

## **2011 Joubert Syndrome Biennial Scientific Conference summary**

The NIH- and ORDR-funded Joubert Syndrome Biennial Conference: Advancing Translational Ciliopathy Research, Enhancing Clinical Care took place at the DoubleTree Hotel Conference Center in Orlando, FL July 13-14, 2011. Investigators in Genetics, Nephrology, Developmental Medicine, Ophthalmology, Neurology, Radiology, Genetic Counseling, Basic Neuroscience and Gastroenterology, as well as patients with JS and their families attended the meeting.

The first day featured a keynote talk summarizing the History of JS by Eugen Boltshauser, MD, (Children's Hospital, Zürich) who has contributed to the understanding of JS for more than 30 years. Additional talks included JS Neuroimaging (Andrea Poretti, MD, Johns Hopkins University), Genotype-Phenotype Correlations in JS (Dan Doherty, MD/PhD, University of Washington), JS Kidney Disease (Meral Gunay-Aygun, MD, Johns Hopkins and NHGRI), JS Liver Disease (Theo Hiller, MD, NIDDK), Biology of JS Nervous System Abnormalities (Russell Ferland, PhD, Albany Medical College), Respiratory and Sleep Abnormalities in JS (Maida Chen, MD, University of Washington) and A New Gene for JS (Ji Eun Lee, PhD, UC San Diego). In particular, Dr. Chen's talk spurred a lot of interest in sleep disordered breathing and circadian rhythm abnormalities. Attendees included all investigators, representatives of the JSRDF Board, at least one primary pediatrician and many families of children with JS.

During meetings over lunch and before dinner, Dr. Doherty led structured discussions to update the healthcare recommendations for patients with JS based on new information generated since the original recommendations made in 2004. Natural history data, access to care, predictive value of monitoring, cost, and the availability of treatments to modify the course of the disease were all taken into account. These discussions highlighted major gaps in knowledge of JS that should be the target of future studies including: mortality (causes and ages of death), sleep disordered breathing, and behavioral/mental health disorders. Information about these issues will directly impact counseling regarding prognosis, as well as the details of when and how to monitor for progressive kidney, liver and retinal complications. By the end of the discussion, the group had agreed on a framework for summarizing and evaluating proposed recommendations. This document is in draft stage and will be distributed to participants later this year for additional feedback and refinement. Once complete, Dr. Doherty will prepare a manuscript summarizing the healthcare recommendations for submission to the American Journal of Medical Genetics in early 2012. The paper will be posted on the JSRDF website ([www.joubertfoundation.com](http://www.joubertfoundation.com)).

The second day of the meeting was dominated by focus groups bringing together families and investigators to identify areas of mutual interest for future research. In addition, an informal survey of challenges experienced by patients and families revealed a variety of concerns, most prominently behavioral issues. Talks featured JS Eye Disease (Avery Weiss, MD, University of Washington) and Biology of JS-related Kidney Disease (Friedhelm Hildebrandt, MD, University of Michigan). Structured discussions focused on outlining a research agenda for the future and planning the next JS Scientific meeting. Input from families stressed the impact of behavioral issues on quality of life for people with JS and their families. Strong support was voiced for exploring the possibility of a Rare Diseases Clinical Research Network proposal to study JS or ciliopathies in general. The research agenda will be reviewed by the participants and then distributed to meeting attendees, NICHD, NIDDK, NIE, NINDS, ORDR and posted on the JSRDF website. Consensus was achieved for scheduling the 2013 JS Scientific Meeting in conjunction with a major medical research meeting to allow for much greater participation by investigators and trainees. Grant funding will be budgeted to ensure family representation at future scientific meetings. The next meeting will focus on research toward therapies that will benefit patients with JS.