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Dyskeratosis Congenita Outreach, Inc.

May You Never Be Alone

The DC Outreach? (you name it)

We are trying to come up with a name for our newsletter and would like YOUR help. If you have any ideas please send them to Nancy Cornelius at snacs6@aol.com and put DC Outreach as the subject. Some suggestions have been "The DC Outreach Companion," "Observer," " Advocate," " Champion," " Outlook," " Frontier." We'd really like your feedback. This newsletter is for you.

President's Corner:

With the beginning of the new year, we find ourselves reflecting upon our growing support group – where we have been, and where we are now.

In the two-plus years since we started Dyskeratosis Congenita Outreach, we've been nothing short of amazed at how people have connected with this organization, with their time, their donations, and their good will.

Two and a half years ago DCO consisted of little more than a website so obscure that even Google hardly knew our name. Yet just a few months ago, we hosted an international meeting of families affected by this disease, which is so rare it occurs in fewer than one in one million people in the U.S. Most people who have it have never met anyone else who does outside of their own families.

At camp we hosted six of the world's leading experts on Dyskeratosis Congenita, including renowned trailblazers like Dr. Inderjeet Dokal of London, England, and Dr. Blanche Alter of Bethesda, MD. Our hope is that these clinicians will form even stronger bonds than they already have, catalyzing further cooperation in research amongst themselves and in the wider community of scientists.

Medical research, public education, community-building: these are the goals we set out to accomplish for this most unusual community of families three years ago. Through literally thousands of hours of work by our all-volunteer board and more than ten thousand dollars in charitable contributions to date, we're getting there.

To everyone who has supported us so far, you have our utmost appreciation. All we have accomplished would not have been possible without you.

To everyone who has been touched by this organization already, we'd like to thank you for validating our mission and our efforts. If just one family has been helped in some way by DC Outreach, then we have meant something.

To everyone who is still thinking of reaching out to us to make a donation or find out how you can get involved, now is the time to act!

And to the families who need us, who are the reason we exist, we urge you to get in touch and tell us what we can do to help you.

Maybe it's finding someone to talk to. Maybe it's getting advice on finding doctors or comparing medical notes. Or maybe you just want to make sure you're the first to hear the details about our next event at Camp Sunshine, which is tentatively slated for September, 2012.

For more information, please go to our website, www.dcoutreach.com, or contact Nancy Cornelius, at snacs6@aol.com

Thanks again – we wish you all a brave New Year.

-Joe Carlson, DCO President, 2008-2010
-Nancy Cornelius, DCO President, 2010-2012

My Camp Sunshine Experience

By Robin Huiras

Throughout my life, I've lived with a sort of isolation. A loneliness that stems from living with a disease so rare that meeting another person with Dyskeratosis Congentia edges on impossible. Several of my family members have been diagnosed with DC or died from it, and I'd heard about others with it, but coming face-to-face with someone affected had always been out of reach.

Until this past fall. In late September, that sense of isolation dissipated as I met and quickly connected with not just one or two people with DC. At Camp Sunshine in Casco, Maine, 12 families from the U.S. and Canada came together to learn from doctors and each other about the disease and its implications.

For three days the adults listened to world-class medical experts explain ongoing research to diagnose, treat and cure some of DC's most devastating symptoms. While we grown-ups got to know one another and absorb the latest DC news, the kids spent the hours romping around the camp's verdant grounds with the most excellent volunteer staff. Each youngster was paired with a counselor and encouraged to take advantage of Camp Sunshine's amenities -- swimming, canoeing, gaming, relaxing, painting, dancing, singing, joking, and, of course, eating.

From the moment we arrived on Sept. 30 to the time we left on Oct. 3, we were engaged. In fact, I felt fueled by a group energy -- well, that and the ever-present coffee. For me, it was an enveloping force that that filled me with warmth and comfort. I was at ease just being my quirky self. And I relished the fact that I was appreciated for my ideas and opinions -- a far cry from the reaction I get upon meeting a new doctor.

For all of us campers, the focus was on what's possible, instead of what is not.

Camp afforded me the opportunity to share, comfort and laugh with those whose lives have been affected by DC. By bringing families together, camp created a venue where connections were forged and lives inexorably changed. I will never forget the looks of joy on the faces of the kids as they danced at the costume party. Or the exuberant voices of campers and counselors singing, "hands up, baby, hands up" during camp's awards ceremony.

And more than anything, I cherish having been gifted with an extended family of men, women and children whose eyes belie that same optimism as mine -- we will not let this disease dictate who we are or what we can do.

And just like the kids who left Camp Sunshine asking their parents when they could come back, I can't wait to revisit this special place to strengthen the bonds with those who've so immeasurably affected my life.

Camp Sunshine: The Kids' Perspective

By Mari Fearon with some help!

The way the teens tell it, Camp Sunshine was one of the most fun places in the country.

It transported them away from the stresses of their real lives and the pressures of school, away from the constraints of living with a tough disease or belonging to a family coping with Dyskeratosis Congenita.

Just for a time, they got to laugh, play, dance, swim and make new friends with other people who understood them, without explanations. They climbed trees, went canoeing or danced till they dropped. They got love and attention that was like a warm blanket. Many cried when they said goodbye to new friends and the wonderful counselors who only three days before were strangers and who now wept with them.

Our teen and preteen campers said it was a special bonding experience that helped them and their families return to their lives knowing others out there coping with DC were now only a phone call or a click away.

We asked them to write about their experiences. Here are a few of their voices:

Aileen, age 14: Everyone was wonderful from the kitchen staff to the counselors and doctors who gave speeches to the adults while the kids went off and did activities. We did many fun things: swimming, miniature golfing, arts and crafts and more. The counselors my sister and I had were great. They played with us, took us to activities and sometimes even to get hot chocolate. I always love going to places where you can truly be yourself and you don't have to try and impress anyone or get anyone to like you because everyone there not just likes you, but loves you just the way you are.

Andrea, age 13: The Camp Sunshine weekend was a family event so I got to miss two days of school to go! At first I really didn't feel like going but there were other girls my age there and we got to do all kinds of really cool activities, like swimming, archery, mini golf, ropes course, arts and crafts, even a costume party and talent show. I never expected to have such a fun time and I made some friends who I would like to stay in touch with. I hate to say that my mom and dad were right about the weekend, but they were.

Kaitlin, age 11: I had so much fun at Camp Sunshine. I liked "Just Dance" for the Wii and the hockey puck table. They had activities like playing mini golf, swimming, arts and crafts and outdoor activities. I liked playing with the counselors, the food and getting to climb a tree. I loved the talent show and the costume party with dancing. I loved sleeping in cabins and getting to meet so many wonderful people. I loved coming to breakfast in my pajamas, getting to spend time with my friends and all the people were so nice. Most of all I got to be there with people just like me and I didn't feel

(con't inside)

like I'm the only one going through this and no one minds what you look like and what you can and can't do!

Maeve, age 12: There are things in life that you look forward to for ages and you're so excited for. Then when it's over, you're sad. That's what it was like for me at Camp Sunshine.
You become so close to your counselors and to other kids in your group. Leaving there was so hard. I had become close friends with some counselors in such a short period of time. They were like your parents for the weekend only better! It's so nice to know that other people there have been in the same situations as me whether as a sibling or as a child with Dyskeratosis Congenita, like my sister.
When you arrive at Camp Sunshine it's almost like you leave reality for a little while and just get to have fun. It's like your own perfect world. When you're at Camp Sunshine, all your worries fade away. Thank you Camp Sunshine!

Ryan, age 19: Camp Sunshine is a good time. They have a lot of fun things to do. They always plan the most fun things to do all day. Every day you are there they have rock climbing, indoor pool and all kinds of games. It's the best place you could stay at. Everyone there is nice no matter who you are. You can't go to Camp Sunshine and not have fun!

Coping with DC: Be Proactive, Do Your Homework, Know Your Options

By Dr. Jakub Tolar

Where in some diseases, a diagnosis brings the relief of finally knowing what to expect, the diagnosis of dyskeratosis congenita launches the affected person and family into a new world of uncertainty. The more resources that can be put in place to provide help and support, the better.

It can be difficult under the best of circumstances to navigate the physician egos, the insurance companies, and everything else involved in managing a complex illness. Children and adults with DC can be stable or have no obvious signs for long periods of time. However, experienced families already know that life with DC frequently becomes a cycle of calmer times alternating with crisis situations. As difficult as it is to sacrifice some of the calmer time, this may be the optimal time to identify a health care team.

There are physicians who, despite extensive training and commitment to some parts of medicine, may not be helpful because they have little interest in DC, lack the ability to feel sympathetic with the patient, or simply don't like to treat a chronic illness that they must manage rather than cure. They may be good doctors, but they are not the right doctors for your team. On the other hand are physicians who, while having excellent medical knowledge and expertise in their own field, will listen closely, provide support to the family, and be willing to learn about what makes DC signs and symptoms relevant, not to the disease, but to your child or loved one. It doesn't matter so much what the doctor's field of specialty is, as long as you feel that you have found the right partner. They can help you build your medical team by identifying other like-minded physicians.

Another way to locate good physicians is through a DC support group. The family support groups are, in my opinion, irreplaceable. The opportunity to speak to someone who has gone through the process that you or your loved one may need to go through helps both in gaining more education and in relieving some of the anxieties that come with it. Another strength comes from the fact that families can share knowledge and become more proactive in gathering the information from their doctors, nurses, and other health professionals.

Along with finding a medical team and a support group, it is important to learn about possible treatment options. Your physician can guide you in this, your support group can educate you, and you can also do research on your own. Today, anyone with a computer and internet has a huge amount of information available to them; most of us are also familiar with how careful we must be to look at the source of the information to see if we can trust the information itself. With some self-education and guidance from a genuinely involved physician, you can learn all that's needed to know to make the decisions that are optimal for each individual family. Medicine does not need to be hopelessly complex; it just has to be explained in an open and logical way. Anyone can understand the relevant details of disease origins, and the risks and benefits of treatment interventions.

In DC, these fall into three categories: 1) supportive care with transfusions and antibiotics, 2) medications that increase the blood counts (such as white blood cell growth hormones and anabolic steroids), and 3) exchanging the faulty blood-forming stem cells all together by means of bone marrow transplantation.

Each of them has, as anything in medicine, side effects. The supportive care with transfusions leads inevitably to iron overload, which can damage the liver and heart. It will also lead to an immune response to white blood cells from other people, which may diminish the success of a bone marrow transplant later on. The anabolic steroids can cause dysfunction and tumors in the liver, and can also change the delicate metabolism in the body, making heart disease more likely in later life. The big problem of these two kinds of interventions is that they are rarely permanent. At one point, the balance of favorable and unfavorable effects tips in a way that the side effects are more prominent and the pattern of diminishing returns occurs.

The third intervention, the transplantation of blood-forming cells from someone else, is one of the most serious procedures in medicine. It is based on the elegant possibility that stem cells—the all-purpose, multi-talented mothers of all cells that live in our bone marrow—can be taken from a person without DC and put into the body of someone who does have DC, and in this fashion replace all the non- or poorly-functioning blood-forming stem cells. If engrafted successfully, these stem cells live for a person's entire lifetime and will restore the blood counts to functional levels, which will in turn prevent further drops in red blood cells, prevent bleeding, and prevent infections. The drawback of stem cell transplantation, which can be achieved by either bone marrow transplantation or transplantation of umbilical cord blood, is that the medications that are needed to make space for the new donor blood system and the immunosuppression used to prevent the immunologic complications of the transplant, have many side effects. Every patient I have ever taken care of who has undergone a bone marrow transplant has had significant complications. Some of them do not live through the transplantation process.

Treatment improvements are in the research pipeline. Many are still in the very early stages of testing, but hold the hope of correcting the disease without requiring the extreme consequences of transplant preparation and maintenance.

The disease path for families affected with DC is marked with grief, loss, and uncertainty at every juncture. Almost every moment and every aspect of the lives of people with DC is overshadowed by this disorder. This can lead to isolation, denial, protest, and disorganization in the lives of families. Equally strong, however, is the desire to take the challenges of the disease head on and balance the sense of loss by maintaining hope and sustaining a semblance of normal family life.

Modern medicine is incredibly helpful to people with DC, and we know with considerable certainty that the recent successes in medical research will be followed by new approaches holding the promise of curing both blood-forming and other defects in other DC individuals. This research is what advances clinical care and provides hope for us all.

Telomere Research

By Dr. Suneet Agarwal

For MDs:
Dyskeratosis congenita: a disease of telomere biology
Telomeres are protective, repeating DNA/protein structures at the ends of chromosomes. In most cells, the telomere repeat length decreases with every cell division, and when the telomeres reach a critically short length, the cell can no longer divide (senescence). The senescence response to shortened telomeres is important to maintain genetic integrity in the cell and to prevent malignant transformation. Telomerase is an enzyme in stem cells that restores telomere length, which allows continued cell division (self-renewal) for the replenishment of tissues throughout the lifespan of the organism. Dyskeratosis congenita (DC) is caused by mutations in genes that encode telomere or telomerase components, which results in an impaired capacity for self-renewal, and premature degeneration of multiple tissues in DC patients.
For patients:
Dyskeratosis congenita: a disease of telomere biology
The body is made up of cells that have a certain lifespan, which are replaced by other cells that divide and grow. Cells have a built-in mechanism (telomeres) that determines how many times they can divide before they die. Telomeres are long repeating structures at the ends of the genetic material (DNA) in the cell, but every time the cell divides, telomeres get shorter. When telomeres reach a critically short length, the cell must stop dividing. This is part of normal aging, and prevents cancer. Some specialized cells called stem cells are responsible for restoring the cells in our body throughout life. They are able to divide indefinitely because they make telomerase, which restores telomere length. Patients with dyskeratosis congenita have defects in telomerase or telomeres, which results in premature "aging" and degeneration of many cell types (such as the skin and blood) in the body.

The DC Outreach, Inc. Board is:

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Please check us out at www.dcoutreach.com. If you wish to keep receiving our newsletters, you must register with our database so that we have a mailing address for you. For any of you who couldn't join us at Camp Sunshine in the fall of 2010 we would like to ask you to take 2 minutes to go to our website and fill out a quick survey to help us better plan Camp Sunshine 2012. And everyone, please start planning to join us at Camp Sunshine in September of 2012. We look forward to meeting you all there!
--The DC Outreach Board

Camp Sunshine 2010