners; and is a Faculty Member & Text Book Author for the Illinois Institute for Continuing Legal Education (IICLE).

Brian feels the tremendous responsibility of not only being the parent of a child with special needs, but also as an attorney with the knowledge and ability to assist others in special needs future planning needed to secure the future of children and adults with special needs.

Benjamin A. Rubin

Attorney Benjamin (Benji) Rubin, Brian's youngest son, Mitchell's "little/big" brother, graduated from the University of Illinois College of Law Magna Cum Laude, received his undergraduate degree from Northwestern University, and currently is pursuing his Graduate Law Degree, an LLM (Tax) at Northwestern University. Benji is a member of the Academy of Special Needs Planners, an affiliate member of the Special Needs Alliance, serves as the President of SIBS (Supporting Illinois Brothers and Sisters, the Illinois chapter of the national Sibling Leadership Network, is a member of the Board of Directors of The Arc of Illinois, is a member of the Clearbrook Associate Board, and also serves on the Leadership/Steering Committee of Encompass (formerly, the Supported Community Living Initiative), a joint venture of the Jewish Federation of Metropolitan Chicago, the Jewish Child and Family Services, and Keshet, regarding residential and day programming for adults with intellectual and developmental disabilities. Benji is also a Faculty Member for the Illinois Institute for Continuing Legal Education (IICLE). Having Mitchell as a brother profoundly shaped who Benji is today, and thus the type of law he chose to practice. His personal experiences as a sibling offer a unique perspective into the responsibilities that come with caring for a sibling with special needs. Now, as an adult, those sometimes present and future responsibilities he will share with his older sister regarding his brother's care are a concern that he shares with all brothers and sisters of individuals with special needs.

Laura Houston

I have had 18 years experience specializing in the birth to three population, as well as preschool and school-age children. I have spent numerous hours gaining additional continuing education with concentration in the areas of oral and/or verbal apraxia or dyspraxia, feeding and swallowing disorders, auditory processing and comprehension disorders, and aural rehabilitation. I have worked with children having a variety of delays, disorders and diagnoses including feeding and swallowing problems, receptive and expressive language delay or disorder, verbal apraxia, dysfluency or stuttering, central auditory processing disorder, word retrieval difficulties, Down Syndrome, Joubert Syn-

drome, Cleft lip and palate, Turner Syndrome, DiGeorge Syndrome, Asperger Syndrome, Autism and Pervasive Developmental Disorders, cochlear implant and hearing impaired, and others. I am certified to administer newborn feeding evaluations using the NOMAS® feeding scale. I have helped children successfully transition from tube feedings to oral feedings. In the last three years, I have added vibrational healing and use of therapeutic grade essential oils in my treatment with children and families. I recently completed coursework and earned certification as a Certified Pediatric Massage Therapist (CPMT) and Certified Infant Massage Instructor (CIMI). I currently am working for certification as a Board Recognized Specialist in Swallowing disorders awarded by the American Speech Language and Hearing Association.

I began a private practice as well in 1996, which afforded me the opportunity to continue working with children after they aged out of the birth to three program. I also obtained a credential to perform evaluations and treatment for the Illinois Early Intervention program. I joined with a group of therapists to establish Birth To Three Therapies in Grayslake, IL. Our business grew until we needed more space and more therapists. We moved to Lake Villa in 2004. In 2008 we changed our name to "The Therapy Tree Pediatric and Adult Wellness" to express our expanded focus on the mind-body connection and wellness for the entire family and/or caregiver unit.

Barb Thomas MEd, CSE, JSOC

Barb has 35 years of teaching experience, and for the past 16 has been Director of TECS Education Center (Thomas Educational Consulting Services). Under Barb's Direction, TECS offers Education with a focus on Social Skills Development and Healthy Sexuality in the following ways:

- One-on-One Education
- Small Group Instruction
- Educational Support Classes
- Travel / Vacation Classes
- In-Class Sessions for Schools (either here at TECS or at your location TBD by Director / Schedules)
- In-Service / Workshops for Teachers and Other Professionals

Also an Educational Consultant, Barb has a Masters Degree in Special Education, is Certified through the University of Louisville as a Juvenile Sex Offender Counselor and is Certified as a Sexuality Educator through AASECT, the American Association of Sex Educators, Counselors and Therapists.

Matthew Smith

Matthew has served five years in leadership with the JSRDF to provide support for families, influence research, and expand education on the JSRD. Matthew serves on the Growth & Development, Conference and Scientific Committees of the JSRDF, and he is a primary grant writer for the Foundation.

Matthew is Principal Investigator of the patient-powered research registry for JSRD called Joubert Syndrome Link to Information and Family Exchange (JS-LIFE). As PI, he oversees the privacy-centric gathering of participant-reported information from the Joubert syndrome community into the JS-LIFE registry with great care, helps to plan and execute research studies using reports from JS-LIFE, and disseminates outcomes to patient families, healthcare providers, and other researchers. Matthew's passion for this work arises from his desire to advance support and research for all individuals in the 'Joubert family' and with other rare conditions worldwide. He has a seven-year-old niece, Haley, affected by Joubert Syndrome.

By trade, Matthew is a Research Specialist and Laboratory Manager at the University of North Carolina. He studies the genetic and biochemical underpinnings of various forms of cancer, cancer metabolism, and epigenetics, and he has co-authored 12 peer-reviewed articles in highly respected journals. He has also been consultant to many genetic education initiatives in the United States.

Matthew is also a Professional Consultant for Genetic Alliance, an international health advocacy nonprofit of more than 10,000 health organizations, of which 1,200 are disease advocacy organizations. Genetic Alliance engages individuals, families, and communities to transform health.

Matthew has extensive involvement with other disease advocacy organi-

zations on shared aspects of their challenges and journeys. He serves on the Rare Disease Task Force of the Patient Centered Outcomes Research Network (PCORnet) and is Patient Representative for nearly 120,000 patients in the Community Engaged Network for All (CENA), a Patient-Powered Research Network (PPRN) of the PCORI initiative in the United States.

Jennifer Rhein

Jennifer Rhein is an Occupational Therapist at Aspire Kids, where she has worked for the past 3 ½ years with children and families in Early Intervention as well as children over the age of three in a clinic setting. She has experience working with children with a variety of diagnoses and a wide range of ages. Jennifer has worked with a child with Joubert Syndrome and her family for the past 2 years. She has a special interest in visual development and how it impacts a child's ability to interact with and learn from their environment.

Amanda Tulumalo

Amanda is a mom to Devin (16) and Jaden (11), who both have Joubert Syndrome. She has formerly served as a member of the Board of Directors for the Joubert Foundation. Amanda has been making music ever since she can remember! She began singing in children's choirs at church and at school, and then learned to play the piano and the clarinet. Amanda continued with her Clarinet studies in college, graduating from The College of New Jersey with a BS in Elementary Education and Music. She left her K-5 Music Teaching position to care for her children but has continued teaching private music lessons, which she has been doing for over 20 years. She has also been teaching Kids' MusicRound classes, as well as providing birthday party entertainment for the past 7 years. Amanda has played the clarinet with the Eastern Wind Symphony, sings and plays the piano on her church's worship team. Amanda feels that early introduction to regular music classes and basic music instruction through Kids' MusicRound has had a tremendous impact on Devin and Jaden's development. She cherishes the opportunity to nurture all children in the love of music and to develop their musical skills.

Bernardo Isacovici

Bernardo Isacovici is a yoga instructor, lawyer, lover of life, nature and all that is! When he is not spending time with his amazing wife and two kids, 2 and 4, he is either teaching one of his 10 weekly yoga classes, representing people's rights in criminal court, running or blogging. In 2010, while training for his second marathon, he stepped on the mat for the first time and never stepped off. Yoga saved his life, and he is happy to share his love and passion for the practice. Originally from Quito, Ecuador he now makes northern Illinois his Home. After having completed 9 marathons, 15 half marathons, completing yoga teacher training and losing 100 pounds in the journey, Bernardo has made it his life priority to share his passion for yoga, health, peace and love with those who need it the most.

Denise Mack

Denise Mack is a current member of the JSRDF Board of Directors, serving as the Secretary to the Board and Executive Team (in a non-voting role). She has held that position since 2011, and prior to the Minneapolis conference was a long-serving conference volunteer, managing vendor tables and sales tables. She also serves on the conference team for the Chicago 2015 "PossAbilities Project". Denise has a passion to support the Foundation from a service perspective as well as a financial contributor. Izzy's Birthday Wish is an annual campaign held in October where she invites friends and family to help her reach her \$1000 goal towards the Foundation. Most recently she kicked off a one-year, 5K-run fundraising program designed to raise awareness and money for the Joubert Syndrome Foundation by running in a different race (locally and/or nationally) each month. To date, she has run six races, and raised almost \$1,500 for the cause.

Denise is Manager of Membership Data and Demographics at the College of American Pathologists. She holds a Bachelors Degree in Public Health and Community Awareness from Northern Illinois University and her MBA in Healthcare Management from Keller Graduate School.

Her most important role is mother and wife. She is married to Stephen Mack, who has also dedicated himself to the JSRDF as President (2013-2015). They have three children, Isabella (10/JS), Christian (5), and Abigail (3). Together they enjoy traveling with their children and making wonderful memories every step of the way. Denise feels that while her journey with Joubert Syndrome has been challenging, the rewards are endless and have made her a better mother as well as wanting to do more for the Foundation.

Kortney Peagram

Dr. Kortney Peagram is the President and Owner of Bulldog Solution. Bulldog Solution is an advocate for providing schools with social skills training, as well as reducing bullying and cyberbullying. Bulldog Solution provides schools and communities with the resources, programs, curriculums, and result-oriented action plans to facilitate positive change. Dr. Peagram works with corporate organizations, schools, and camps to provide programs on leadership, conflict management, team building, group management, diversity, bullying and cyberbullying. Dr. Peagram has developed training programs on evidence based research and behavioral models to increase the overall training effectiveness.

Dr. Peagram consults, writes, facilitates, and presents on topics that focus on the emotional and behavioral development. She also focuses on leadership, communication, diversity, and digital citizenship. Dr. Peagram's passion is to reduce bullying and violence in schools, with the goal to start making a difference by using her skills as a facilitator. Dr. Peagram is also an Adjunct Professor at The Chicago School of Professional Psychology.

Due to her experience in the field, Dr. Peagram was featured as an expert on anti-bullying and program development on Fox 32 News Chicago in May 2014 and January of 2015. By blending her education, teaching, and passion, she delivers professional and effective programs, as well as projects to schools and communities.

Kim Walton

In the past 25+ years of working with a variety of special children and families who experience stress and trauma I have learned many valuable lessons – most important of which is that "the care of the child cannot exceed the care of the parent or caregiver". Our children are extra special and sometimes we wonder where we will get the stamina and the courage to take the next steps. As a parent, grandparent or friend of a special needs child, come listen, learn and share as we focus on resiliency, stress management and self-care. As the flight attendant says – "it is time to put the oxygen mask on your own self first."

Barry Katz

Barry R. Katz is a partner in the Business and Real Estate practice groups focusing in the areas of real estate and corporate law areas. He represents Fortune 500 companies, national lenders, developers, syndicators, condominium associations and management companies located both in the Chicago area and nationwide with respect to the acquisition, sale, financing, syndication, operation and management of real estate. Mr. Katz has been involved in numerous multimillion dollar transactions concerning the purchase and sale or financing of real estate, and has represented many lenders in multimillion dollar transactions secured by liens upon real estate, equipment and inventory. He has extensive experience representing landlords and tenants in the leasing of a wide variety of retail, commercial and industrial properties. Mr. Katz also has been involved in the merger and acquisition of businesses and the financing of such transactions, including the representation of lenders extending credit facilities and the representation of the buyer or seller of the business.

In the corporate law area, Mr. Katz represents companies ranging from small privately held corporations to publicly held Fortune 500 companies. He also represents several not for profit corporations.

Presenter Bios

Tom McGuire

Thomas McGuire is a partner at Arnstein & Lehr LLP and is also a Certified Public Accountant. His practice is concentrated in the areas of estate planning, probate and trust administration, and taxation. Mr. McGuire has substantial experience representing a broad range of individuals, financial institutions and tax-exempt organizations in connection with their estate planning and administration needs. He also is involved in obtaining and maintaining tax-exempt status for charitable organizations. Mr. McGuire regularly handles complex issues involving individual and fiduciary income tax, estate, gift and generation-skipping transfer taxes. A substantial portion of his practice is also devoted to business and succession planning for family-owned and other closely-held businesses. He is involved in handling a diverse range of matters in the Elder Law area, including guardianships; healthcare and property powers of attorney; living wills; revocable and irrevocable trusts; Medicaid, Medicare and social security issues; and the review of agreements with nursing homes, retirement homes, and "life care" facilities.

Sabrina J. Pippin, M.A.

Sabrina Pippin is a recent graduate with a Masters in Forensic Psychology from The Chicago School of Professional Psychology. Additionally, Ms. Pippin obtained a Forensic Science Certificate from Northwestern University. Previously, Ms. Pippin assisted with play therapy with families with disabled children for two years at Lekotek Center. Her research and professional interests include an attachment theory approach to mental health treatment, psychotherapy with individuals and families with disabilities, traumatic stress, and media psychology. Ms. Pippin will be featured as a co-author for an upcoming chapter regarding mental illness and BBC's Sherlock. In her leisure, Ms. Pippin enjoys volunteering at Lekotek with disabled children, performing physical exams for animals at the Anti-Cruelty Society, and enjoying time with her rescue dog, d'Artagnan.

Rebecca Cain

In 2003, I had the good fortune to partner with a group of very talented women to create Birth to Three and Beyond Pediatric Therapies. We all met while providing home health services through the Lake County Early Intervention Program. Our business quickly grew, and we expanded into the spaces next to our original office space. But even with our expansions, we felt that our plans and dreams for the services that we wanted to offer were much bigger than our space could accommodate. So in 2010, we re-branded to become The Therapy Tree, and bought a beautiful old building next to Cedar Lake in downtown Lake Villa, which is our current home. We created a wellness center on the upper floor, and opened our therapy clinic on the lower level.

Working at The Therapy Tree is not just a job for me; I consider it an ongoing journey of discovery, with endless possibilities. Each day brings new challenges and triumphs that I am privileged to share with the families that come to TTT in search of guidance, support, and healing. I look at each client as a unique individual, not a diagnosis or label that fits them into a box with a pre-determined destiny. I am a life-long student, and am always searching for the best approach to help my clients and their families reach their highest potential.

The field of OT is a perfect fit for me, as it allows me to treat clients with a truly holistic approach. OT's consider all aspects of a person's success and/or struggles across all of the environments and activities that are meaningful to them. This means considering the complete person, physically, emotionally, and psychologically. It also means considering the influence of all of the people that are an important part of my client's life. When I formulate a plan of care with a family, I make it a priority to consider the interdependence of all of the parts that contribute to the health, happiness, success, and well-being of the whole person.

I am always searching for innovative ways to expand my knowledge and skills. This has led me to reach beyond the traditional training of my profession, and incorporate modalities such as yoga, aromatherapy, craniosacral therapy, and lessons in mindfulness into my treatment plans. I have found that awareness of our emotions has a tremendous effect on our physical bodies, and awareness of our physical bodies has a tremendous impact on our emotional well-being. It is exciting to follow the near-daily discoveries in brain plasticity research, which are making the vague term, "Mind-Body Approach" a measurable, concrete, evidence-based and essential framework in our healthcare system.

I feel privileged and thankful each time I am entrusted with taking a role in the life of a child. It is a great responsibility, and I consider each child and their family to be my greatest teachers.

Thank You

Jackie and Jenni would like to extend a huge thank you to Denise Mack. When the Chicago location was announced, Denise stepped in as third Conference Co-Chair without even having to be asked! She has done an amazing job finding, coordinating and scheduling childcare volunteers, is fully stocking the child care room with toys, games, etc., has taken time out of her personal life to participate in site visits and conference planning meetings, she fully planned the medical and executive dinner, she has located and secured several of the Non-MD Presenters for this conference as well as the entertainment pieces of the schedule that we'll get to enjoy this week! Additionally, she is presenting two workshops of her own to share with us some tools that have worked well for Isabella! Thank you, Denise, for being simply amazing!

The Board of Directors and Executive Team

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\$500 - \$1000

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People In Business Care
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Robert & Christine Olson - Ice Bucket Challenge, in honor of Andrew Olson*

We would also like to extend an extra special thank you to those who donated their time, talents and/or made donations to make our conference a success:

*The Therapy Tree
The Kid's Music Go Round & Amanda Tulumalo
Melissa Fields
Kim Walton
Brian & Benjamin Rubin - Rubin Law
James Harlan - The Answers, Inc.
Tom McGuire - Arnstein & Lehr LLP
Kortney Peagram - Bulldog Solution
Jennifer Rhein - Aspire Kids Chicago
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Our Families



We honor all our families for the love, dedication, passion, and drive that we see every day in the stories that you share!

Families not pictured:

- | | | | | |
|-----------|------------|--------|---------|---------------|
| Ahmed | D'Angelo | Helwig | Kostos | Redmon-Bogusz |
| Altamura | DeSchepper | Hudson | Lamm | Richter |
| Amatriain | Douthwaite | Hyde | LaRocco | Starks |
| Banares | Dowdle | Keller | McGlynn | Veillette |
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