YOU ARE NOT ALONE

ISSUE NO. 90 SUMMER 2021

Chromosome Disorder Outreach Newsletter

Letter from the President

We at CDO hope everyone is enjoying a great summer and staying safe. As the world gradually transitions to a 'new' normal, we are starting to reconnect with friends, family, and activities that were put on hold over the last 18 months. And that has been wonderful! Most of us will never look at things we took for granted in the same way again.

But many of our CDO members have children that are more vulnerable to illness and infection, and their 'new' normal may look a bit different than others. So those in this group may wish to maintain their high level of vigilance. With the rapid expansion of vaccine availability and the continuation of other safety precautions, there is cautious optimism for a better future. As parents and family members of the medically fragile, we have increased responsibility. But by keeping up the great work, we can collectively beat the pandemic and move ahead.

Again, everyone at CDO wishes you all a fantastic summer!

Warmest wishes, Linda Sorg, CDO President

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

The 2021 CDO Calendar
Still available for purchase

We Do It For You

CDO is a Great Nonprofit

New Media/ Books

What to read and watch

Living with a Rare Disorder – A Family's Story

Every morning there is a small celebration in our home: we go into Jaimi's room, peek over his bedside, and if he is already awake, we get a million dollar smile and a high five. Oh, that high five…. the arm has to be pulled all the way from underneath the blankets to come up, so that is a real gift.

It can happen too that we are woken up by the sound of giggles; Jaimi usually takes life easy.

He just turned 21, which felt like such a milestone! I had envisioned a real party, a reunion with all the caretakers who mean so much to us. We all know how that went during these strange times. We were grateful, though, to celebrate it with a few dear friends, and grandpa and grandma visiting. One of the things I learned so well over the years is to cherish the small things… and always have a well-stocked pantry (so I can?) to make some good food, soup or a cake.

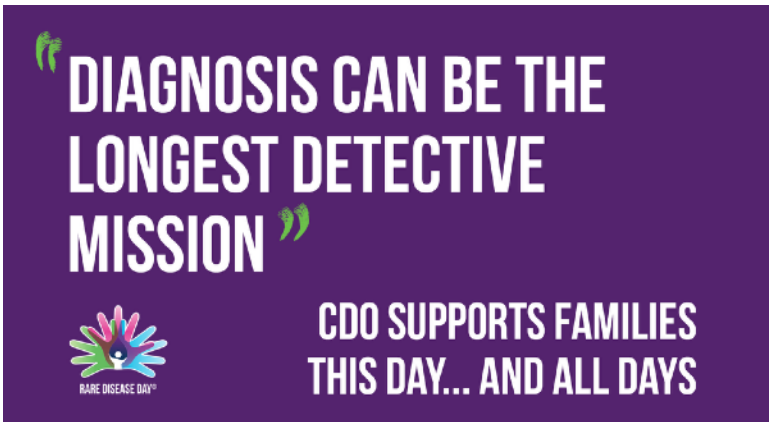
Jaimi is home full-time. His life is mostly about being, not doing. We took him out of school at 14 years old because it cost him so much of his energy. School was a lot about repeating what they think might be good to learn, and he was not much into that. He learns more by experiencing and has little interest in doing things with his hands. He got plenty of practice in school and would come home smelling of the shaving cream that they used to stimulate tactile senses. I realized when he was 11 or so, that he wasn't into exploration with his hands anyway.

Even though it is often an uneventful life, it has worked out well for us. We are enjoying each others' company, just by sharing. Going for a long walk, folding clothes, working in the garden: life, adventures, chores… it is all just more fun when he is there, sharing it with us.

When I am cooking, he is at a fantastic standing table (Easystand). I highly recommend this and feel so grateful that we got this through our health insurance.

We had a lot of snow in February, and it was really cold, but we managed to bundle him up and go for walks everyday. After a few walks he really started to enjoy this new white world; he loved it very much, as you can see in the pictures. He has a wheelchair with big mountain bike wheels and a detachable bigger wheel in the front, and this has made it into a real all-terrain wheelchair. I am not sure if he is bothered by it being a bit boring sometimes. I see he is overall happier than most people I know, including me.

He/we managed, and he became very healthy. Home-cooked and home-grown food, often organic, herbal medicine, being outside a lot… and also learning that our personal wellness is just as important for his wellness. From being sick often until the age of seven, knocked out by a cold for two weeks, even going to the hospital because his oxygen was low, now he rarely even gets a cold! We think we all might have had COVID last year in February, without too many problems and this winter no colds or sickness at all. No medicine either, except for



some homeopathic drops. He drinks liquids by spoon, always needing time to adjust to this, so the first sip is pretty small.

We have a good life but it's not always an easy one. He is starting to have less energy and likes to stay in bed until midday. He is very content until around 8 PM, when he wants to go back to bed. His blood work is perfect. We sense that the body just has a hard time with it all, not being optimally formed, and showing us that the strong years are over.

Even though our days are often not so eventful, we have managed to travel to the Caribbean several times in the past, and we visit Sweden as much as we can. We always look for ways to stretch what is possible. Jaimi has stayed quite small and light (5 ft and about 80 lbs) so it is quite doable to go on a bit rougher terrain. We go into the forest, for walks or to pick berries and mushrooms in Sweden. He gets parked in a nice spot, and we circle around him. You get a big smile upon returning with a bucket full of blueberries.

So what was initially such a heartbreak, his birth and discovering all the "disabilities" and then the severe delays, has turned into an often wonderful life of learning and discovering many things I would never have experienced otherwise. Jaimi has a special wisdom; he doesn't do jealousy, guilt, dishonesty and all that, and doesn't connect to people who do… how smart! He takes life as it comes, and I feel very grateful being his mother. We see him as a very special gift in our life.

Jaimi (46 XY del (13)(q31)) and his family live in the Netherlands. They have been members of CDO for over 20 years.

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.

Ring chromosome 20 syndrome: genetics, clinical characteristics, and overlapping phenotypes

Ring chromosome 20 syndrome – the genetics of inheritance and the main symptom: seizures. Ring autosomes are relatively rare, but ring chromosome 20 may be the most common type of all autosomal rings.

7p12.1 microdeletions the IKZF1 gene and cancer predisposition

Broad phenotypic spectrum of germ line 7p12.1 microdeletions encompassing the IKZF1 gene includes predisposition to acute lymphoblastic leukemia. Proximal deletions of the short arm of chromosome 7 are uncommon. The authors report two Polish patients with deletions 7p13p12.1 (in a girl) and 7p11.2p12.2 (in a boy).

Cardiac evaluation of patients with 22q11.2 duplication syndrome

Cardiac symptoms in patients with 22q11.2 duplication syndrome. Patients with isolated duplications of 22q11.2 segment show a very wide spectrum of manifestations from severely affected to practically asymptomatic persons. Some patients with duplications of 22q11.2 may have congenital heart defects (CHD).

Feingold syndrome

Feingold syndrome is the association of learning disability, microcephaly, facial dysmorphism, short stature, brachymesophalangy (shortness of the middle phalanges of the fingers) and other digital anomalies. There are two types of Feingold syndrome: many patients with FS type 1 also have gastrointestinal defects, mainly esophageal or duodenal atresia.

Two new reported cases of 16q22.3q23.3 duplication syndrome

Two new reported cases of 16q22.3q23.3 duplication syndrome and intrafamilial variability. The authors report a 15-year-old male with developmental delay, attention deficit disorder, autism, dysmorphic features (midface hypoplasia, prognathism), absence seizures, leg discrepancy (left leg being shorter than the right one) leading to scoliosis, and polymicrogyria.

Great Non-Profit Campaign

CDO is a great non-profit! We have received this award many years running. Please support our efforts again this year. It only takes a few minutes. If we have been able to help, please consider reviewing our efforts on greatnonprofits.org - doing so raises awareness of rare chromosome disorders, helps others to find us and allows CDO to make an even bigger impact on our community. Thank you! 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

Q: I have 2 children who were diagnosed with a 2q13 deletion. I understand that 2q13 deletions are rare but can you tell me just how rare they are? Do you know how many people in the US (or in the world) are known to have this genetic condition? Also what are the most likely health conditions associated with 2q13 deletions and what is the probability of having more than one sibling with it ?

A: Deletions of chromosome 2q13 are relatively common. There are no official statistics regarding this condition, but at least 220-230 persons with all kinds of deletions 2q13 have been reported so far. Of course, only a small percentage of patients are being published.

Clinical manifestations may vary very significantly - from normal to patients with multiple birth defects and numerous neurodevelopmental problems. The attached articles may offer a better understanding of the possible health issues caused by this deletion.

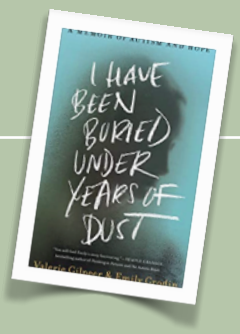
If a family has 2 children with del 2q13 it means that one of the parents is a carrier of the same deletion. Cytogenetic testing of both parents is necessary not only to explain the occurrence of this deletion in two sibs, but also for the planning of further offspring. If the family has a healthy child (or healthy children) they also have to be tested, because (if they are asymptomatic carriers) they may have their own children with the same deletion.

CDO has many articles in our library discussing 2q13 deletions. For more information on any article please email info@chromodisorder.org

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

MEDIA CORNER

I HAVE BEEN BURIED UNDER YEARS OF DUST, A MEMOIR OF AUTISM AND HOPE



Valerie Gilpeer

A memoir by a mother and her autistic daughter… long unable to communicate, a miraculous breakthrough reveals a young woman with a rich and creative interior life.

THE REASON I JUMP… NOW STREAMING ON NETFLIX

The diverse experiences and emotions of five young people with autism in an illuminating documentary.

Did You Know?

Each year, CDO's networking program helps hundreds of individuals and families connect with others who share similar disorders or medical concerns. CDO Co-President, Darlene Axelsson, explains more about exactly how much goes into preparing each networking list.

"Many of you have probably requested networking (a list of members with a similar disorder) at some point, usually upon joining, but some renewing members also request it. I totally enjoy connecting members with other members. It is a process and can take anywhere from 20-30 minutes to complete one networking list (some members have more than one disorder, so those may take longer).

Often when someone joins CDO, I need to email them for the exact chromosomal results. For example, a member may submit on their application that their child has a 15q deletion. In order to see if CDO has any similar members, I need to know specifically which part of 15q is deleted. Once I have these results, I search our database for similar members which involves plugging in given values of the disorder (the majority are deletions) into the database, and I am given a list of 'matches.' I then copy and paste those into a welcoming (or renewing) letter, password-protect it, and email it to the member.

I have been doing networking for CDO since 1999; our database has grown over the years! For years, there were times when we had no matches for a member. Now it's unusual to not find at least one match for a member."

This month, Julie Hofer is stepping down from the CDO Librarian position she has held since 2002. During these many years, Julie interacted with almost 300 families and catalogued and sorted CDO's many thousands of publications. We are truly grateful for the time Julie donated to help families and all those affected in any way by a rare chromosome disorder diagnosis. Thank you, Julie! We wish you all the very best in the future!

Chromosome Disorder Outreach

You Are Not Alone.

CHROMOSOME DISORDER OUTREACH

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CDO is a 501C3 charitable organization.

(Florida Registration #CH11200)

SUPPORT CDO

DONATE NOW

If you can, please help support CDO during this difficult time.

CDO is a Great Nonprofit