10th Biennial 11q Research and Resource Conference - June 26-30th, 2016, San Diego, California

By Paul Grossfeld, MD

In July, 1995 during my first month of pediatric cardiology fellowship at the University of California San Diego (UCSD), I took care of a patient with Jacobsen Syndrome (JS), a rare chromosomal disorder caused by deletions in the long arm of chromosome 11. My interest in this syndrome was heightened by the possibility of identifying a novel gene for left-sided obstructive lesions, including HLHS (Hypoplastic Left Heart Syndrome), which we subsequently discovered occurs at a higher frequency in JS than for any other known human genetic syndrome. Fast forward to June, 2016: serving as the Chief Medical Advisor for the past 15 years for the *11q Research and Resource Group* (www.11qusa.org/), a support group for patients and their families with JS, I worked with the group's President, Linzee Carroll (mother of two children with JS), to organize the *10th Biennial International Conference* held in San Diego June 26-30th. Fifty-one families and 15 invited faculty from nine countries representing four continents, gathered for the four day conference. Just over half of the families were attending for the first time.

While my initial interest was and continues to be in the genetic basis of HLHS and other congenital heart defects that occur in JS, through the encouragement of the families I have established collaborations that have focused on many of the other major causes of morbidity and mortality in JS including: a congenital platelet disorder, varying degrees of intellectual disability, behavioral problems including autism and ADHD (Attention Deficit Hyperactivity Disorder), and life-threatening immunodeficiencies.

Day one of the conference began with the keynote speaker, Ms. Peyton Goddard (www.peytongoddard.com), who depicted her struggles as a severely autistic and non-verbal child. She described how using facilitated communication allowed her to overcome her limitations, as well as the discrimination, abuses and biases of others to accomplish, among her many remarkable achievements, graduating valedictorian from her college. The remainder of the day included talks on several of the recently identified major medical issues including immunodeficiency and brain hemorrhages. The afternoon featured a riveting talk by Reverend Ryan Sey, the head chaplain at Rady Children's Hospital of San Diego, on coping with having a child with a rare genetic disorder. The opening evening dinner featured every child receiving a "rare bear," graciously provided by Dr. Chris Waters from her non-profit organization, Rare Science (www.rarescience.org). That was followed by a talent show displaying the remarkable talents of several of the JS children and young adults.

Tuesday's sessions were highlighted by several talks on neurologic and behavioral problems, including a hands-on demonstration by chiropractor Dr. Tommy John III on how improving movement can help overcome some of the physical limitations imposed by the syndrome. The afternoon session included a lively panel discussion on potential gene-based therapies for intellectual disability and autism, with the hopes of launching at least one pilot clinical trial by the

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end of 2016. The day culminated by attending the San Diego Padres game that evening, with tickets graciously donated for all of the families by the Padres.

Wednesday began with a comprehensive overview of endocrine problems that can occur in JS, followed by a number of breakout sessions led mostly by family members of affected children. Perhaps the highlight and most moving session of the conference was the Wednesday afternoon panel discussion consisting of adults with JS, as they fielded questions about what it is like to live with JS. The remainder of the afternoon consisted of more breakout sessions, including another gripping discussion led by Reverend Sey entitled, "Shut Up About Your Perfect Child."

Thursday, the final day of the conference, was highlighted by an interactive music therapy session organized by Dr. Barbara Reuer and three of her music therapists (www.resoundingjoyinc.org). The emphasis was on participation, and people of all ages with JS found themselves engaged and thoroughly enjoying the moment. The conference concluded with a picnic in which families had one final opportunity to say good-bye to each other until the next conference, tentatively scheduled for the summer of 2018.

Numerous on-site assessments took place during the conference, including: cognitive and behavioral, three-dimensional craniofacial imaging, physical exams, and one-on-one family meetings with me. These assessments have been instrumental for acquiring new knowledge, ultimately leading to improved therapies for people with JS.

Financial support was provided, in part, by the Jeffrey Modell Foundation (jmfworld.com), Vivint Gives Back (www.vivint.com/company/gives-back), and the UCSD Department of Pediatrics (www.pediatrics.ucsd.edu). The Sulpizio family kindly donated Magali wine for all of the speakers. In addition, many families did their own fundraising to afford the cost of attending the conference, and thanks to some generous anonymous donors, limited scholarships were made available to families in financial need. In one case the father of a child from England, who sold his guitar in order to afford the costs of bringing his family to the conference, was given a brand new handmade guitar graciously provided by Pepe Romero Jr., of the famous Romeros family of classical guitarists.

Ironically, it was my interest in the broken hearts of people with JS that initially got me involved with this remarkable group of families. To that end, we have made some exciting strides in identifying the gene that causes congenital heart defects in JS, a second genetic locus that can prevent heart defects in mice lacking the gene that causes heart defects, and most recently an animal model for HLHS using the commonly studied frog *Xenopus*. Despite these very gratifying successes, I never anticipated that 20 years after seeing my first JS patient, it would be the strength and beauty of their hearts that could teach me so much about the human condition, and why I remain involved with this unique and extraordinary group of families.

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Paul Grossfeld, MD
Professor of Pediatrics
Division of Cardiology
UCSD School of Medicine
Tel: 858-966-5855; Fax: 858-571-7903
www.littleheartsbighopes.org
pgrossfeld@ucsd.edu