ISSUE NO. 86 SPRING 2020

Chromosome Disorder Outreach Newsletter

Letter from the President

CDO Supports Families... Always.

CDO is here for you. These are stressful and uncertain times - made even more so for those with a chronic or rare disorder. Everything we relied on changed overnight, and we accept new ways of doing the most basic things.

As we learn to live with new terms like 'social distancing,' we wanted to let you all know that CDO is still here, and here for you. You can reach us any time via email or phone. We are also working to create more Facebook group live events to help with the isolation that many may feel even more than before. We will keep you posted. Join our private group at www.facebook.com/groups/chromosomedisorderoutrreach/

Remember you can email or call, whichever is best for you. Please be safe, stay healthy and let us know if there's anything we can do to help.

-Linda Sorg

Phone: *(561) 395-4252*

Email: *info@chromodisorder.org*

CDO Calendar 2020
Our art calendar is still available at zazzle.com

Remeber CDO When You Shop
Choose CDO for your Amazon Smile donation

RCDA Week
Coming soon...
June14 - 19, 2020

Every storm in your life is followed by a rainbow…

Stay connected, stay with family, stay safe.

See resources at cdc.gov... search "stress and coping"

INFORMATION CORNER

Transition services are intended to prepare students to move from the world of school to the world of adulthood. Here are some resources which might prove helpful if you have a high school age child. You also may be able to locate state specific resource pages online as well.

The Organization for Autism Research has developed a comprehensive guide to resources. Although the guide is specific to autism spectrum disorders, it includes general information helpful to anyone parenting a young adult child. Visit OAR to learn more.

The **Center for Children with Special Needs** page**,** sponsored by Seattle Children's Hospital and Washington State Department of Health, contains numerous links to helpful resources such as a transition toolkit, guardianship and supplemental needs trust info and more.

Florida Department of Education has created The Family Guide to Secondary Transition Planning for Students with Disabilities.

Center for Parent Information and Resources: https://www.parentcenterhub.org/ transitionadult/ General information as well as links to educational info, work connections, IDEA, independent living and more. Spanish information also available.

Related reading: Wall Street Journal, Jan 26-27 2020: Lost Labor Now Found - Disabled Americans are joining the workforce at the highest rate in years.

And Ticket to Work is a program providing social security disability beneficiaries access to free employment services.

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.

Q: We received a prenatal diagnosis of mosaic trisomy 20? Can you provide more information?

A: There are many published articles on mosaic trisomy 20. In several publications the authors report the birth of healthy children or children with minor (mostly cutaneous) abnormalities. However, several authors also report serious birth defects. Unfortunately, it is virtually impossible to predict the outcome of a pregnancy with mosaic trisomy 20. Development of the fetus should be monitored by ultrasonography. *CDO has many articles available on this condition, contact info@chromodisorder.org for more details.

Q: Can you help me understand the life expectancy of a child with distal 18q deletions?

A: Life expectancy for patients with distal deletions of 18q is considered to be normal, at least if the patient does not have life-threatening birth defects. Distal deletion 18q is not a progressive condition, and the status of the patient does not normally worsen as the person gets older. However, usually they show no significant improvement.

There is a group in San Antonio, TX, which specifically studies all aspects of this deletion. chromosome18.org *CDO has many articles available on this condition, contact info@chromodisorder.org for more details.

Q: Can you provide more information on 3p26.1 deletion?

A: Only 5 patients with deletions 3p26.1 as an isolated chromosomal defect have been mentioned in the literature for the last 5 years (see below), In 4 out of 5 publications patients with del 3p26.1 were just briefly mentioned among other patients reported in these studies. For example, Lowther et al. studied patients with schizophrenia and one of them had del 3p26.1. Rojnueangnit et al. studied patients with cleft lip and palate and one of them had del 3p26.1, etc. These patients have not been described in details. **There is no evidence that the disorders in these patients were caused by this deletion** (or that the deletion was a random finding unrelated to patient's problems). *CDO has the articles mentioned in our library.

FAMILY STORY

The Woodburys

To share your family story, email info@chromodisorder.org

"Your child will teach you more than you could ever teach them." Ain't that the truth. We often take for granted the little things in life, such as your baby hitting milestones and smiling at 8 weeks old, because, you know, the pediatrician says it should happen that way. And all the other things your baby does like rolling, crawling, walking and talking. Tyler was born at 37.5 weeks and everything about my pregnancy and delivery was pretty flawless. We were released from the hospital after the typical 2 days for Caesarean deliveries. Around 12 weeks old, Tyler was still not smiling. He would stare at the wall or lights and wouldn't respond to our voices. It wasn't until he was 16 weeks old did we see something that resembled a smile. As time went by, we noticed he wasn't hitting even more milestones and consulted birth to three specialists. After numerous appointments with specialists and tests including a brain MRI and multiple hearing tests, we learned that Tyler has a chromosome micro deletion. What that means is he is missing 5 genes on chromosome 1. The exact location is 1q42.11-1q42.12. Tyler is globally developmentally delayed, has brain anomalies, an arachnid cyst on his brain, a large head, short stature, low muscle tone, a visual delay, potential hearing loss, a sub-mucous cleft palate, has had two seizures to date and displays some autistic behaviors.

After researching about Tyler for what seemed days and nights, I stumbled on what's called Skraban Deardorff Syndrome. We later received confirmation that this is what Tyler has. Tyler is missing a gene named WDR26 which displays clinical symptoms identical to Tyler. Kids like Tyler with Skraban Deardorff Syndrome are also globally delayed with mild to severe intellectual disabilities, have dysmorphic facial features, seizures, low muscle tone, trouble balancing/walking, autistic behaviors, and the biggest impact to these children is speech. Some kids are non verbal. Tyler is 18 months old now and can sit independently for about 5-7 minutes. He still doesn't crawl, walk, or have any words. But what these kids ALL have in common is they are the most friendly, happy, social and the sweetest kids ever! Everyone who meets Tyler falls in love with his sweet smile. He loves to smile at everyone!

Life with Tyler is busy. It isn't easy. But we remain strong and consistently research ways to help him and advocate in his best interests. We don't take things for granted and we celebrate the smallest of milestones with him. The work he puts in every day is impressive, he is a fighter. The love I have for this sweet boy is like no other. I never imagined being the mom of a special needs child who will probably rely on me for most of his life, but I would never change a thing because he brings so much JOY.

New Chromosomal Research:

A new article analyzes the neurological and psychiatric characteristics of 56 adult patients with 22q11.2 deletion syndrome. Myoclonic epilepsy, parkinsonism, schizophrenia and left-handedness as common neuropsychiatric features in **22q11.2 deletion syndrome**. 22q11.2 deletion is one of the most common microdeletion syndromes. The clinical features of this condition are broad and highly variable across physical, developmental, and mental health domains. **Read more**.

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.

A small 7p22.3 microdeletion: Case report of SNX8 haploinsufficiency and neurological findings. 7p22.3 microdeletion, a small deletion within the short arm of chromosome 7, has been reported in the literature before, but the genotype-phenotype correlation remains uncertain. Clinical features in previous cases include developmental delay, intellectual disability, congenital heart disease, and dysmorphic features. **Read more**.

An adult female with **chromosome 5q34-q35.2 microdeletion**: A rare syndromic presentation of left ventricular non-compaction and congenital heart disease. Microdeletions in the distal segment of chromosome 5q have rarely been reported in the literature with less than 20 published cases. Common features that have been identified in distal 5q deletions included failure to thrive, microcephaly, developmental delay/intellectual disability, abnormal craniofacial phenotypes, and cardiac anomalies. **Read more**.

Genetics diagnostics can be costly and complicated. New research shows it is possible for patients with **Emanuel and Pallister-Killian syndromes** to be diagnosed with high accuracy using only a facial photo of an affected person.

Neuropsychiatric phenotypes and a distinct constellation of ASD features in **3q29 deletion syndrome**: results from the 3q29 registry The phenotype of 3q29 deletion syndrome is characterized mainly by neurodevelopmental and psychiatric features.

Read more about these and other new research publications by visiting

https://chromodisorder.org/latest-research-articles/

SUPPORT CDO

DONATE NOW

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

CDO is a Great Nonprofit

Your hands are washed. You are physically distant.

Now power the fight to end COVID-19. Share your experiences to help scientists understand COVID-19 and how communities can best prepare for the future. Join us in the #FightToEndCovid! #covid19response

fighttoendcovid.com

Download these *Calming Podcasts* on Apple Podcasts, Stitcher or wherever you get your podcasts:

Tara Brach, psychologist and meditation teacher

10% Happier with Dan Harris

Free Remote learning / Homeschooling resources

Check your local library's website for resources like Tutor.com, Kanopy Kids and Learning Express Library.

123 Homeschool4me

Resources are organized by subject and grade. Helpful education activities to keep kids learning and printable worksheets when you need them to sit and get some work done.

All Kids Network

This free and rich content platform provides access to thousands of fun kids activities like children's crafts, worksheets, coloring pages, printable mazes, dot to dot, hidden pictures and more.

Readworks.org

If you're looking to really zoom in on reading comprehension, Readworks is going to be a great fit for you, providing content from kindergarten all the way through 12th grade.

Arcademics.com

Arcademics make multiplayer educational games for students from K to eighth grade, from free math games to language games. Arcademics combines the excitement of video games with educational content to produce a high rate of learning

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Chromosome Disorder Outreach

You Are Not Alone.