April 2017
Volume 3

CDG CARE
Where CARE Grows...

Announcing New CDG CARE Grant Programs

We are excited to announce two upcoming grant programs that will soon be available to CDG families!

The first is the CDG CARE Family Travel Grant Program. Due to the generous donation from WaterStone and the Richard and Ann Anderson Family Foundation, the first grant under this program will be awarded in the amount of $500.00 to support a CDG family from the USA to attend the 3rd World Conference in Belgium this summer. Applicants for this grant opportunity will be accepted April 1 through May 31, with the award being announced by June 16, 2017.

Later this year, CDG CARE will launch the Family Assistance Grant Program. These grants will be available to CDG families seeking financial assistance for expenses that are not traditionally covered by insurance, including but not limited to, treatments, services, therapeutic equipment, communication aids, supplies or other eligible expenses that will improve the overall quality of life for the individual diagnosed with CDG.

CDG CARE is very grateful for the individual and corporate donations that make these programs possible. We are actively seeking grants, sponsorships and contributions to be able to significantly expand upon both of the above

Be sure to check out the global updates provided in this newsletter!

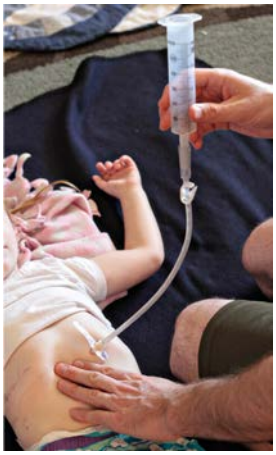
Read about Unstoppable Oliver in this volume of our newsletter!

CDG
Awareness

May 16th - Congenital Disorders of Glycosylation (CDG) Awareness Day!

programs and specifically to make funds available to support families interested in attending the upcoming CDG Scientific and Family Conference in 2018.

Grant program guidelines and applications will soon be available on the CDG CARE website. To support CDG CARE and our mission to continue to provide information and support to families affected by CDG, consider making a tax-deductible donation TODAY!



Read our Welcome to the Tubie World feature.

New CDG Clinic Opening in April 2017

By Christina T. Lam, MD

We are excited to announce the opening of a Congenital Disorders of Glycosylation focused clinic at Seattle Children's Hospital. The clinic will initially run once a quarter by medical staff experienced in glycosylation and deglycosylation disorders with the inaugural clinic on April 28th, 2017. The goal of this clinic is to provide recommendations and guidance to the home care team regarding appropriate management, testing, and monitoring for individuals affected or highly likely to be affected with CDG. At the clinic, we also will provide families with information about their diagnosis and the opportunity to ask questions. In addition, our connection to the CDG research community and family advocacy group may provide support for these individuals and their families. For more information regarding this clinic and how to make referrals, please contact us at CDGClinic@seattlechildrens.org, or call us at 206-987-3012.

CDG Awareness Day - May 16th

"Awareness" is becoming harder and harder to generate in an increasingly info-filled world. With Facebook, Instagram, and Twitter feeds chocked full of stories, causes, politics and cat pictures, it can be hard to stand out in the crowd. CDG Awareness Day is an opportunity to grab the spotlight a little bit and allow that spotlight to edify our friends, families, and communities about the important and complex world of CDG.

World CDG Awareness Day is May 16th. Here are some ideas to help raise awareness:

- Take a selfie with a heart drawn on your hand or face and tag your photo with #WorldCDGDay and #CDGAwareness
- Click and like the World Congenital Disorders of Glycosylation - CDG Awareness Day page on Facebook
- Donate to CDG CARE
- Purchase items that support CDG funding:
  - Order CDG Tea by e-mailing christa.doot@gmail.com
  - Order Awareness items from the CDG Community Store
  - Bravelets to support CDG research
- Sign-up for our newsletter on our homepage www.cdgcare.com

Moments You'll Never Forget

By Melissa Schlemmer

Everyone has moments in time that they will never forget. It may be a fleeting second or an experience that seems endless. Ever since our journey began I can vividly remember these moments that completely changed me, our path, and our lives.

Our family was sitting in our son's neurologist's office when she told us that she believed our perfect little boy had congenital disorder of glycosylation. I held his tiny little body in my arms and thought that if I poured all of my love into him her words would rewind and our lives would go back to "normal." I looked at him and envisioned myself running out of her office; as far as I could go. I was terrified. I didn't know what those words meant for our son and for our family. And to be honest, as much as I knew there was something going on with our little boy I knew saying it out loud made it even more real. I was so fearful of what the future would bring.

That moment in her office our lives changed. Receiving our son's rare diagnosis was just the beginning of living a life full of unknown and worry. Our son has had numerous hospitalizations for common illnesses. He's tube-fed continuously due to slow gastric emptying and hypoglycemia. He's globally developmentally delayed, nonverbal with severe hearing loss, and also visually impaired. He gives us plenty of reasons to worry but he gives us even more to rejoice.

Our son has given me so many moments in time where I can take a step back and am reminded what life is all about. He was over a year old when he started lifting his head off of the floor while laying on his stomach, and I can tell you that I think my entire neighborhood could hear the cheers in our living room. Every accomplishment is magnified and celebrated. He has the most beautiful smile that will change your mood immediately; he may be nonverbal but his facial expressions and eyes speak volumes. He has reminded me over and over to never take anything for granted. He has taught me to live a life full of gratitude.

His diagnosis has changed our lives.

I had no idea when we were sitting in the neurologist's office that next to the unknown and worry was also a life full of joy. It was the beginning of a life full of wonder. A life full of inspiration.

2018 North America CDG Scientific and Family Conference

By Andrea Berarducci

SAVE THE DATE, February 23-25, 2018!

Highlighting the successful collaboration between CDG CARE, Sanford Burnham Prebys (SBP) and NGLY1.org in offering a groundbreaking educational event for families and professionals in 2016, the preparations are already underway for the next North America Scientific and Family Conference. Join us in San Diego to Celebrate Rare Disease Day 2018 to focus on CDG: Moving toward the treatment horizon!

Participation in this 3-day event will center on Congenital Disorders of Glycosylation and De-glycosylation, involving models and therapies for PMM2-CDG, novel approaches to other types of glycosylation disorders, and NGLY1: A Disorder of Glycosylation. A multi-faceted poster session will present emerging science alongside patient and advocacy tools and education. There will be multiple opportunities for professional and parent networking, as well as a fun, family-friendly social activity at a local venue to be announced soon.

Once again, SBP will offer the innovative half-day, doctor-is-in session, allowing physicians and scientists to meet small groups of like-minded family members in an informal roundtable format. The full day offering of family-focused sessions will facilitate the meeting and collaboration of medical professionals and families in order to share knowledge and experience, and discuss common issues of patients living with CDG. The preliminary program is designed to promote the exchange of education and resources, and present alternative and cutting edge treatment and therapies to maximize the overall quality of life and health outcomes for children, youth and adults affected by CDG.

Keep an eye out for this exciting program agenda and early-bird registration that will be released later this summer!

No CDG Left Behind: Why this mom cares about the CDG Natural History Study at NIH

By Cristina Casanova Might

Back in June 2014, when I was 39 weeks pregnant with my 3rd child, I moved heaven and earth to pack my oldest son, husband and mother-in-law on a plane to the National Institutes of Health (NIH) so that my son Bertrand could be one of the first participants in the newly formed CDG Protocol and Natural History Study.

Despite the risk of my husband missing the birth, the difficulty in my high-risk pregnancy, and the challenge of both working and caring for our 3 year-old daughter by myself for a week, the choice to participate in the Natural History Study was a no brainer because the long term benefits for our subtype and greater CDG community were so great.

It's since become evident that a lot of my fellow CDG parents may not fully understand why the study—despite the hardship and commitment—is so critical to our community.

In less than 4 years since it's discovery, our subtype now has four distinct treatments being developed and multiple pharmaceutical industry partners in large part thanks to our tiny community's participation in the Natural History Study.

Yes, you read that right: 4 years, 4 treatments being developed. Even better? Two are already FDA approved.

My hope is that by sharing the NGLY1-CDDG experience, more families will begin to care as much as I do about the Natural History Study and, as a result, accelerate the understanding and the development of treatments for kids with ALL the CDG subtypes. #NoCDGLeftBehind

The Road to NIH

In 2012, when researchers identified two loss of function mutations in a gene called NGLY1 in my son, it had never been identified with a human disease.

By 2014, we'd identified enough patients worldwide that we held our first NGLY1 scientific and family conference in La Jolla, California courtesy of Dr. Hudson Freeze. It was there that we met folks from the NIH. One in particular, Lynne Wolfe was the study coordinator for a new CDG Protocol and spoke about it at the conference. Lynne saw our kids with NGLY1 in person at the conference. Their features were all hallmark CDG, so she said, "Let's see them."

When I was a little girl, my father (a neurologist) was doing his fellowship at NIH. I grew up hearing stories of the miracles made possible there and the importance of basic research and natural history studies. While I hung onto every story and lesson, I never expected that to play a role in my personal life.

When Lynne and Dr. Bill Gahl, the Accountable Investigator for the CDG Protocol, made the off-hand offer to study our NGLY1 kids, I leapt at the chance. And, I followed up (repeatedly), but didn't hear anything until a few months later in May 2014. Someone else with a spot in the study had canceled, and Lynne wondered if we were interested in taking the spot, even though it was last minute. The only possible answer was "YES!"

Understanding for us, Understanding for all

Bertrand was not just the first NGLY1 patient they brought into the CDG Protocol, but one of the first patients period. The questions we asked, the tests they ran, and the features they found informed and expanded the protocol as a result. Because of questions we, as crazy parents, had about his lack of tears and sweat, they added that to the study and have since found evidence of this to some degree in all CDGs.

With every new CDG subtype examined, discoveries and insights are made for all.

About a Natural History Study at NIH

There is a ton of information out there on the importance of Natural History Studies for rare diseases in particular. According to the FDA:

Natural History Studies track the course of disease over time, identifying demographic, genetic, environmental, and other variables that correlate with its development and outcomes in the absence of treatment. Thorough understanding of disease, natural history is the foundation upon which a clinical development program for drugs, biologics, medical foods or medical devices is built.

https://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/OrphanProductsNaturalHistoryGrantsProgram/ucm487336.htm

I won't belabor the the technical points, but I will add a few things:

NIH is the Gold Standard. The Clinical Center is a world class facility for world class researchers. With the instruments, technicians, and clinicians being held constant across the study, it is absolutely ideal when it comes to Natural History Studies. The FDA loves this.

Dr. Gahl & his team are more than just world renowned researchers. In order to be a part of their team, it must be a job requirement to be funny, compassionate, and genuine in addition to brilliant. That is to say, while science is important, they always put people first. They never forget the child and family at the center of their work.

Unlike what is often experienced with academic institutions, at NIH they don't sit on information. They share it. They present and publish findings as quickly as possible. They are fair and open with the work they've done on behalf of tax payers, and share equally with industry, academic centers, patient groups, other agencies, and more.

Given the small amount of funding the CDG protocol has, the outcome in such a short amount of time has been truly remarkable. Dr. Gahl's team has enabled our community to leap frog decades of effort through their generous contributions of materials to researchers across the globe, deposits to a public biobank (Coriell), discovery of diagnostic biomarkers, and critical mechanistic and therapeutic insights.

Hello, Pharma!

Thanks to the Natural History Study there now exists thorough phenotyping, improved mechanistic understanding, biospecimens, biomarkers, and now therapeutic avenues for NGLY1. As a result, pharmaceutical companies are starting to work on treatments specifically for our disorder. The foundation laid by the Natural History Study was critical.

The Future is Today

When I spoke with NGLY1 families about participating in the Natural History Study, I told them that there was very little chance it would help their own child(ren) but the discoveries would certainly help future generations. Our selfless NGLY1 families jumped on board regardless.

Fortunately, it turns out that I was wrong. The benefits of the study were far more immediate than we could've ever imagined. Participants left with actionable information for their home medical care teams, adjusted supplements and medications, and more.

The future generations benefiting from the study are here now. Thanks to the discovery of a urine biomarker in our NGLY1 kids, one child has already been identified from a urine specimen analysis rather than a blood draw. And due to therapeutic insights in collaboration with other researchers, there is work being done with repurposing FDA approved compounds that will benefit patients in a matter of months rather than in a matter ofdecades.

How You Can Help

While my son is reaping the benefits of the CDG Protocol and our subtype is set, it blows my mind that the broader CDG family hasn't rallied behind this.

Last week I heard Dr. Hudson Freeze talk about the 125 difference CDG subtypes and how the list keeps growing.

Again, you read that right: ONE HUNDRED & TWENTY FIVE different CDGs.

Our community is much bigger than we realize. Our impact can be much bigger as well!

I believe that children and families with all 125+ CDGs deserve understanding, treatments and cures. I feel it in my bones like a moral imperative. I also know that the first step on the path to achieving that is a robust natural history study for all.

So, as a community, let's make this happen!

You don't have to be a hormonal pregnant woman to make a difference—you don't even have to leave your house! Here is how you can make a differencetoday:

- Contact your federal delegates to support NIH funding! Give Dr. Bill Gahl's CDG Protocol a shout-out if the spirit moves you. Here is how: http://rareadvocates.org/advocacy-tools/
- Thank the folks at NIH for their hard work. This includes Dr. William Gahl, Lynne Wolfe, and NIH leadership (i.e., Dr. Francis Collins), and encourage them to keep funding this critical initiative! Hint: firstname.lastname@nih.gov works pretty well. :)
- Not everyone can participate but everyone can donate. Consider contributing to a fund at either CDG CARE or NGLY1.org that goes directly to the CDG Protocol. At NGLY1.org this fund will go to fund a research nurse for the CDG Protocol which benefits all CDGs, and is not specific to an individual subtype.
- And last, but not least, consider participating in the study!

We are stronger together and we can begin to change the future for CDG TODAY.

#NoCDGLeftBehind

About the Author

Cristina Casanova Might is the Executive Director for NGLY1.org and a proud board member of CDG CARE. Cristina believes that partnership between patients, families, researchers, clinicians and legislators is essential to accelerate science for rare diseases—understanding, treatments and cures. She can be reached at cristina@ngly1.org.

See the conference agenda and learn more about attending the event.

Unstoppable Oliver

By Claudia Graetsch-Vasquez

Oliver Hans Vasquez was born on July 7, 2007 weighing an even 7 lbs. All 7's, Our lucky boy. Oliver was deemed a healthy baby and we welcomed him home with proud open arms. It was somewhere around 5-6 months when we started to notice milestones delays and other odd behaviors. There was no urgency from his pediatrician, and some therapy was prescribed with the notion he would catch up. But as time went on, our concern grew. At 15 months, and after several months of referrals, blood tests, and scans, Oliver was finally diagnosed with CDG 1a (PMM2 CDG) We were relieved to finally have an answer but frightened by the uncertainty that comes with a rare disease diagnosis.

Early in our quest to find a diagnosis, we were referred to OT/PT with our private insurance and with an early start program through the state. The therapists we worked with (and some whom we still see today) were not only Oliver's therapists, they were ours. Their support and words of encouragement still ring with us today. They saw the determination and drive in Oliver, that we all saw at home. They really knew and understood our smart, brave and determined boy.

Life became busier. We welcomed Oliver's brother Eli into the world, Oliver had started school, and it became our new norm to have our calendar filled with therapy and specialist appointments. We had heard of intensive therapy from other CDG families, and were curious about its results. But since insurance didn't cover it and the calendar was full, it's thought was put it on the back burner. We continued with private and state therapies and fit in the normal activities we enjoyed. We always looked for ways for Oliver to join in, whether in a jogger stroller, on his trike or tagging along in his walker. Oliver's Physiatrist once gave words of advice when we expressed our concerns of not being able to fit in an intensive therapy program with Oliver. He said that the best therapy for Oliver is "to just be a kid". He advised us to get him into adaptive swimming, adaptive team or individual sports like skiing, whatever we could get our hands on. And so we did. I allowed myself to take the pressure and guilt off of getting him into an intensive program. We even scaled back on his traditional therapy, to allow Oliver to do more "regular" kid stuff and give him (and us) a rest. It felt great. Oliver continued to flourish and we could all breathe a little easier. As time went on our interest in intensive therapy renewed and grew with the help of social networking. Following families worldwide, who were sending their kids to Intensive programs and having positive results. We now knew it wasn't a question of if, but when.

As Oliver's younger brother Eli started to grow, move and walk, Oliver took notice. I often say that Eli is Oliver's best therapist and motivator. Oliver and his brother are quite competitive and for the past year Oliver tells us (almost on a daily basis) that he "wants to walk like Eli". This statement makes us smile but also breaks our heart. As private therapy decreased and state therapy switched to maintenance mode (once monthly), we began to seriously pursue and research getting Oliver into an intensive therapy program.

This past Christmas, through the actions of great friends and the generosity of many others, a fundraiser was held to send Oliver to an intensive therapy program and cover all its expenses. With this gift, Oliver will be attending "Now I Can" program in Provo Utah this summer. We are excited for this opportunity and its potential impact on Oliver's continued progress. Daily therapy might be challenging, but I am trying to prep Oliver by reminding him of the goal he speaks of almost daily - "I want to walk like Eli"

Stay tuned…

Welcome to the Tubie World

By Andrea Berarducci

A big warm welcome to the feeding tube world! Odds are, if you're reading this, you are either faced with the decision of placing a feeding tube for your child, you have recently undergone feeding tube placement surgery, or you are an experienced tubie parent.

My daughter had her feeding tube placed at 18 months old, after 6+ months of pondering the decision. With many emotions leading up to finally moving forward with the procedure, the reality is that she has had it now for 5+ years and this one decision, literally and undoubtedly saved her life.

First of all I know how scared you are. The feeding tube world can feel strange, un-natural and you feel totally de-attached to the "real" world. Although there is no promise that your child will eventually eat in the traditional way, what I can promise is that it does get easier with time.

The feeding tube world is filled with long medical words and terms, including abbreviations such as NG, NJ, PEG ,MIC-KEY, G-TUBE, and more. You will come across even scarier terms such as stoma irritation, granulation tissue and possible infections. You undoubtedly are feeling very overwhelmed, but, if there is any consolation, this is exactly how we all feel at the beginning of our feeding tube journey.

Your child may also need a feeding pump. You will look at this medical device as the most complicated equipment you have seen with all the buttons and the many different beeps it has. However, because CDG parents can conquer anything, you will quickly learn every different beep, port, screen message and reset capabilities in a matter of seconds. It is also likely that your child may at some point be bolus fed. This enters an entirely new language of extension tubes, gravity feeds, liquid and blended diets, and learning (and you WILL learn) every medical equipment company that makes syringes, and exactly who makes the best, where to order them, and in what manner is the best cleaning method for all of the above. For a wealth of information, resources and support for all things tube related, visit www.feedingtubeawareness.org

FEEDING TUBE AWARENESS FOUNDATION

You will know the feeding tube world better than your Doctor or consultant. You will be able to vent your child and have them primed ready for a feeding in a matter of seconds. At home, on the go, waiting in line, sitting at the stoplight, you will instantly know what to do in any tubie situation and be a helpful resource to all those entering this seemingly complex culture. The feeding tube world will become second nature to you, your family and all of your child's caregivers.

This next part of the feeding tube world does take time to adjust to and personally can be the hardest part of the feeding tube journey. The "outside world". There is no avoiding the fact that you will have many stories to tell about the looks, questions and stares that you will get from everyone outside of your "inner circle". Some may be polite and try to understand what the tube is for, even if they get it completely wrong, at least they are making an attempt. It is important that you take these opportunities to educate and spread positive awareness of feeding tubes. Most importantly hold your head up high and be proud of the alternate method in which you feed your child, because you certainly have NOTHING to be ashamed or embarrassed about. You are doing an amazing job, for a very special child, who is a thriving tube fed child, because of the love and abilities you have overcome to provide the very best for them.

The lingering question you may never get an answer to is: How long? Will it be forever? Will they ever eat? Many children have a feeding tube for many different reasons, but for how long? The answer is; that each and every child is different and they may or may not ever be able to eat on their own. So, my most important advice is that you just take each day, week, year, milestone as they come. Regardless of your tubie timeline, you will always look back on your feeding tube journey and be very happy that your child was or is tube fed and that you are able to see how much they have thrived and grown because of this decision you made for them. You and your tubie child will be okay. Life will not stop because your child is tube fed. Your entire family will adapt so easily to the feeding tube world and just learn how to live life and enjoy all of the activities that you did before entering this strange world.

So, here we are, welcome to the feeding tube world! Welcome to, yet another, elite group of special needs parents that you never dreamed you would be a member of! We are all in this together, give yourself a huge pat on the back, and know what you are doing an amazing job each and every day!

Two new CDG subtypes; ATP6V1E1 and ATP6V1A

By Fiona Waddell

Prof Callevaert and his team, from the Center for Medical Genetics, Ghent University Hospital, Belgium and the Nijmegen umcRadboud University team in the Netherlands, led by Prof Wevers and Prof Morava described two new Congenital Disorders of Glycosylation (CDG) subtybes; ATP6V1E1 andTP6V1A, which encodes the E1 and A subunits.

Seven patients from five different families have been reported. All patients have skin wrinkling and sparse subcutaneous fat. They had a similar progeroid facial gestalt, which is the mimicking of the ageing of the face, with a 'mask-like' triangular face, a short forehead, an abnormally increased distance between the eyes, inward tilted lower eyelids, low-set ears with malformed folds of the ears, a beaked nose with a broad nasal base and narrow nostrils and a short and pointed chin. At birth, several patients had congenital heart defects. One patient had a severe speech delay and suffered from a generalized tonic-clonic seizure (a seizure that affects the whole brain) at the age of 14. Most patients suffered from severe hypotonia (low muscle tone). Additional, but less frequent features included hip dysplasia (deformation or misalignment of the hip joint), multiple congenital contractures, kyphoscoliosis (abnormal curvature of the spine), marfanoid habitus (symptomes resembling those of Marfan syndrome, which includes long limbs), inguinal herniation (a hernia decending into the scrotum) and testicles which are not descended.

Glycosylation screening, by means of transferrin isoelectric focusing (TIEF; a laboratory test), showed a pattern indicative of a CDG type II by patients with ATP6V1E1 mutations. N-glycosylation was affected to a lesser extent in patients with ATP6V1A mutations. Apolipoprotein C-III screening, however was abnormal in patients, even when routine blood screening was normal.

Now we know of three gene defects (ATP6V0A2, ATP6V1A and ATP6), all causing sagging skin and wrinkling (cutis laxa) at birth. The patients show improvement of skin symptoms with getting older, even their hear disease might improve. Their most important burden is extreme joint laxity and learning disabilities.

CDG CARE Global Corner
News from Europe!

Authors: Dr Rita Francisco **(**CDG Patient Advocacy Group Manager and researcher for Portuguese Association for CDG (APCDG) and Vanessa Ferreira (APCDG founder and volunteer, coordinator of the international research network for CDG (CDG & Allies PPAIN)

The European Reference Networks (ERNs) – Connecting experts, sharing knowledge and information, improving medical care for Rare Disease Patients

In Europe, as in the US, approximately 30 million people suffer from rare diseases.

Rare Disease Patients are globally dispersed and have different care opportunities depending on their home country. The European Reference Networks (ERNs) aim to tackle these hurdles and allow every single patient to get the best possible care by establishing connections and shortening distances.

On 28th February, taking advantage of the Rare Disease Day celebrations, the ERNs were officially launched. This is a joint initiative of the European Commission and Member States with support from the European Parliament (learn more HERE) in full cooperation with EURORDIS (learn more HERE).

Currently, there are 24 ERNs each specialized in a group of rare diseases (to know more visit HERE). Among these networks is the ERN for **Rare Hereditary Metabolic Disorders (MetabERN)** where CDG are included and represented. The ERN's structure and governance ensure patient involvement. To achieve this EURORDIS created the ePAGs (European Patient Advocacy Groups). There is an ePAG representative for each disease group. CDG are represented by Vanessa Ferreira PhD (APCDG Founder, Sister to a CDG Patient). To know more visit HERE.

These virtual networks connect nearly 1,000 healthcare providers across Europe with the aim of facilitating and promoting information exchange, improving treatment and healthcare access, ultimately, accelarating CURES.

Learn more about the tools created by the ERN's HERE.

See EURORDIS Press Release about the ERN's launch HERE

*****CALL FOR HELP*****
Volunteers are welcome to voice CDG patient and families needs!
For further information please contact rita.francisco.28@gmail.com

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Save the Date - 3rd World Conference on CDG for Families and Professionals

On 15-16th July let's all meet in Leuven together "United Shaping the Future for CDG"
The registrations for the 3rd World Conference on CDG for Families and Professionals (learn more HERE) are now open. REGISTER HERE!

Where is it? The conference is taking place in Leuven (Belgium). The conference venue is the Park Inn by Radisson Hotel in Leuven, Belgium (more information HERE)

Who is going? The 3rd World Conference on CDG for Families and Professionals will include19 expert speakers (meet them HERE), among them are physicians, researchers, healthcare professionals and parents. Topics will range from Therapies to Disease Management, from Diagnosis to Patient Reported Outcomes, among many others. Curious? See the conference AGENDA HERE.

Why can't you miss it? This is a unique event that brings together Professionals and Families. It opens networking avenues and reinforces bonds among the different Community stakeholders. The most up-to-date and state-of-the-art knowledge will be shared.

Important information & documents: Please see the conference Booklet HERE

Leuven is the birthplace of CDG. Nothing better than to draw inspiration from the past to plan and help shape the future for the CDG Community!

CDG & Allies – PPAIN
CDG & Allies - Professionals and Patient Associations International Network

CDG & Allies- Professionals and Patient Associations International Network (CDG&Allies PPAIN):
CDG & Liver Working Group

We teamed up to lead a unique research --- the team is HERE

ACCOMPLISHMENTS

- A revision of literature on "Liver involvement in congenital disorders of glycosylation (CDG). A systematic review of the literature". (available HERE)
- Based on that research work, a patient-friendly document was done for families and professionals (available HERE)
- To complement these projects, a new methodology to increase CDG knowledge was set-up –
  - **The Liver electronic questionnaire (eLCDGQ)** (visit HERE)

RESULTS SNEAK PEEK

- ✓ 203 participants
- ✓ The majority of represented patients was under 10 years-old
- ✓ The number of female and male represented patients was similar
- ✓ 2.6% of represented patients still do not have a full diagnosis (unknown CDG type)
- ✓ The vast majority of participants were Mothers

Dr Dorinda da Silva, is currently analyzing the data from the eLCDGQ. We estimate to be able to submit the article on July 2017.

Thus on September 2017 we hope to have all the results publicly available.

CDG CARE (Community Alliance and Resource Exchange) is a nonprofit organization founded by parents seeking information and support for a group of disorders known as Congenital Disorders of Glycosylation (CDG).

Our mission is to promote greater awareness and understanding of CDG, to provide information and support to families affected by CDG, and to advocate for scientific research to advance the diagnosis and treatment of CDG.

Visit us on the web at www.cdgcare.com **or send an e-mail to** info@cdgcare.com**.**

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