Wednesday July 19

2:30-3:15pm  Conference Orientation - Welcome to Hope Shines Bright!
Provides an overview of conference events and introduction of Conference Committee and Executive Team. Coronado Ballroom

3:15-4:30pm  Life with Joubert Syndrome – Panel Discussion
Hear from a group of teenage and adult individuals (and their families) affected by Joubert syndrome speak about how their milestones, challenges and just lives in general have been affected by growing up with Joubert syndrome. Coronado Ballroom

4:00-6:00pm  Treehouse Open House
Meet the Treehouse staff, get a sneak peek at the amazing activities your kids will be involved in and complete any open paperwork. Arizona II and III

4:30-6:00pm  Social Hour
Join in some pre-dinner fun. Be on the lookout for a special guest! Loggia and Paradise Lawn

6:00pm  DINNER Coronado Ballroom

After Dinner  Pool Gathering
Join us at the pool to relax and unwind after an exciting first day with music and swimming! Main Pool and McCormick Courtyard

Thursday July 20

7:30-8:30am  BREAKFAST Coronado II, III, IV, V and Paradise Lawn

Times TBD  Research Opportunities at the 2017 Conference - Learn and Enroll
Research studies are important because they contribute to knowledge and progress on diseases and disorders, in our case Joubert syndrome. It furthers our knowledge of JS and ultimately helps families. We have 3 groups conducting research at the conference this year. Participation is voluntary and there is no cost to participate. Look for specific times in your registration materials.

• Jennifer Dempsey - research coordinator at the University of Washington Hindbrain Malformation Research Program

• Meral Gunay-Aygun, MD, Pediatric Geneticist at the National Institutes of Health

• Nick Mahasuwan - Assistant Clinical Research Coordinator, Division of Craniofacial Anomalies, University of California, San Francisco

9:00-10:15am  Research Update: 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, comprehensive blood and urine tests to evaluate kidney and liver function and growth and re-analysis of brain MRI images. Coronado I

9:00-10:30am  Empowerment is the Name of the Game – Darrel Christenson, Ability 360
Attendees will hear about the importance of learning to speak up for yourself and getting your needs met in life. Don’t settle for sitting on the sidelines of your life – get in the game and participate. Feel the power of empowerment. JSRD individuals who would find this valuable are welcome to attend! Arizona I

10:15-  Research Update: JS and Kidney Disease
11:00am - Individualizing Treatments and the use of Precision Medicine - John Sayer, Professor of Renal Medicine
In his talk, Dr. Sayer will detail studies on human cells grown from urine from Joubert syndrome patients with CEP290 mutations in order to explain how this ciliopathy affects the kidneys. He will discuss the problems related to cilia and CEP290 and how personalized molecular treatments can be used to rescue this defect. He will also show how these new treatments can be given to mice as a prelude to treatment in patients with JS and kidney disease. Coronado I

10:30am-  Wanted: Cheerleaders! -
12:00pm  Darrel Christenson, Ability 360
This session is intended to encourage young kids and their families to learn how to be a cheerleader for yourself and for your children. Hear a first-hand story from someone who was born with multiple disabilities. Don’t give up – believe in yourself – be all you can be! JSRD individuals who would find this valuable are welcome to attend. Arizona I
**10:30am-1:15pm**

Calling ALL Grandparents...Let’s Talk! –
Kim Walton, MSN, APRN

Oh, the blessings...and the challenges of having a very special grandchild with JS. So, how do we support our families – our children, our grandchildren and take care of ourselves? Bring family pictures and let’s talk about what works and what we have learned on our grandparent journeys. Following the talk, join us for a grandparent luncheon. Yuma

**11:00am-12:00pm**

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. Coronado I

**12:00 - 1:00pm**

LUNCH Coronado II, III, IV, V and Paradise Park Lawn

**1:15-5:00pm**

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 AZ 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). Times may change and appointments can be scheduled at the registration desk beginning Wednesday. Rooms TBD

**1:15-2:15pm**

Ophthalmic Manifestations of Joubert Syndrome, Wadih Zein, MD

Dr. Zein’s presentation will cover the ocular (eye) manifestations of Joubert syndrome with an attempt at correlation with molecular findings. In addition, he will address recent advances in research for therapeutics of hereditary retinal degenerations. Coronado I

**2:00-3:00pm**

Anat Baniel Method (ABM)– Ed & Carla Dassie, New Connections Movement Works LLC

Learn about who Anat Baniel is, what the Anat Baniel Method is and the 9 essentials that Anat Baniel has created to define how her work can assist in treatment for adults and children with special needs. Arizona I

**2:15-3:00pm**

Research Update: Genetics of JS - Friedhelm Hildebrandt, MD

Joubert syndrome is a rare childhood disease that may involve many different organs, causing multiple health problems. By ‘whole exome sequencing’ a genetic cause of JS can be identified in over 50% of patients. Discovery of more than 50 genes, each of which may cause JS has allowed the first insights into disease mechanisms of JS. Coronado I

**3:00-4:00pm**

Making Sense of Genetic Testing for Joubert Syndrome, Melissa Parisi, MD, PhD

Dr. Parisi’s talk will describe what is known about the genetic basis of Joubert syndrome and the genes that cause Joubert syndrome and their general function. She will also discuss current genetic testing options for the condition, including when to pursue testing for individuals with Joubert syndrome and when to consider testing in relatives or family members of the person with Joubert syndrome. Coronado I

**3:00-5:00pm**

Small Group Discussions –

**3:00-4:00pm**

Kidney Concerns, Arizona I

Teens/Young Adults - Ideas for Independence. Board Room

Behavioral Concerns - Young Children. Arizona I

**4:00-5:00pm**

PT Brainstorming for Younger Children. Board Room

Behaviors, Puberty and Hormones in Older Kids. Arizona I

Orthodonture with Special Needs Individuals. Yuma

**6:00pm**

DINNER Coronado II, III, IV, V and Paradise Park Lawn

**7:00pm-9:00pm**

Poster Session / Meet and Greet with Cilia Researchers

The Federation of American Societies for Experimental Biology is also holding a conference locally, entitled “The Biology of Cilia and Flagella,” and we are delighted to invite them to our resort for an informal poster session and meet and greet. Please join us to learn more about current cilia research! McCormick Courtyard

**7:30pm**

Dads Night

Being the dad of a child with JS has its own unique challenges. Come and enjoy the night connecting with other dads that experience a similar journey while you kick back and relax with some yard games and music. No wives, no kids – just the guys. BYOB (Soda and water will be provided) Seclusions Suite
Friday July 21

6:45-7:15am  Yoga in the Lawn  Bring your own water and towel. Community Lawn

7:15-8:15am  BREAKFAST  Coronado II, III, IV, V  Paradise Park Lawn

8:45-9:45am  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 JSRD, discuss the effects of the kidney related problems, and explore what families can do to optimize kidney status. Coronado I

8:30-10:00am  Positive Behavior Support – Leslie Williams, Raising Special Kids  Learn how to reduce unwanted behavior and increase good behavior by using Positive Behavior Supports in your home and in the community. Understand the role you play in your child’s behavior, and how to work with the school when behaviors get in the way of learning. Arizona I

9:00-11:45am  MD Consults and 1:00-4:00pm  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 AZ 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). Times may change and appointments can be scheduled at the registration desk. Rooms TBD

9:45-10:45am  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. Coronado I

10:15-11:45am  Journey to Adulthood – Janna Murrell, Raising Special Kids  This workshop gives parents information on how to get ready for when their child goes through puberty and adolescence. Parents will learn about the physical, emotional and social changes that come during this time of their child’s life. Parents learn the skills and tools to help them through their child’s journey. Arizona I

10:45-11:45am  Reproductive Options and Joubert Syndrome, Ian Glass, MD  Dr. Glass will discuss the range of reproductive options for families of children with Joubert syndrome including adoption, sperm/egg donor, preimplantation genetic diagnosis, and prenatal diagnosis by genetic testing and imaging. Coronado I

11:45am-12:45pm  LUNCH  Coronado II, III, IV, V and Paradise Park Lawn

1:00-1:45pm  Research Update: UW: JS Genes, Cells and Resulting Outcome - Dan Doherty, MD, PhD and Julie Van De Weghes, PhD  Genetic testing results and clinical features (including mortality) in >500 families affected by JS will be presented. Work by Univ. of Washington and others has shown that mutations in >35 genes cause JS, and we are now determining what cellular defects are shared across patient cell lines with different genetic causes. This work will identify the central mechanism(s) underlying JS and pave the way for designing precision treatments of the future. Coronado I

1:00-2:30pm  IEP Training – Leslie Williams, Raising Special Kids  Have a better understanding of the IEP process. You will learn who is a member of your child’s IEP team, how to track your child’s progress, and what to do if the team does not agree. Arizona I

2:00-2:45pm  Research Update: UW Mortality Matters – Dan Doherty, MD, PhD and Julie Van De Weghes, PhD  While a hard topic to process, learn about recent research on JS and Mortality. Coronado I

2:45-4:15pm  Resilient Relationships – Janna Murrell, Raising Special Kids  This workshop is designed to help couples make and keep a healthy relationship while raising a child with special needs. Learn what resilience means and how to decide what comes first. Understand how women and men handle situations differently. Learn about ways to handle unexpected changes and what might cause those changes. Arizona I

3:00-4:00pm  Benefits of Osteopathic Manipulation (Cranial Sacral Massage) for Patients with Joubert Syndrome – Dr. Greg Heller  A case in which Osteopathic Manipulation was used to successfully treat a patient with Joubert syndrome will be presented as well as a physiological explanation will be given for how patients with JS can benefit from Osteopathic Manipulation (Cranial Sacral). Coronado I

4:30-5:15pm  JSRDF General Meeting  Learn important information about the JSRD Foundation, our mission and our vision for the future. Meet your executive team and board members who tirelessly serve our foundation. Arizona I

5:15-6:00pm  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) Coronado I

7:30pm  Moms Night Out  Moms work hard, frequently with little praise or thanks! This night is about you! Come relax with other moms (sorry ladies – no kids, please) BYOB (Soda and water will be provided) Seclusions Suite
Saturday July 22

7:30pm Adults with JS Gathering
Too old for the teen activities but want to connect with other JS adults? This is for you!
No topic – just a social gathering. Bar Six 40 area

2:00-3:00pm Hippotherapy and Equine Assisted Activities – Saebra Pipoly, Horses Help
Ms. Pipoly will discuss EAAT programs and what that means, the difference between Equine Assisted programs and who is suitable for each type of program. She will also discuss the benefits of EAAT programs, safety concerns, how to find a program in your area and what to look for in an EAAT provider and finally the typical industry standards for billing and whether or not programs are covered by insurance. Arizona I

3:00-4:00pm Fundraising 101 – Jenni Swenson, JSRDF Fundraising Committee/Conference Co-Chair
New this conference, this session will cover official foundation logo use guidelines and why it’s important to keep the JSRDF brand consistent. We’ll also cover the difference between personal vs. JSRDF fundraising opportunities. We’ll also be rolling out two incredible fundraising opportunities that will begin in October! Learn how to earn FREE Conference fees! You don’t want to miss this! Coronado I

Sunday July 23

9:30am Decompression Session
Brainstorm with conference organizers with ideas for 2019 and learn ways to get involved. Bring your own coffee!!
Patio Outside Bar Six 40

3:00pm-4:00pm Building Your Child’s Voice – Denise Mack, JSRDF Secretary & Conference Co-Chair
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. Arizona I
**Teens: JS and Unaffected Individuals Ages 13-18+**

**Wednesday**

2:30-3:30pm  
**Teens - Getting to Know you Activity - Mummy Game!**  
“Wrap up” some fun with the affected and unaffected teens at the conference. *Seclusions Suite*

**Thursday**

9:00-11:00am  
**Teens – Games, Games, Games, plus a Special (Secret!) Project**  
Board games, card games, dice games and more! *Seclusions Suite*

11:00am-12:00pm  
**Teens – Photo Scavenger Hunt**  
Our teens will set off on an adventure taking pictures of fun things around the hotel. They will bringing back digital photos of places and things. In doing this, they will capture good memories and also have some fun working together as a team. Volunteers will walk the kids to Coronado to meet you for lunch. *Seclusions Suite*

1:15-2:45pm  
**Teen Time – Photo Scavenger Hunt, Continued**  
Part 2 of this fun adventure. *Seclusions Suite*

2:45-5:00pm  
**Teen Time – Pool Time – (must provide own supervision)**  
Cool off after the photo scavenger hunt with your new friends – *This is an unsupervised activity - parent/adult supervision is required!* *Main Pool*

**Friday**

8:30-11:45am  
**Teens – Art Project**  
This morning we will be working on a cool art project. Wear grubby clothes! Volunteers will walk the kids to Coronado to meet you for lunch. *Seclusions Suite*

1:00-3:00pm  
**Teens and Young Adult Topics**  
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. *Seclusions Suite*

3:00-5:00pm  
**Teens – Movie and Snack Time!!**  
Movie selection TBD but it may be PG13 – *Seclusions Suite*

**Saturday**

9:00-11:00am  
**Teens – Memory Book!**  
You’ve made new friends – and now you will have a memory book to take home as a memento. *Seclusions Suite*

11:00am-12:00pm  
**Yoga for Affected Individuals – Sandy Wendelin**  
Yoga is becoming more popular every day – not just for adults! Perhaps more important, for special needs children, it can be used as an alternative therapy. Yoga can help increase concentration, flexibility, and strength. It can also develop coordination and reduce stress and anxiety. Bring a caregiver to maximize participation. *Arizona I*

1:00-4:00pm  
**Teens – Afternoon of Fun**  
Freeze Dance, ice cream social and karaoke - start practicing your dance moves for the closing dinner! *Seclusions Suite*
**Siblings: Ages 5-12**

### Wednesday

**2:30-3:30pm**  
**Welcome to The Sibling Retreat! – Led by Melissa Fields & Kim Walton**  
Open house and crafts – the retreat room will be open for families to drop by and meet Kim and Melissa. Kids can stay for craft time if they wish.  
*Pima*

**3:30-5:00pm**  
**The Sibling Retreat Kickoff**  
Join us as we all get to know each other with games and ice breakers as well as one of our favorite craft projects!  
*Pima*

### Thursday

**9:00-10:30am**  
**The Sibling Retreat – Siblings and Challenges – It Will Be a Blast!**  
We will have facilitated conversations with the sibs about how they are affected by JS. Conversation to include: Tell us about your sibling. What does JS mean to you? What do you love about your brother or sister? If you could change one thing about him or her; what would it be and why?  
*Pima*

**10:30am-12:00 pm**  
**The Sibling Retreat – Game Time**  
Game time! Kick back and relax with some cards and board games. Coloring and craft time also available.  
*Pima*

**1:15-2:00pm**  
**The Sibling Retreat – Don’t STRESS me out!**  
Kim will be leading an informative and interactive discussion with the kids about stress and how it affects kids. Kids will have the opportunity to identify stressors in their lives.  
*Pima*

**2:00-3:00pm**  
**The Sibling Retreat – YOGA**  
A great way to calm yourself!  
*Pima*

**3:00-5:00pm**  
**The Sibling Retreat – Stress Management for Kids**  
We will talk about and PRACTICE ways to reduce STRESSORS. We will be making stress balls, having a dance party, and enjoying some art therapy with Carrots, Cookies and Canvas…don’t worry. Healthy snack and cookies included!  
*Pima*

### Friday

**8:30-11:45am**  
**The Sibling Retreat – Helpers and Heroes**  
This morning will be packed with fun as we look at how we can be Helpers and Heroes at home with our JS siblings. We will also talk to sibs about how we can best take care of ourselves. Don’t forget about games and crafts!  
*Pima*

**1:00-3:00pm**  
**The Sibling Retreat – Crafts & Music**  
*Pima*

**3:00-5:30pm**  
**The Sibling Retreat – Movie & Popcorn**  
*Pima*

### Saturday

**9:00am-12:00 pm**  
**The Sibling Retreat – Practice Makes Perfect!**  
The kids have a special surprise they will be preparing for today! Shhh! We can’t ruin the surprise!  
*Pima*

**1:00-4:00pm**  
**The Sibling Retreat – More Practice**  
More practice for that special something. Kids will also be making a favorite take home!  
*Pima*