Dr. Paul Grossfeld, Associate Clinical Professor in the Division of Cardiology of the Department of Pediatrics at the University of California, San Diego, is the preeminent U.S. expert on Jacobsen Syndrome and 11q chromosomal disorders. He collaborates with other genetic-specialist colleagues world-wide to conduct state-of-the-art research. In turn, we gain invaluable insight into our children’s unique development, behaviors, and needs.

Our research on Jacobsen syndrome continues to focus on two major areas of investigation: cardiac and cognitive/behavioral. Several years ago we identified a gene in 11q, ETS-1, that is the likely cause of at least a subset of the congenital heart defects that occur in JS. Recently, we have determined that deletion of this gene in mice causes an even wider spectrum of congenital heart defects than what we originally observed, paralleling those observed in JS children.

Interestingly, the presence and type of heart defect is dependent on the strain of mouse studied, implying that there are specific genetic factors that can influence whether or not there is a congenital heart defect. It is likely that these same factors determine whether your 11q child has a heart defect. We are currently embarking on a strategy that could help us to identify these factors.

In order to understand the mechanism of how loss of ETS-1 causes heart defects, we have identified at least two populations of cells that are critical for normal heart development: The cardiac neural crest and the endocardium. Recently we have determined that loss of ETS-1 impairs normal migration of cardiac neural crest cells, an essential process for normal heart development. We now have some preliminary evidence indicating that ETS-1 is essential in the...Continued on page 2
earlier stages of heart development, prior to the time cardiac neural crest cells migrate to the developing heart. One possibility is that the loss of ETS-1 endothelial and neural crest cells in combination is required to cause heart defects. We are currently testing that hypothesis.

In addition to congenital heart defects, we have identified several genes in chromosome 11 that cause cognitive and behavioral problems. One of these genes, Neurogranin, is essential for memory and learning. Interestingly, patients with Alzheimer’s disease have decreased levels of this same protein in their brain. Towards that end, we are hoping to embark on a drug development strategy aimed at boosting the level of this protein in patients with memory and learning disorders, including potentially those with Alzheimer’s disease and other related memory and learning disorders.

Most recently, we have determined that half of children with Jacobsen syndrome have autism. We have identified a new gene in chromosome 11 that likely causes autism in these children, and we hope that by understanding the function of this gene in normal brain development, better therapies can be developed.

We are currently utilizing very recently developed technologies to reprogram cells from skin fibroblasts obtained from 22 11q- patients at the 2012 conference into developing heart and brain cells. These studies will allow us to assess the molecular and cellular defects underlying the heart and brain problems in 11q- patients, hopefully leading to greater insights into the pathogenesis of these problems in 11q- patients. We are very excited about this and are optimistic about what we hope to learn.

Finally, as you may recall from the 2012 conference, there are multiple lines of recent evidence suggesting that a subset of 11q- patients have varying degrees of immunodeficiency. In collaboration with the European Chromosome 11 Support Group, we are hoping to initiate a study to better understand these issues, and hopefully develop better treatment strategies as they relate to infections and immune function.

None of this work would be possible without the participation of your 11q- children. We are also grateful for the fundraising efforts on the part of several 11q- families. Without your support, none of this research can continue!
WHAT'S NEW WITH 11Q?

Plans are underway and it promises to be a great week!

A more detailed schedule of speakers and sessions will be sent out as the event gets closer, but the general schedule is as follows.

**Monday, June 23**

Early bird sessions will begin around 11:00am and continue through the early afternoon.

A sibling workshop for children ages 6 and up will be held in the morning.

Official registration and check in will begin Monday afternoon followed by a dinner and keynote speaker in the ballroom Monday evening. Adults and children are invited to all sessions on Monday.

**Tuesday-Thursday, June 24-26**

Conference sessions will be from 8:30am to 5pm with a break for lunch each day. These days are packed with speakers, both medical and non-medical, as well as several parent led break-out sessions where you will have the opportunity to share ideas with other parents, grandparents, and families working through similar issues.

**Wednesday evening** there will be an adult reception out on the patio of the hotel. Appetizers will be served and there will also be a cash bar available. It is a great time to relax and socialize with other parents and grandparents.

**Friday, June 27**

On Friday afternoon, the conference culminates with the 11q family picnic at the park adjacent to hotel which is always one of the highlights of the conference. You don't want to miss it!

Fees for conference attendance are $50 per adult, $25 per child, and as always, 11q children are free.

Childcare will be offered during sessions on Tuesday, Wednesday, and Thursday for a small fee. The childcare takes place in the room adjacent to the conference sessions so you can check on your child frequently.

Please email Linzee Carroll at 11qusa@gmail.com if you have any questions. We hope that you will be able to join us for an amazing week of education, sharing, support and friendship! We look forward to seeing you in June!!

**Courtyard Marriott San Diego Airport/Liberty Station**

2592 Laning, Road, San Diego, CA 92106
619-221-1900

Group room rates of $159/night are available, but limited. Click here to make a reservation or mention that you are with the 11q group when making your reservation.

The hotel offers complimentary shuttle service to and from the airport as well as free parking.
Thank you!
First of all, I would like to extend a heartfelt thank you to those families who have organized and hosted fundraising events in support of 11q Research and Resource Group as well as for Dr. Grossfeld’s research (see sidebar). Your dedication, generosity, time and effort are at the core of what makes our 11q family strong, welcoming, and supportive.

Some of the events hosted this year include online auctions, silent auctions, art auctions, yard sales, Causes.com and similar fundraising web pages, painting parties, and selling 11q stickers. Families have raised $100s to $100,000s of dollars for 11q R&R and Dr. Grossfeld’s research, and it all helps so much! Whether the funds help provide a scholarship for a family to attend conference or allow Dr. Grossfeld to conduct patient studies or purchase lab supplies, every dollar makes a difference!

- $50 pays for the preparation of two DNA samples.
- $100 pays for one kilogram of lab chemicals.
- $150 pays for one night of a family’s stay in the 11q conference hotel
- $250 pays for a skin biopsy of one 11q child.
- $1000 pays for the preparation of a skin cell culture.
- $5000 pays for the mapping of a patient’s entire genome.

The research done by Dr. Grossfeld and his team help not only those with JS but also larger populations affected by heart defects, Autism, Alzheimer’s, ADHD and other behavior and cognitive disorders.

FUNDRAISING FAMILIES
- Kurt & Stephanie Christensen (Sydney)
- Tim & Cindy Pellett (Vanessa)
- Dean & Joy Robertson (Kailani)
- Todd & Lori Sorbo
- Charity & Mike Stanfield (Zion)
- Geoff & Krista Walsworth (Catelin)
- Gaby Wantah
- Craig Thomas & Rebecca Alson-Milkman (Elliot)
- The Williamson Family (Grace)

UPCOMING EVENTS
- $11 for 11q (see Page 5)—Kickoff Nov 1
- Help Little Hearts Heal—Nov 9, Los Angeles, CA
  Organized and hosted by Craig Thomas & Rebecca Alson-Milkman with friends Carter & Denise Bays, Pamela Fryman and Alan Grossbard, Robyn Murgio & Chris Miller, 20th Century Fox Television and the cast of How I Met Your Mother. For more information about this event, email Rebecca at Jacobsen.fund@gmail.com

11q stickers designed by Gaby Wantah. Stay tuned for more information on ordering.
$11 for 11q beginning 11/1

Written by Heather Wainwright

With November being the 11th month and the holiday season in full swing, we thought it would be a great time to launch an $11 for 11q: Support Research and Awareness campaign to raise funds for Dr. Grossfeld’s research.

The motivation
Dr. Grossfeld needs funds to keep his lab open so he can continue his groundbreaking research.

The venue
Each of our personal Facebook pages

The plea
To all of our Facebook friends and family, please donate $11 (or more) in honor of my great 11q family member and to support Dr. Grossfeld’s research.

How it will work
There will be a copy-able “status” on the 11q R&R Facebook page on November 1 to announce the campaign. The status update will include a direct link to the University of California San Diego Foundation (the home organization that processes funds for Dr. Grossfeld’s research). My hope is that most, if not all, of our 330-member-strong group will then copy and paste the status update onto our personal Facebook pages on November 1—and as often throughout the month as you think is reasonable. Our friends and family can then simply click on the link in the status; they will be directed to the UCSD website with just a click of a mouse, where they will be able to make their $11, $111, or $1,111 (or any other amount) tax-deductible donations and receive a receipt/confirmation.

To sustain this campaign throughout the month, I will post additional “Did you know…?” posts that reveal a factoid about Dr. Grossfeld’s research on 11q disorders and/or Jacobsen’s syndrome, some of the recent news he’s shared with us regarding the understanding and prevention of heart defects and possible medicinal therapies that might help our loved ones in addition to the larger populations of those contending with ADHD, autism, and other cognitive/memory disorders. The factoid will be followed with a reminder about the $11 for 11q campaign and a link to the donation site. Again, the idea is that each “status” can be reposted on our personal Facebook pages to encourage our friends and family to donate.

Corporate Match
Many businesses and corporations match employees’ donations as part of their community outreach. 11q Research and Resource Group is a 501(c)(3) charitable organization, so it qualifies as a beneficiary for such programs. For more information, contact your company’s matching gifts office. For information about designating 11q R&R Group as your recipient organization, contact Linzee Carroll at 11qusa@gmail.com.

THE ONLY WAY FOR US TO CONTINUE TO MAKE PROGRESS TO HELP YOUR CHILDREN IS FOR US TO WORK TOGETHER, AND TO HOPEFULLY FIND BETTER WAYS TO OBTAIN ADDITIONAL FUNDING FOR THIS VERY EXPENSIVE AND TIME-CONSUMING RESEARCH.
~DR. PAUL GROSSFELD, AUGUST 2013
The need continues

Written by Heather Wainwright

Dr. Grossfeld’s research is long-term, cutting-edge, and therefore expensive.

In fact, he needs to secure $2 million to move forward with his research on the genetic components of heart defects and cognitive/behavioral disorders. Unfortunately, the funding that keeps Dr. Grossfeld’s lab open and allows him to serve as the 11q Research and Resource Group advisor as well as sponsor our biannual conference has become harder and harder to secure through federal grants.

Due to government cutbacks, funding agencies have more proposals to consider than ever before, making competition for award dollars extremely time-consuming and not often fruitful for those who attempt to navigate the cumbersome process. Although Dr. Grossfeld spends as much time as he can—time which takes him out of his lab—identifying and applying for funding, both he and the 11q Research and Resource Group need our help!

We hope that the Help Little Hearts Heal event and the $11 for 11q campaign are successful in raising funds for Dr. Grossfeld’s research. We have more fundraising and awareness plans underway that we will be sharing with you in the coming months, including a promotional video about the 11q R & R Group, ways for your family to raise funds to attend the 2014 conference, collaboration with other rare disease organizations, and more plans to support Dr. Paul and the 11q R & R Group. Please stay tuned to our group’s facebook page and to your email for updates.

Where you can find us...

11q Research and Resource Group- www.11qusa.org
11q Research and Resource Group Facebook Page- www.facebook.com/groups/25120356262/
Jacobsen Syndrome Facebook Page- www.facebook.com/groups/jacobsensyndrome/
$11 for 11q: Support Dr. Paul Grossfeld’s Research Facebook Page- www.facebook.com/SupportDrGrossfeld

SHARE YOUR FUNDRAISING IDEAS WITH US...

If you or someone you know is sponsoring a fundraising/awareness event to support the 11q Research and Resource Group, Dr. Grossfeld’s research, or your family’s trip to the 2014 conference, please share that information with Heather Wainwright. If you have hosted an event already and you’re not listed in this newsletter, we apologize. Please send us your information so we can include it in future issues. We would love to hear your ideas and let other families know that you contributed. Please don’t hesitate to contact Heather Wainwright at 2boys4joy@gmail.com, 662-769-4897, or via Facebook if you have any questions or ideas to share.
2014 Conference Registration Form

June 23-27, 2014

Courtyard Marriott San Diego Airport/Liberty Station

11q Child Name and Age  ________________________________________________

Diagnosis (Please include deletion/duplication size) ______________________________

Parent(s) or Guardian _______________________________________________________

Address  ________________________________________________________________

Phone ________________________________________________________________

Email Address __________________________________________________________

Please list all the people attending in your party (please include ages of all children)

________________________________________________________________________

________________________________________________________________________

Registration Fees

$50 per adult $25 per child (3 and under are free)

All 11q children are free.

# of Adults x $50 =  # of children (over age 3) x $25=

# of 11q children x $0=

Total enclosed =  ______________

Please mail this form along with your registration fees (please make checks payable to the 11q Research and Resource Group) to-

11q Research and Resource Group
c/o Rich Syretz
12004 Trona Court
Manassas, VA 20112
The first 11q family conference was held in Dallas, Texas in 1996. Over 20 families attended from the US plus one family from the Netherlands. The rest, as they say, is history.

Since then, the organization has continued to grow, and we now have hundreds of members from all over the world. In 2005, the 11q Research and Resource Group officially became a non-profit 501(c)(3) organization. The organization now hosts a conference every two years for families of children or adults with all 11q disorders, where families have the opportunity to learn from medical experts, participate in ongoing research, and meet other families dealing with similar issues.

The mission of this group is to provide support and organizational efforts for the parents, as well as friends and family, of all children with 11th chromosome abnormalities, including deletions (monosomy), duplications (trisomy) and translocations.

Through networking, cooperation with the research community, and the sharing of information, we will be better able to meet the challenges brought to us as parents, families and friends of 11q children.

The 11q Research and Resource Group has no paid employees and all of the board members are volunteers. 100 percent of the funds raised and donations made to the group will be allocated toward supporting research and funding of conferences that provide families of affected children with the ability to learn more about the syndromes from doctors and professionals from around the world.

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11qusa.org
EIN: 04-3840155

Click here to visit us on Facebook!

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11q Research and Resource Group Board Members

President- Linzee Carroll
Vice President- Jessica Fekete
Secretary- Melanie Johnson
Treasurer- Rich Syretz
Chief Medical Advisor- Dr. Paul Grossfeld
Fundraising Co-Chairs- Heather Wainwright and Sarah Williamson
General Board Member- Lori Richardson
Newsletter- Krista Walsworth

Thank you to those who volunteer for particular committees throughout the year!

We are currently looking for volunteers for the conference planning and fundraising committees. Please email Linzee at 11qusa@gmail.com if you are interested.

If you would like your child featured in the 11q Kid Spotlight or if there are any topics you want discussed in future newsletters, please send your thoughts to kckwalsworth@gmail.com.