

Denver Conference Overview

Personal review of the KS&A 2008 Mile High Family Conference on X and Y chromosome variations (August 2008), written by Mrs. Mina Avery

Let me start by saying that if you have never attended a conference be forewarned. This was my husband, Don's, second conference and my third. We joked that first timers should be given the "gut-wrencher's" discount as it is an information overload much of which is somewhat difficult to accept. I don't know how it will be for most but for Don and I the first conference was really painful.

Don said the conference was much less distressing to him this time. For me I found myself being able to use the information as a check list, "O.T. evaluation, done: Special Needs Trust established, do that when we up-date our will: Apply for SSI, do when she is 14... and so on."

So here goes, hope this gives you a glimpse of my perspective of the conference.

Friday was clinics.

We were traveling without the girls so we did not attend any clinics. We heard that they were very valuable.

Saturday:

Kick off with Dr. Tartaglia. Nicole explained the development of the eXtraordinary Kids Clinic at the Children's Hospital in Denver. She emphasized that the clinic was developed in response to parent requests for a place to go where professionals knew about this population. She said, "This is your clinic. You come see me when you want." I loved that as I was thinking of it as the research clinic that would call us when they wanted. She said she would meet with our daughter until she was 30 or beyond. I want to schedule Rowena to see her next year as she is doing really well right now. I think if Nicole sees her during the great times she will have a better foundation for helping her through the difficult times.

Nancy Raitano Lee presented the preliminary finding from the NIH Trisomy X brain study. As you know our girls tend to have reduced white matter. Here is the break down they are seeing on the MRI's.

Brain Area	Function	Trisomy presentation
Temporal Lobe	Social, Lang, emotion, memory	Reduced sized in both gray and white matter
Basal Ganglia	Organization and Prioritizing	Slightly smaller
Occipital Lobe	Visual center, orientation, color, size, shape	White matter reduced
Ventricles cavities in the middle of the brain	Contain spinal fluid and have been noted as being enlarged in some disorders	Enlarged
Frontal Lobe	Impulsivity, organization, expressive language, personality and emotions	Reduced white matter
Parietal Lobe	Sensory function, spatial processing	Reduced white matter
Cortical Layer	Outer layer, measured for thickness	Thicker in the orbital frontal cortex and anterior cingulated area. Thinner in the fusiform gyres, lateral temporal, and lateral inferior frontal area.

The white matter affects the speed of transmission. So, I spoke to a father after the presentation and he said, "O.K., so now how do we help them? "

Of course I had no answer but I did think about an adult Klinefelter's man I had talked to at breakfast. He had said, "You need to get trisomy X listed on the disabilities act as it will be easier for your daughter to get services then."

If we can have proof positive physical data, won't that make recognition as a disability easier?

Nicole addressed the trisomy group separately. (At the Kansas City conference the trisomy sessions found five moms in a room. It was valuable but imagine how thrilled I was to find two dozen or more people in the trisomy sessions. Hooray!) Nicole did a survey of individuals and found that anxiety is a big concern for our girls. She also found that aggression is very low. She felt the lack of aggression issues may be a sign of passivity. She noted that the girls suffer from fatigue which has been attributed to low muscle tone. She noted that she will continue to look in to the source of the fatigue as she doesn't see this level of fatigue in other patients with low muscle tone.

She noted that the girls tend to be “quietly inattentive” and may be under treated for attention concerns. She said that she has had some improvements with anxiety and attention with certain medications.

She noted that the girls have abdominal pains, constipation, are prone to allergies, pneumonia, dental problems, lazy eye and kidney disorders. They are also at risk of heart malformations. She discussed tremors and seizures. She also mentioned that she and Lennie Wilson are currently reviewing literature on trisomy X and hope to complete a review by December. This may include recommendations for treatments. She displayed a slide showing overlapping conditions with trisomy X, bi-polar, autism, seizures, attention concerns, anxiety, etc. She warned that the reduced bone age common in our girls can delay puberty. She recommended that we not let them be delayed. She thought they should seek care if they are later than (I think) 13 or 14. She felt keyboarding could help with school as handwriting is slow, laborious and frustrating for many of the girls.

Jay Lucker did a great presentation about auditory processing, how it presents, what to look for, and how to accommodate the girls and boys. Jay noted that once the Auditory processing deficit is diagnosed you must find out what system is accounting for the problems and treat the problems by addressing that symptom. He noted that the XXY's suffer from Auditory Phonemic extraction, auditory memory problems, auditory temporal extraction, general auditory attention, organization and sequencing and auditory integration concerns. He then discussed how to address each area. He has a book out called “*Don't You Get It?*” I plan to order it directly from Jay today as I love autographed copies. (drj@ncapd.org)

The support group meeting was amazing. We had all introduced ourselves to each other during breaks, meals and sessions but it was fun to have a time to compare experiences.

Sunday

Rich Boada explained the process of reading development, how it goes wrong and what to do and not do to remediate. He cautioned against quick fixes and encouraged long term programs.

Karen Riley walked us through RTI to help us understand how the schools can help our children. She encouraged parents to work as partners with the school in helping them understand the diagnosis and the best ways to help the children. Her presentation was powerful.

Laura Pickler gave step by step instructions on how to help your child get in the driver's seat of their medical management. She has a booklet that she created

to help parents keep medical paper work organized and ready for the child to use as they transition in to adulthood. You can get a copy at www.cdphe.state.co.us/ps/hcp/transition/workbook.pdf. I plan to get this.

The awards ceremony is amazing. Dr. Bruce Bender worked with Dr. Robinson and Mary Linden on the original Denver Study. He gave a compassionate speech about how bad the information about these conditions had been before their study and how passionate they all were to give the world a more complete picture. He was amazing.

Nicole Tartaglia and Susan Howell hosted a panel discussion with two young adult's with extra Xs. The trisomy girl was amazing. She made me laugh and cry and celebrate the potential that my daughter has. The young Klinefelter's man was articulate and clear sighted. He was amazing. If you buy the conference DVD only to see this session it will be worth your money.

Rene Beauregard led a session called "Planning is Power." She explained that all parents have a Plan A for children. (Give them roots and give them wings) She said some of our children will have that, but we need a Plan B for when those things don't come easily. She explained how and why to get continuous medical coverage, the necessity of services and supports for adulthood, the tricks to navigating the system, and much more. She handed out a workbook to use as a check list for getting ready for adulthood.

I did not attend any of the sessions for the Klinefelter's or XYY boys. I was told they were very good.

I have already ordered my copy of the DVD's. I would encourage you to think about finding a couple of people to go in with on a set of DVD's. You can watch them, share specific sessions with O.T.'s or teachers and then send them on to the other people you are sharing with.

I attended these sessions but, as you can imagine, my brain only processed a fraction of what was being said. The DVD sets give me a great reference when a professional wants more information.

The conferences are amazing as they build such a sense of community and hope. It also gives you great resources for when you need a little help.

I hope I will meet all of you someday at upcoming events. We have much to face but we also have much to celebrate.

Mina