

CDG One-to-One Interviews, comprises a series of interviews with some of the most active and influential minds, who make this world a better place for CDG Children and Adults. This is an initiative led by the **Portuguese Association for CDG (APCDG, www.apcdg.com)** and coordinated by Vanessa Ferreira.

Andrea Berarducci: A Mother & Advocate who has an immense CDG care!

CDG changed her life, so she decided to change CDG lives.

2nd November 2016, By Rita Francisco (CDG community social manager. Email: sindromecdg@gmail.com).

Introduction

My name is Rita Francisco, from the Portuguese Association for CDG (APCDG, www.apcdg.com). In this CDG One-to-One interview, I am very lucky to be accompanied by Andrea Berarducci, whose impressive work as a CDG Advocate has resulted in great improvements in the lives of CDG Families. Welcome Andrea—it is most delightful for us to you at CDG One-to-One!

Chapter 1 – Receiving the unexpected diagnosis: It's CDG

Rita Francisco: Six years ago, your life changed. You became a Mother to beautiful Bianca and a few months later you received the medical diagnosis of Congenital Disorder of Glycosylation (CDG). What have been the main difficulties you have faced after the announcement of the diagnosis? How do you manage to overcome them?

Andrea Berarducci: When Bianca was diagnosed there was very little information available and very few resources to contact locally, regionally and through the internet. Although Bianca's CDG diagnosis occurred rather quickly as a result of her physical presentation (failure to thrive, strabismus, inverted nipples, etc...) it was unexpected and devastating. Bianca was 13 months old when her diagnosis was made.

After the diagnosis, many facets of life changed. I would say that the main difficulties were finding the best care providers, both therapeutic and medical to guide us along Bianca's journey. We switched providers, medical institutions and even travelled long distances geographically in order to find the therapists and specialists who would continue to recognize and encourage Bianca's progress and not "give up" on our daughter.

Rita Francisco: How did CDG impact your family and professional life?

Andrea Berarducci: As a parent of a child with a rare disease, priorities changed immediately. It was a very emotional and transitional period. I was unable to continue in my professional capacity as a non-profit lawyer, and I quickly became full-time mom, caregiver, therapy coordinator and medical scheduler.

I took it upon myself to learn everything there was to know about CDG, read every bit of literature that had been published since CDG was discovered by Dr. Jaeken and contacted as many global medical professionals as I could find to ensure that I was providing Bianca every opportunity to receive the most appropriate therapies and treatments available. For a long period of time... years, travel plans

were put on hold, family gatherings became sporadic and friendships strained as my time was consumed with therapies, medical appointments and caring for Bianca.

Due to the nature of CDG, the frequency of illness and impact that hospitalizations had on Bianca both physically and emotionally, my goal for Bianca's well-being is now centered around her quality of life... keeping her as healthy and happy as possible.

Chapter 2 – A CDG Advocate is born

Rita Francisco: You have an immense experience on non –profit organizations, and rapidly you became an active member of the CDG Family Network. What were the 3 major gaps and opportunities that you immediately identified as urgent among the CDG community?

Andrea Berarducci: My parents joke that I was born an advocate! I have always had a passion to fight for what I believe in. Although the subject matter of my advocacy has transitioned over the past 35 years, my commitment to the cause that I represent is my stronghold.

I became involved with the CDG Family Network when Bianca was diagnosed in 2011. At that time, the CDG Family Network had been serving CDG USA families, holding conferences and advocating for CDG for 15 years. The 2012 CDG Family Network Conference was held in Chicago, Illinois.

Reflecting on the success of the conference, it was recognized that there were opportunities to expand the vision and direction of the organization. Specifically, to develop partnerships and collaborations globally with research, patient and professional organizations that were increasing momentum and awareness efforts for CDG.

Rita Francisco: In 2014 you started a new non-profit to represent and help the CDG Community – CDG CARE (Community Awareness Resources Exchange). How was that idea born?

Andrea Berarducci: In 2014, after 18 years of committed leadership and expertise, the CDG Family Network restructured and merged into the new non-profit organization, CDG CARE. Under new governance and leadership, the vision, mission and collaboration of CDG awareness, education and advocacy initiatives in the United States began to expand regionally, nationally and globally.

Rita Francisco: Could you tell us about the mission, goals and briefly the 3 relevant projects that CDG CARE is developing at the moment?

Andrea Berarducci: Yes, CDG CARE's mission is to: 1) Provide information and support to individuals diagnosed with CDG, their families, and the professionals who work with them, 2) Increase public awareness and understanding of CDG, 3) Develop a resource of programs and services to help individuals build strengths and meet challenges from early childhood through adulthood, and 4) Encourage and support research into a wide range of issues related to CDG.

We have a number of strategic initiatives that are ongoing and in the development phase. Currently, we are preparing our planning and fundraising committee to focus on the 2018 CDG Family Conference. We are also continually working with our Medical Advisory Board and international allies to bring the most current research, collaborations and patient stories to the global community through

the publication of the CDG CARE newsletter. And in addition to organizing CDG Awareness Day efforts for May 16, 2017, we are developing a resource database consisting over 200 US medical institutions and clinicians to increase CDG awareness and news distribution and facilitate educational webinar opportunities to make 2017 the second official CDG Awareness Day recognized nationwide.

Rita Francisco: In your opinion, why are collaborative initiatives between patient groups and the scientific – medical community so important for CDG?

Andrea Berarducci: Both patients and the scientific/medical community hold the key to CDG research, therapies and treatments. For families to understand the importance of CDG research and how they can contribute, they typically rely on organized patient groups for education and networking opportunities. Patient groups serve as the trusted liaison between the families and the scientific/medical community. This is the most efficient framework for setting goals, aligning initiatives, supporting funding and research efforts, and finding a cure for CDG.

Rita Francisco: What is the relevance of creating awareness about CDG and related rare diseases?

Andrea Berarducci: Awareness fosters education among families, communities and medical professionals. Through the promotion of awareness an early and accurate CDG diagnosis can be made, and with a diagnosis, families are able to reach out for support and resources specific to their rare disease. Additionally, awareness generates interest, which leads to research opportunities, advancement in therapeutic options, and ultimately a cure for many individuals diagnosed with CDG.

Rita Francisco: What can people do to get involved and help the CDG cause?

Andrea Berarducci: We all need to work together. CDG is such a rare disease that in order to make therapeutic advances to benefit all of those diagnosed with CDG, we must work collaboratively to increase educational opportunities and raise funds for this cause. CDG CARE needs volunteers. CDG CARE has several strategic initiatives that are ongoing and in the development phase, but we cannot continue to make a difference without funding and volunteers.

It seems that over the past 3 years, individual efforts to create CDG non-profits in the US has tripled and we need to all recognize that in the current philanthropic climate, funding is limited and competitive. It is my hope that families and professionals will understand that competing against ourselves will only set us back and limit the impact that we can make as a collective group. The passion of our CDG community members is strong, and if we join together, we can make a difference and discover the advances that will lead our children to a cure.

Chapter 3- Future Perspectives of a CDG Mother and Advocate

Rita Francisco: What are their main needs, hopes and wishes of the CDG community?

Andrea Berarducci: It is my hope that enough awareness can be raised around CDG so that families are diagnosed sooner. With greater awareness, and more collaboration among family and patient

groups, we can consolidate efforts to support research and make advancements in therapies for individuals affected by CDG.

Within the US, I hope that we can increase funding and support for the CDG natural history study so that WHEN treatments are discovered, we have the data available and timely to proceed with obtaining the required FDA approvals as quickly as possible. If we can position our medical professionals with this data, for all CDG subtypes, we will be able to avoid delays in connecting families with the resources they need to treat their children.

Rita Francisco: What are the most important lessons that CDG, CDG patients and families have taught you?

Andrea Berarducci: That we all have the same goal of providing the best care possible for those affected by CDG and that we are all committed to openly communicating, sharing and learning from the experiences of each other. The private social network tools that have been created for our CDG families is invaluable and full of resources that we simply would not be able to find on a local level.

Rita Francisco: What do you hope to accomplish for CDG in the short term?

Andrea Berarducci: For CDG CARE to be a resource for clinicians throughout the US to provide to newly diagnosed families. It is also my hope that over time, CDG CARE can develop the resources to provide assistance to families to attend national and international conferences so that we can increase the number of families having the opportunity to meet each other and that more families will be able to develop their own personal resource network for their child and for their family as a whole.

Chapter 4 – Family

Rita Francisco: How do you balance your family life with your role as CDG patient advocate? Are they distinguishable and separate or are they intertwined, and really one and the same, issues?

Andrea Berarducci: I believe every parent is an advocate every day. As a parent of a child with CDG, trying to keep them separate would definitely be a challenge! However, with regard to the volunteer work that I do for CDG CARE, I make every effort to keep my time committed to these tasks separate from my family activities. But, depending on the status of each initiative and event that is being planned, this turns out to be a difficult goal at times!

Rita Francisco: Last but not least, could you share with us some of your family hobbies?

Andrea Berarducci: As a family we enjoy any type of activity that gets us outside! Bianca loves the fresh air, so be it a trip to the park, zoo, or even a weekend camping excursion, we try to plan our weekends around these types of activities. Living in Colorado, it is an easy drive to waterfalls, mountain hikes and just strolling local shopping... fortunately, with over 300 days of sunshine annually, we have the opportunity to enjoy being outside year round!

Who is Andrea Berarducci?

Andrea Berarducci is a committed CDG Advocate and Mother. She has been involved with non-profit organizations since 2003 and has great expertise on the matter. Moreover, since 2011 she has been working as a volunteer in the CDG cause. In 2014, with the goal of improving the impact on the CDG community, Andrea created a new non-profit dedicated to CDG awareness and education - CDG CARE. Since then, she has been presiding over the organization and dedicating her time to developing several strategic initiatives to improve the quality of life of CDG patients.

