ISSUE NO. 91 WINTER 2021

Chromosome Disorder Outreach Newsletter

Letter from the President

Dear Friends of CDO,

CDO's 30-year anniversary is coming up! In February 2022, CDO will celebrate 30 years of providing support and information to those affected by the rarest of chromosome disorders.

For those who may not know our history - Chromosome Disorder Outreach was started in 1992 by 7 parents of children with chromosome deletions. CDO grew slowly in its first few years - only a few hundred families found the organization. But in 1996, when the website launched, membership took off. Today - just since 2007 - we have added almost 8,000 members. And we now offer support for all rare chromosome disorders, as well as many gene mutation disorders.

CDO's primary mission has always been to make sure no one ever feels alone. And through our expansive networking programs and large Facebook community, we hope this has been accomplished. Additionally, over the years, CDO's medical advisors have worked tirelessly to provide information to families who otherwise may have had little to go on. Our library contains well over 10,000 publications, with 40+ new research articles added monthly.

We hope to continue CDO's mission well into the future. And you can help too. CDO always need volunteers in many areas; won't you join us? Please email info@chromodisorder.org if you have time to donate. We would love to hear from you. Warmest wishes to all for a very happy holiday season!

-Linda, CDO President

Phone: (561) 395-4252 Email: info@chromodisorder.org

The 2022 CDO Calendar
Available for purchase at zazzle.com

We Do It For You
CDO: 2021 Great Nonprofit

New Books
Three recommended books for reading

The Nogueria Family – Infinite Gratitude

This is the history of the Nogueira family. We are from Brazil, more precisely from the city of Porto Velho in the State of Rondônia, located in the Brazilian Amazon region.

Our sweet Ravena Nogueira came from a high risk pregnancy and complications in the umbilical cord, which meant that she was born prematurely at 33 weeks.

Weighing 1 kilo and 180 grams, Ravena came into the world by an emergency Cesarean, as it was necessary to interrupt the pregnancy due to the mother's life at risk due to pre-eclampsia. Ravena was rushed to the neonatal unit, where she stayed for 180 days.

In her first weeks of life, Ravena was diagnosed with congenital heart disease: Inter-Atrial Communication (IAC) and Interventricular Communication (IVC). It took 6 months of an intense struggle against time to keep Ravena alive through medications and cardiac monitoring, until she gained enough weight for heart surgery but without success. Every day Ravena lost even more weight, even weighing less than at birth.

The doctors had no idea why, even though she was fed by enteral tube 24 hours without interruption, Ravena was just losing weight and gaining little.

Until she was 6 months old, Ravena's little heart reached the limit of what was bearable, and already in a myocardial effusion, her body could no longer support adjustments in the dosage of medications for the heart. Her only chance was surgery, we rushed Ravenna to the Children and Maternity Hospital of São José do Rio Preto in São Paulo, more than 2,055.17 km away from our house, as there are no cardiac hospitals in the northern region of Brazil.

After nearly 9 hours of surgery, on March 2, 2020 Ravena left the operating room and went straight to the post-surgical unit, where she spent 15 days in a coma due to post-surgical complications due to acquired pulmonary sepsis.

In the preoperative period of cardiac surgery, in the midst of a battery of tests, Ravena was diagnosed by the geneticist physician through a karyotype test, with Syndrome 49, XXXXX (Pentasomy X).

In the beginning, even though cardiac surgery was a success, we were very scared due to the discovery of this rare syndrome and that doctors and geneticists in Brazil do not know anything about it. So, with the diagnosis in hand, we searched all over the internet and around the world, sending out emails to genetic and research centers around the world.

We found CDO, who welcomed us and helped with all possible information about Syndrome 49, XXXXX (Pentasomy X), in addition to connecting us with other families from other parts of the world, having daughters with the same syndrome.

We are infinitely grateful to CDO for their help since then, as with this valuable information, we went to the doctors of our sweet Ravena Nogueira, and with that, they may have a way to go to help us take care of her.

Our sweet Raven is now 2 years and 1 month old, undergoes therapies to help her development and is the greatest joy of our lives!

We would love to help other families with children like ours. We even have an INSTAGRAM profile, @meumundinhoravenistico, which we use to help other families with their atypical motherhood.

- Richárlisson Nogueira & Alexia Nogueira

LASTEST RESEARCH

For more information see….

chromodisorder.org

Important new research articles are selected monthly by Dr. Iosif Lurie, MD PhD and summarized for publication on our website.

Clinical and genomic characterization of 8p cytogenomic disorders.

Congenital heart defects were the most common internal defect reported in 2/3 of invdupdel(8p) patients and in almost all patients with "proximal" or "proximal + distal" deletions, but very rare in "distal" deletion group. Constipation and gastro-esophageal reflux were frequent in all sub-groups. Loss of the GATA4 gene is considered the main factor causing heart defect in patients with 8p abnormalities. However heart defects in invdupdel(8p) patients were relatively minor compared with individuals with "pure" deletions involving GATA4.

Characterization of the phenotype for Phelan McDermid syndrome.

Patients with larger deletions of chromosome 17p have more severe delays in speech and motor development. Aggressive tendencies were reported in ~25% of studied patients. More than half had self-injurious tendencies. Non-psychoneurological symptoms included hypotonia, kyphosis, gastro-esophageal reflux, frequent infections of ear, respiratory or urinary systems. Seizures and epileptic-like episodes were reported in some of the examined persons. Relatively mild kidney anomalies including asymmetric or duplex kidney were reported in 18 patients. The wide variability of manifestations indicates that some persons with deletions of this area have very mild manifestations and escape being diagnosed.

Interstitial duplication of 20q11.22q13.11.

Interstitial duplications of chromosome 20q are very rare. Less than 10 patients have been reported in the literature. The reported patients with similar duplications of 20q showed that developmental delay, dysmorphism and shortness of hands were common manifestations for most of these persons.

CDO is a Great Non-Profit… Again!

And all thanks to our wonderful members. Here are a few things people are saying…

"When we were finally given the diagnosis before my daughter's birth of a rare chromosome abnormality, we immediately started searching the internet for information. This was the first information we found. Just knowing we're not alone was a big support for us. It's great being able to read about other parents challenges, updates, and support. Thank you."

"Hearing that your child has any disorder can be difficult, but when you hear that they don't know what this means for your child can be scary. This agency not only found information our doctors did not, but helped me find others with similar situations. Thankful we found them!"

https://greatnonprofits.org/org/chromosome-disorder-outreach-inc

Ask the Doctor

CDO's geneticists answer hundreds of questions from members and website visitors each year. Below is a sampling of a few recent inquiries. Some details have been changed to preserve privacy.

To learn more about our program, view archived questions and answers or submit an inquiry, visit chromodisorder.org/ask-the-doctor or email askthedr@chromosdisorder.org

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Q: Unbalanced translocation of chromosomes Xp and 11q - although translocations are very unique to the individual, would you please provide what information you can?

A: The patient has a deletion for the almost whole short arm of X-chromosome and trisomy for a large ~62 Mb segment of the long arm of chromosome 11. This is an unbalanced translocation X;11. Normally a trisomy for so large a segment of 11q is either lethal or leads to numerous significant birth defects.

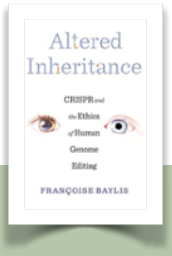
The situation in this patient is very different, because trisomy 11q is associated with a deletion of the X-chromosome. Females have 2 X-chromosomes, and only one of these two remain active in each cell (and the other one is inactivated). Usually inactivation is random: ~50% of cells have an active maternal X-chromosome and ~50% have an active paternal X. In girls with large structural defects of one X-chromosome this abnormal X-chromosome is predominantly inactivated. If such an abnormal X-chromosome carries an additional piece of another autosome this inactivation spreads also to this autosomal segment. So in this girl in the vast majority of cells additional material of the long arm of chromosome 11 remains inactive and that explains her relatively mild (for trisomy for such large segment 11q) manifestations. CDO's geneticist, Dr. Iosif Lurie was able to locate only one article and two abstracts in the literature dating from 1981 about patients with similar duplications 11q in association with X;11 translocations. These materials have been added to the CDO Library. Of course, no one can predict the real consequences of such a chromosome abnormality, but generally an association with a deletion of the X-chromosome is the only option for a relatively favorable prognosis for such a patient.

Many unbalanced translocations X;11 are results of a balanced t(X;11) in mothers. Genetic counseling is recommended.

The CDO Library contains information documenting this.For more information on any article please email info@chromodisorder.org

Iosif Lurie, M.D., Ph.D. Medical Geneticist CDO Medical Advisor

BOOK CORNER



Altered Inheritance: CRISPR and the Ethics of Human Genome Editing by Françoise Baylis

Haben by Haben Girma

RARE CHROMOSOME DISORDER AWARENESS WEEK

Save the Date… June 2022

If you are interested in serving on a committee to increase awareness during this special week, please email info@chromodisorder.org

SNEAK PEAK: BOOK CHAPTER FROM RAISING JESS: A STOY OF HOPE BY VICKIE RUBIN

On April 11, 1982, Easter Sunday, I was twenty-four years old and had the chicken pox. And I was about to deliver my first child. For a week, I had complained to my doctor about a rash, and for a week he had said, "The baby is settling. Do not worry" or "Just put some lotion on. It must be dry skin." I was naïve, pregnant, unaware, and truly wanted to believe it was nothing.

And then my water broke. We arrived at the local hospital, and as I told a few doctors about my rash, each dismissed it as unimportant, until finally an astute nurse (probably an experienced mother) said to me that my rash looked like chicken pox. My mind went into overdrive, racing with anxious thoughts. How could this be? I was in labor and I was twenty-four years old. Didn't I already have all the childhood diseases? They called in other doctors to confirm the suspected diagnosis, and since nobody was sure, a physician ordered a biopsy of my rash to confirm that I indeed had chicken pox. I clearly remember a labor pain and a scalpel removing a pox happening simultaneously. Welcome to motherhood.

Once the chicken pox diagnosis was confirmed, the local hospital had trouble dealing with the ridiculous news. My husband, Mitch, had the misfortune of a new resident telling him that it was unlikely the baby and I would survive the birth and delivery. He actually said that. Perhaps in the late 1970s and early'80s, medical schools didn't include the class on how to talk to patients and families. Perhaps the resident was so overwhelmed with chicken pox, labor, and a frantic mom-to-be that he just blurted out what he was thinking rather than considering what he should say. I wonder if he is still in medicine and if he ever thinks of this incident from Easter Sunday in 1982.

Mitch contacted my parents, who were in Florida and were unable to fly to Buffalo on such short notice on Easter Sunday. They would have to wait until Monday. My in-laws were at the hospital with us, praying for a positive outcome.

The attending doctor decided that I should be moved to Buffalo Children's Hospital (now called Oishei Children's Hospital of Buffalo, Kaleida Health). One would assume that I would be transported by ambulance, but Mitch drove the twenty-five-minute route with me laboring beside him. My labor was at the beginning stages, and since it was my first child, I believe the doctors at the first hospital thought I was not in danger of giving birth on the highway. In today's litigious world, I think they would not have taken that chance.

(continued)

Meanwhile, my father-in-law was sent on a mission to get what we were told was the last vial of the varicella-zoster vaccine (for chicken pox) to administer to the baby as soon as she was born. The problem was that on Easter, the only pharmacy that had this drug was Roswell Park Cancer Hospital, and they were closing within the hour. We were told it was the only place to get it in the city. Again, it's hard to believe that in 1982 the hospital would send my father-in-law to pick up crucial medication. What if something happened to his car, or he dropped the medication and it shattered, or he encountered any number of random acts of delay? Buffalo was not a rural town in the 1980s, but the way this day was unfolding, one would think we lived in Mayberry (a small town in a 1960s sitcom, for you millennials).

Mitch and I arrived safely at Buffalo Children's Hospital, and to our surprise, the medical staff was not alarmed about my circumstances. They admitted me into an isolation room so as not to spread chicken pox in the labor and delivery unit or among fellow newborns. My father-in-law was able to get the medicine while I labored for eight more uncomfortable hours at the hospital.

Jessica was born on April 11, 1982, at 8:15 p.m. and our friend Jackie promptly dubbed her our Esther Bunny, her nickname for a Jewish baby girl born on Easter. Jessica did not have chicken pox and was immediately administered the drug my father-in-law had fetched from the cancer hospital. Jess was taken to the neonatal intensive care unit (NICU) for monitoring, and since this was 1982, I was unable to see my firstborn child. There was no cell phone, Face Time, texting, Facebook, Instagram—nothing. The 1982 version of instant photos was a Polaroid camera. Mitch bought a camera and showered me with photos of our newborn; however, I was desperate to hold Jessica in person. My mom-hormones were screaming to hold my newborn while the physicians reminded me that I was still contagious.

I was released from the hospital within thirty-six hours, but was unable to see our baby or bring her home until a pediatrician in the community declared that I was chicken pox free. In hindsight, I question why I wasn't referred to an infectious disease specialist at the hospital or perhaps to a dermatologist, as it was skin related. Fortunately, I found a pediatrician who cleared me and sent me on my way home to motherhood.

Jessica weighed less than five pounds when she was born, although she was not delivered early. In retrospect, it was obvious that Jess had unusual facial features and other anomalies, but as first-time parents, we were somewhat clueless, and the doctors did not say anything to us. We had a baby nurse who had been in the profession for over thirty years and who assisted us for the first month. She said Jess was the tiniest baby she had cared for, but she didn't remark on any other differences.

In Long Island, New York where I was raised, baby nurses were common-place when a mom came home with her newborn. My mom was not a hands-on parent, but she was extremely generous with funds. Mom insisted that I have a baby nurse for the first month and that she would pay. I didn't have to think twice about that offer; however, this was not a common practice in the suburbs of Buffalo where we lived. I searched high and low for a baby nurse and was repeatedly asked if I had a disability or some problem that prevented me from caring for my newborn. I replied that unless being spoiled by my parents is a disability, I was okay. We selected a nurse after an out-of-town friend referred her to us.

As the months passed, the pediatrician continued to assure us that Jess was developing normally, and I continued to question all her delays. Interestingly, in reexamination, an inexperienced mom like me knew more about her child than an experienced physician.

I recently came across a letter written by my father that applauds Mitch's and my ability to manage the difficult birth and circumstances. He concluded in his letter that this was a brief hiccup of a start and that he looked forward to seeing Jessica develop into a wonderful daughter whom we could be proud of as he was of me. Jessica has developed into a wonderful daughter and we have much pride, but her story is very different from mine.

Little did we know that chicken pox was the least of our worries.

CHROMOSOME DISORDER OUTREACH

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info@chromodisorder.org

CDO is a 501C3 charitable organization.

(Florida Registration #CH11200)

Chromosome Disorder Outreach

You Are Not Alone.