

Chapter 29

Navigating Telomere Biology Disorders

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Introduction

When faced with the diagnosis of a rare illness, one of the greatest challenges is understanding what you can and cannot control. Not only do you not know what you are dealing with, but more often than not the medical care providers are also at a loss. Everyone responds to this situation in different ways. This chapter is designed to help you navigate this complicated disease, advocate for yourself or your loved one, and care for yourself and other members of your family to sustain yourself for the journey ahead.

DC is part of my life, but DC does not define me.

—Young adult with DC

A diagnosis of Dyskeratosis Congenita (DC) or a Telomere Biology Disorder (TBD) comes with a complexity of concerns. Living with a rare diagnosis, with an illness course that is not fully understood and can vary greatly from person to person, is filled with layers of uncertainty. A challenge for many people newly diagnosed with DC/TBD is how to embark on a journey with an uncharted path and few fellow travelers. The ever-evolving scientific landscape and a committed cohort of scientists and clinicians afford hope and great promise for the treatment and management of DC/TBD.

Whether it is regarding the “triad” of DC (i.e., abnormal nails, skin pigmentation, and oral leukoplakia), short stature, bone marrow failure, lung disease, cancer, etc., the clinical characteristics of DC can cause considerable discomfort, require regular attention, and impact one’s well-being, self-esteem, and quality of life. Those things that cannot be seen (e.g., low blood counts, fertility issues, liver disease, etc.) may dictate many aspects of the life of a person with DC.

Depending on one’s coping style, adapting to an illness with multiple, varied manifestations can create unprecedented anxiety. Not knowing what the road ahead holds can be immobilizing and can create self-imposed limitations. There is the possibility of pulmonary fibrosis, liver disease, cancer, and other life-limiting complications. In addition, the need for bone marrow or solid organ transplantation, or other serious intervention, may be looming. Coping with DC involves learning to adapt to those future possibilities, but not becoming overwhelmed by them. Part of coming to terms with DC/TBD is about acquiring the knowledge to manage the illness, and not having the illness manage you.

When families facing DC/TBD come together, a subtext is revealed: regardless of what is shared in common, there are also many differences. Each person’s story is their own,

adding to the emotional complexity of DC/TBD. There can be comfort in the differences as well. Uncertainty is cumbersome emotionally but also holds out hope as one of its components.

The Emotional Journey

When I was diagnosed, I wished it was breast cancer, because it would be something people knew, understood, and for which there were specific treatments.

— Adult with DC/TBD

Although all life-threatening illnesses can cause an individual or family to feel isolated, isolation is more “prevalent” with less “prevalent” diagnoses. The availability of illness-specific organizations can dramatically change the experience of the illness. Team Telomere is that organization for families affected by DC/TBD.

Team Telomere, formally Dyskeratosis Congenita Outreach, was started in 2008 when researchers from the National Institute of Health, including Dr. Blanche Alter and Dr. Sharon Savage, brought together patient families for support and to help research move forward. The premise, set by our founder Nancy Cornelius, was that no one is ever alone. In the years since the forging of the organization, the mission has always remained consistent.

Individuals living with chronic, life-limiting illness develop different coping styles. One can be proactive, coping assertively by choice. For example, one can choose to compensate for limited knowledge about DC/TBD by seeking out all possible relevant medical information. This style may not work for everyone. Some people may choose to know less, managing things as they happen; others may try to ignore the illness altogether. Some will choose to identify someone in their life to become the “keeper” of the most up-to-date clinical information.

The type of “information seeker” you are will influence how you proceed to gain knowledge about DC/TBD and what you do with that knowledge, whether you are an individual with DC/TBD or a family member. In more common illnesses, even a passive person can be inundated with information from media, colleagues, friends, and family. With information not overly abundant regarding DC/TBD, you have to be willing to seek out knowledge.

Part of the barrier of a rare disease is that throughout the journey you always feel alone.

– Parent of a child with DC

The rarity of the illness and the wide range of symptoms do not make DC/TBD easy to diagnose. In many cases, symptoms precede the diagnosis for an extended period of time. Knowing that something is wrong but being unable to quantify it can create uneasiness, loneliness, an ongoing sense of anxiety, and questioning oneself. Even after the diagnosis is established, that period of uncertainty may have established a pattern of coping that persists.

Another common challenge of DC/TBD is having an illness that is difficult to explain to others. Until recently, most people had not heard of a telomere. Telling someone that you have short telomeres does not automatically engender resounding support or compassion.

Because it was unknown, people did not know how devastating, even fatal it was.

– Adult with DC

The Role of the Advocate

To be an advocate for somebody who cannot advocate for themselves, or a patient who is fully occupied with the challenges of their disease and treatments, can be difficult in many ways. This is often the case with DC/TBD; a very few truly understand the disease and what it means to each individual can vary drastically. Engaging with resources upon diagnosis is critical; these can come in many forms. Teaming with medical providers and becoming a partner in care is vital. Some things to consider when advocating for your loved one include:

- **Patient advocacy organizations** such as Team Telomere offer support in the forms of regional access, financial assistance, connection to specialists familiar with the illness, and community connections.
- **Social workers** offer support and mediation between medical/mental health teams, insurance, government agencies resources and family members. Ask the center where your DC/TBD patient is being treated for a referral to a social worker.
- **Palliative care**, often misunderstood, offers support and comfort to the entire family when a loved one is living with a serious illness. It's never too early to start learning about the services and additional support palliative care can provide, as current treatments and plans for care continue. Ask the center where your DC/TBD patient is being treated for a referral to palliative care.
- **Education advocates** have knowledge of your region's educational laws, e.g., 504 plans and individual education programs (IEPs). Note that these can extend through the college years. Ask your child's school for a referral to an education advocate.

Identifying and caring for these relationships is the responsibility of all parties involved. Coming from a place of partnership is key, with collaboration as the focus for the best interest of the individual with the disease.

Staying organized throughout your loved one's medical journey is critical. You will likely be inundated with information from numerous sources, and staying organized can provide a sense of control over the situation. Create an organizational system that works for you to keep track of conversations, medical records, contacts at various centers, next steps, etc. This could be in a binder or notebook, or it could be an electronic record (e.g., Google Docs).

Nobody knows you or your loved one better than you. Never be afraid to ask questions, send inquiries, or be made to feel that you've done something wrong. DC/TBD research and discovery are currently progressing very quickly, and what we knew three years ago seems light years behind us at times. Treatments and research move forward because of those that work for the betterment of individuals with DC/TBD.

Caring for the Caregiver

Being a caregiver creates tremendous demands on the mind, body, and spirit. In line with Team Telomere's founding belief that no one is ever alone, caregivers are encouraged to do whatever they can to care for themselves during what may be a protracted medical journey that is more of a marathon than a sprint.

- **Take time for self-care.** Caregiver burnout leads to patient suffering. Remember the mantra shared by one nurse caring for a DC/TBD patient, "You cannot serve from an empty vessel." Another expression that especially resonated with a parent of a DC/TBD patient is, "Put on your own life jacket first."
- **Stay connected to friends and family.** Do not be afraid to ask for help. One way to visualize a support network is to imagine concentric circles with the patient at the center, the primary caregiver in the next outward circle, supporters of the caregiver in the next outward circle, and so on. Anyone in these circles can lean outward for support. Sometimes well-intentioned supporters make generic offers like, "Let me know what I can do to help." In response to this, many patients and caregivers have reported that it is most effective to request help with very

specific and defined activities, such as providing meals or child care on a certain day of the week or driving the patient to and from scheduled medical appointments.

- **Care for your body in ways that nourish you.** Walking outdoors, exercise, yoga, meditation, warm baths, aromatherapy, reading a favorite book, sleep, hobbies, and taking time to prepare and eat a healthy meal are all examples of self-care.
- **Connect to others sharing the same or similar experiences.** Look to support groups at the center where the DC/TBD patient is being treated and/or through Team Telomere.
- **Mental health specialists** with expertise in illness and grief and/or supporting caregivers can be tremendously supportive. Ask the center where the patient is being treated for referral to a mental health specialist.
- **Gather daily inspiration.** As you go through a long medical journey, along the way you will likely run across quotes, images, text passages, and comments from people you interact with that are especially meaningful and impactful. Collect these in a notebook, Google Doc, or whatever storage format works for you, and continue to refer to these to remind yourself of your goals, vision for success, and the support network around you.

Dyskeratosis Congenita in Families

Attending the DC family meeting allowed us to reconnect as a family.

— Parent of a child with DC

DC/TBD impacts the whole family. The number of affected family members and their ages will influence the emotional profile and needs of a family. The inheritance pattern of DC/TBD presents an emotionally complex story, with the illness affecting multiple generations in some families. The fact that a grandparent, parent, and child can be

dealing with DC/TBD at the same time, or that some can be lifelong asymptomatic carriers, adds to the multiple layers of complexity.

I knew I had what my father had.

– Adult patient with DC

Prior to diagnosis, living without knowing what is wrong, but in some cases seeing that you and other family members share similar symptoms, can be unnerving. Alternatively, it can cause family members to believe that those symptoms carry less significance, because they seem more commonplace when they present in more than one family member. The diagnosis is complicated, both for those in the family who are affected and for those who are not.

I have learned that the 'non-affected' family members are impacted as much by my diagnosis as I am.

– 33-year-old adult with DC

Parents' Journeys

When a child is diagnosed with DC/TBD, orchestrating their medical care, sustaining family life, managing responsibilities and family finances, all while maintaining hope, falls to the child's parents. They must explore and assimilate tremendous amounts of information to stay the course.

The diagnosis itself presents an emotional crisis. It may take time before parents can move from shock and disbelief to a more proactive mode of coping. Many parents feel anxious or depressed upon learning the diagnosis, unsure of what to expect. However, finally having a diagnosis may alleviate some of the existential anxiety which develops when you know something is wrong, but you do not have an answer. The ability to

contain the anxiety, manage the emotions, make decisions, enjoy life, and continue to function, are all skills to be mastered.

If a marriage was previously stressed, difficulties in the relationship may be further exacerbated by the illness. However, in some situations, couples may feel that the strain and the magnitude of the issues they face enable them to become stronger together. Individual parents may cope differently. One parent may need to learn everything there is to learn to plan strategically for the future, whereas the other may choose to stay focused on the moment at hand. One parent may need to talk and to cry, while the other may appreciate silence. Differences in coping styles may relate to gender, culture, age, and personality, and should be acknowledged so that each parent can be supported for their strengths, insight, and ability to adapt during the course of the illness.

Depending on parents' ages at the time of the child's diagnosis, or that of an adult with DC/TBD, the implications for the family are great with regard to having more children. The increased success and refinement of preimplantation genetic diagnosis (PGD) and surrogacy options present methods that can be utilized to conceive a child to be a matched donor for stem cell transplantation for the child with DC/TBD, or for someone with DC/TBD in choosing to have an unaffected child.

Assisted reproduction can be physically, emotionally, and financially draining. Unsuccessful PGD attempts may serve to delay having more children and can create other conflicts. This phase can be an emotional one in the life of a DC/TBD family, as treatment options, as well as additional children, stand in the balance. Successful PGD attempts can set the course of a family towards having a baby and planning a stem cell transplant, creating an unusual dichotomy: anticipating the birth of one child and the transplant of another. Whether you have DC/TBD yourself or have a child with DC/TBD, actively choosing to have a child without DC/TBD can present an existential crisis within the family.

The Journey for Children with DC/TBD

Even though we all have DC children, they are all different.

– Parent of a child with DC

Physical and other differences may set children with DC/TBD apart from their peers and can be factors that cause children to feel isolated, lonely, or depressed, affecting their self-esteem and ability to focus on age-appropriate achievements. Counseling and meeting others in similar situations can be a great benefit. Children also need to feel that they can confide in their parents, their medical team, and important people in their lives when they feel limited physically or emotionally by DC/TBD.

How parents accept and face the illness will influence how children with DC/TBD develop and adapt to it. If parents can create an environment that allows for dialogue, children will find it easier to ask them questions about their illness and treatment and become more active participants in discussions about DC/TBD and its management. This is true whether it is the adult or the child who has DC/TBD (or both).

Children often know much more about DC/TBD than adults might believe. In addition to what they have been told, they have independent interactions with professionals and other children while in the hospital and may also overhear information from ambient conversation. Children tend to be good regulators of their own knowledge base, providing insights about what they know and what they want to know.

Children of all ages and all stages of illness need to be allowed to continue to grow, regardless of the status of their medical condition. Maximizing their capacity and recognizing achievements of all magnitudes will enable the development of emotional strength and support continued growth.

At any age, the educational environment may present unique issues for children with DC/TBD. Upon entering school, children begin to see themselves in comparison to other children. School may be where they learn that everyone does not have DC/TBD or a family member with DC/TBD, not everyone has so many doctor visits, needs blood drawn frequently, or takes medication. This age becomes a time of further inquiry and therefore presents an opportunity for greater understanding and growth. If a child is sick and unable to attend school, if they are unable to participate in activities because of their physical ability or limited stamina, or if they are perceived as different from their peers, they may begin to feel depressed or emotionally uncomfortable in a manner that they may not have experienced previously.

Children need support learning how to adapt, respond, and connect to their peers around matters related to DC/TBD. As school-age children grow, they begin to differentiate themselves from their families and develop increasingly strong relationships with their peers. Physical limitations, or particular treatments (e.g., bone marrow transplantation) may influence a child's social activities and relationships. Each child will need help finding a balance between social and family relationships in the context of living with DC/TBD, allowing the child to feel nurtured while gaining a sense of independence.

Development of a child's sense of self and how that relates to their illness will be influenced by the age and developmental stage at which they learn about their diagnosis. A frequent concern for parents is what and when to tell children about DC/TBD. At each stage of development, children need age-appropriate explanations of the condition and its required treatment. Such information should grow in sophistication as the child grows. Developmentally appropriate explanations and access to information throughout the illness experiences enhance the child's ability to understand and deal with DC/TBD.

As children get older, they begin to assent, consent, and participate in decision-making about their own care. As their children become more active decision makers, parents

may feel some relief that they are now making decisions with, rather than for, their children. At the same time, parents may feel uncertain about their child's decision-making skills compared to their own. Some parents have expressed anxiety about how their children will learn to make sophisticated, well thought out, difficult decisions for themselves. Of equal concern for parents is whether their child will continue to include them as an integral component of their medical care. Taking care of a child's illness is a job that no parent ever wants in the first place, but alternatively, once proficient at it, it is not a job that many parents want to give up.

Additional guidelines for children can be found in Chapter 25, Routine Healthcare for Children with Telomere Biology Disorders.

Adolescents

As children with DC/TBD mature towards adulthood and begin to take responsibility for their actions, the concomitant challenges of age-appropriate development (complete with inappropriate choices) do not evade them. Adolescents have the capacity to understand DC/TBD in greater depth and may need assistance as they work to integrate this knowledge into their daily lives. For adolescents, challenging "the system" is age-appropriate and functional at times, facilitating emotional growth and allowing them to assert themselves as individuals. It can be expected that even those with the mildest of temperaments may rebel against the "rules" of DC/TBD. Adherence to medical regimens may be incomplete and should be given particular attention at this age. Risk-taking behaviors that relate to peer pressure, including illicit drugs, alcohol use, and sexual activity are all components of the adolescent's developmental landscape.

Additional guidelines for adolescents can be found in Chapter 26, Transitioning from Pediatric to Adult Medical Care.

Growing to and Through Adulthood

Many individuals with DC/TBD, who have lived with the knowledge of their illness since childhood, will continue to make decisions as young adults in collaboration with their parents. Having grown up in a “medical partnership” with their parents, they have grown accustomed to having them involved in their medical care. Growth for the individual with DC/TBD can also become a time of growth for other family members. Parents will sometimes need assistance in strengthening their skills at enabling their “aging” children to become responsible for their own care. In turn, parents are crucial in educating and empowering their children, while learning to trust them and their judgment. As with growth in all facets of life, there can be occasional dissonance between parents and children living with DC/TBD. Ultimately, parents will need to learn to support and appreciate their grown children’s choices. Newly emerging adults with DC/TBD will need to learn to trust and engage their families at times of crisis and when they need assistance.

Young adults may find themselves torn between the desire to be proactive about their health and their desire to fit in socially. This struggle can be exacerbated by the complexities of their emotional journey with DC/TBD, combined with a sense that life may be on an accelerated path. As those with DC/TBD age and medical problems emerge, groundwork set in earlier years will encourage them to rely on health care providers for treatment and support.

Finding their own voices, taking responsibility for managing their own illness, becoming primary decision makers while using their parents as partners or consultants, and truly becoming independent, are appropriate and very significant steps for young adults. It is important to help individuals with DC/TBD gain their independence, while helping them understand that they can still rely on their families for information, support, assistance, and guidance. Medical partnerships with parents should be well established before children age out of pediatric care.

Becoming a young adult leads to a more comprehensive understanding of DC/TBD and new intellectual and emotional realizations. Issues that may have otherwise been dormant at other developmental stages will need to be addressed. Young adults who face the most severe manifestations of DC/TBD may, of necessity, remain more physically and emotionally dependent on family members. At each stage, issues of dependence and independence may need to be negotiated.

Becoming a young adult carries with it certain responsibilities for all individuals, even more so a person with DC/TBD. Becoming responsible for one's own medical care begins in earlier stages of development as an individual learns about DC/TBD. Being in charge of one's medical care is best seen as a partnership between the person living with the disease, members of that individual's support network, and the medical team. Taking care of oneself does not mean having to deal with DC/TBD alone.

Growing up with DC/TBD, establishing and mastering life goals, forming relationships and dealing with issues of partnership, sexuality, marriage, children, financial and insurance concerns, while managing a complex illness, organizing a variety of medical specialists in the interest of your care, and dealing with potential medical risks, present unique challenges for adults living with DC and their family members. Adults who have DC/TBD and are the parents of a child with DC/TBD have to negotiate their own medical issues and concerns as they anticipate and take care of their child's needs. In such situations, as children with DC/TBD mature, they will be exposed to the medical trajectory of their parents or even grandparents, leaving them wondering if they will experience a parallel illness course.

Who you are in the world is often amplified by the friendships you create. Whom do you tell that you have DC/TBD, and what and when do you tell them? These are complex issues, as they seem to be inherently related to those you trust, combined with an ongoing evaluation of the relevance of who needs to know, and your sense of what they will do with the information. Each individual must decide how they will incorporate DC/TBD into the structure of their lives. This issue can frame early stages of

relationships with friends, roommates, and romantic partners. When someone who has DC/TBD embarks on a relationship, questions about the nature of the illness, as well as the personal implications for the person with DC/TBD and for the partner, emerge. The revealing of DC/TBD, the short version, and then DC/TBD, the long version, becomes a component of the “dating” process.

Once in a relationship, partners of individuals with DC/TBD may need an outlet for information, expression, and assistance. Aspects of DC/TBD may be understood intellectually, but it is only when a partner’s condition worsens that some of the partner’s own concerns may emerge. Negotiating the caregiver roles of partners and parents for the person with DC/TBD presents an additional developmental and emotional challenge.

Avenues for Support

We had never met anyone with Dyskeratosis Congenita before. We both arrived scared and nervous and are leaving excited and hopeful.

— Parents after the experience of a DC family meeting

Team Telomere has compiled a vast array of resources to help individuals with DC/TBD and their families. The website, newsletter, monthly calendar, chats, educational meetings, fundraising activities, and other events that bring people in the community together are invaluable in sustaining and inspiring hope, joy, and a sense of being connected. They clearly enhance the lives and access to medical care for persons with DC/TBD.

Team Telomere, in collaboration with Camp Sunshine, has hosted biennial sessions for families of children with DC/TBD since 2010. Additionally, there are other opportunities created by Team Telomere to bring families together. All such programs serve as vehicles to help educate and support those with DC/TBD and their families.

Retreat-style, “campferences” or one-day family events blend educational sessions

presented by clinicians and researchers with psychosocial support and recreational activities have proven to be invaluable in empowering and inspiring hope in the lives of individuals with DC/TBD and their family members. This combination of education and the joy of community-based activities provides a successful formula to help meet the needs of families dealing with life-long illness.

Family Comments on Team Telomere Events

- *Camp Sunshine has given us hope for the first time in seven years.*
- *We are not alone.*
- *My son was transformed from being 'the boy with weird skin and nails' to being accepted and meeting two other kids his age.*
- *It gave us a directed path of information and help.*
- *We laughed for the first time in years, truly laughed.*

Siblings

Why did he inherit the DC? It could have as likely been me; the odds were the same.

—Sibling of a teen with DC

Siblings and sibling relationships are exceptionally significant, but may not always be the first priority in a family when a diagnosis of DC/TBD occurs, given the complex nature and the demands imposed by the illness and its treatment. Siblings of children with DC/TBD present their own unique concerns, some more and some less apparent. Siblings may feel guilty that their sibling was diagnosed and that they are healthy. In the case of genetic disorders like DC/TBD, these feelings are often further exaggerated. Non-affected siblings can experience complicated emotions at not having DC/TBD, fear and worry for their siblings, as well as normal sibling rivalry and then remorse at feeling jealous.

Siblings often use each other as reference points in life, defining themselves in relationship to their siblings. They see themselves in a comparative context: “I am one of three; I am the oldest; I have no brothers or sisters.” DC/TBD becomes another defining parameter in the relationship and is potentially even further complicated if there is more than one child in the family with DC/TBD. Sibling relationships can be among the strongest bonds in life, needing to be maintained and nurtured. It is important that affected and non-affected siblings have the opportunity to talk with their parents and with each other.

Specific Issues for Siblings, from a Team Telomere

Family Meeting

- Concern that all attention goes to the child with DC/TBD
- Feelings of neglect or isolation, less loved
- Never meeting or seeing others to relate to in day-to-day life (prior to regional or national gatherings)
- Guilt over not having DC/TBD
- Powerlessness, when a clear avenue of how to help is not apparent
- Wishing they could help more
- Needing to understand DC/TBD more
- Needing help in how to explain DC/TBD to their friends or peers
- Worry for the sibling
- Worry for the impact of DC/TBD on their own future family
- Not wanting to burden parents with their needs
- Wanting to be “good” so as not to create further issues in the family
- Feeling responsible for sibling and parent’s well-being

I was diagnosed after my brother’s autopsy results came back.

— DC patient and sibling

Siblings, whether affected or unaffected, worry about each other and themselves. Depending on the situation, siblings can exhibit emotional responses to the illness equal to or stronger than those who are affected. Anxiety is a dominant emotion experienced by children with DC/TBD and their siblings. Siblings, even understanding the magnitude of their sibling's illness, can still feel left out. Some may feel that they are less important to their parents because they do not have DC/TBD, or because they are not getting as much attention. Having such feelings, even if they are not verbalized, can cause a brother or sister to experience distress, so sometimes all of these emotions remain unspoken.

Open communication, education, and the opportunity to express and process experiences will enable siblings to find solace on the DC/TBD family journey. It is important to address unaffected children's feelings and questions, while including them in illness-related activities whenever possible. Siblings need their own time with parents, to have age-appropriate explanations of DC/TBD, to feel that their voices are heard, and to truly feel how integral they are to the family.

Recommendations for Siblings

- **Afford siblings a voice:** Create opportunities for siblings to meet others in the same situation to talk about common experiences, and to talk with family members
- **Create sibling time:** Simple time alone with siblings, valuing their time and your time with them. Examples could include going for ice cream, or even food shopping
- **Reassure siblings:** Siblings need to know and hear explicitly how much they are loved. It may not seem necessary, but it becomes very important when siblings see the attention that is given to their siblings with DC/TBD.
- **Educate siblings:** Help minimize confusion through education and communication.

- **Create communication:** Keep communication ongoing during complicated medical times, and in times of forced separation. Use social media, Zoom, FaceTime, texting, calling, writing (letters or leaving notes), journal entries (sharing a journal back and forth between parent and child, writing entries to each other creating a private, yet regular form of communication), etc.
- **Honor relationships:** Sibling relationships are unique and play a role in defining the identity of each child. Avoid asking the sibling to carry roles of caregiving outside of age-appropriate tasks.
- **Allow for emotions and their expression:** Fear, loneliness, neglect, jealousy, worry, depression, pride, independence, etc.

Ongoing Impact

This, too, is life. Gently try not to devalue the life you have by longing for another life.

– Adult with DC

In the context of a rare, progressive illness, there is often the concern about what will happen next. It can be difficult to live in the present when worrying about the future. Growing up with DC/TBD, you will inevitably be exposed to someone who dies as a complication of the illness, either in the greater DC community or in your own family. As the DC/TBD community becomes more cohesive and individuals become more connected, they and their families offer each other tremendous support, information, and hope. At the same time, additional exposure to loss and grief emerges. Creating rituals to honor the life of someone who dies from the same illness you have honors not only the individual, but the entire community.

Questions from Adults with DC/TBD

- What will the future bring?

- How can I plan?
- What types of careers can I pursue?
- How do I negotiate DC/TBD and a relationship?
- If I want a family, how should I proceed?
- How would I manage my DC and manage a family?
- How will I integrate my DC/TBD into my life, but not have it stop me from doing things I want to do?
- How do I live with the uncertainty of the future?
- What if my symptoms cannot be controlled?

Family Matters

We cannot thank you enough from the depth of a mother's heart – desperately trying to save her children and their daddy.

—Parent of children with DC

The diagnosis of DC/TBD has a strong impact on the family system. Unlike other illnesses, at times the diagnosis of one family member may lead to the diagnosis of another. At certain points families may find themselves making decisions about experimental procedures and protocols that have been utilized with very few patients. These and many other factors create a unique experience of vulnerability, as individuals are forced to recognize the rarity of DC/TBD and the frailty of life.

Should an individual with DC/TBD deteriorate and alternate treatment options be considered, the family may again be thrown into an emotional crisis. One answer to such an experience can come in the context of support from the Team Telomere community. Being informed, empowered, prepared to take appropriate action, and feeling supported are all critical components, strengthening and supporting the resilience of persons with DC/TBD and their families. The support and assistance of

this community can mitigate against the stress and loneliness of this experience and can help prevent a sense of being immobilized, helpless, or hopeless.

The medical course of DC/TBD and its treatment continues to evolve, allowing for the emotional and physical sequelae also to continue to evolve. At every point there is a balance, endeavoring to prepare for the future and potential next steps, while actively living in the present.

Patient-Caregiver-Physician Relationships

Relationships with physicians are of tremendous significance to families affected by DC/TBD. Finding a physician who has expertise in DC, or is willing to work in collaboration with such a specialist is critical. The quality of these relationships often influences the patient and family's entire experience of DC/TBD and their quality of life. Helping navigate the course of the illness, and thinking through decisions, can help those facing DC/TBD feel much less isolated and much more in control.

Individuals and their family members must truly strive to become experts about DC/TBD and its treatment. Engaging with researchers and physicians who have devoted significant portions of their professional lives in the pursuit of knowledge about DC/TBD improves and inspires the lives of those facing the illness. This connection among patients, researchers, and clinicians creates a hopeful paradigm, which is truly best medicine for patients and their families.

Guidelines for Patients, Caregivers, and Physicians

- An initial psychosocial assessment of an individual diagnosed with DC/TBD (and parents if the person is a child) can serve as a helpful tool.
- Referrals to appropriate counseling and other resources (e.g., Team Telomere, individual counseling, support groups).

- Encourage a dialogue among children or adults with DC/TBD or other bone marrow failure diseases (or other rare life-threatening illnesses) to minimize isolation and enhance self-esteem.
 - Encourage support group attendance for patients, parents, and siblings.
- Present information that is developmentally appropriate for individuals to enhance their understanding of and comfort with the diagnosis of DC/TBD.
- Encourage involvement with Team Telomere to help families develop and maintain an up-to-date knowledge base, to gain support, and to afford families an active role in supporting research. Encourage families to utilize Team Telomere resources:
 - Websites/webinars
 - Scientific sessions
 - Family meetings/retreats
 - Educational programs
 - Support of mentors
 - Group support
 - Team Telomere newsletters
 - Team Telomere and DC Facebook groups
 - Support from programs for persons with other rare illnesses, including other bone marrow failure and cancer predisposition syndromes
- Encourage families to create a working partnership with the physician/medical team.
- Encourage individuals with DC/TBD to become responsible and proactive with regard to their illness and medical care.
- Encourage adolescents and young adults to pursue their (academic, work, social) goals and dreams to prepare them for the transition to adulthood.
 - Help establish obtainable goals, with a next goal ever present.
- Encourage patients/family members to learn and stay abreast of salient treatment options.
- Encourage prevention and proactivity as they relate to illness manifestations.

- Work to make decisions with, and not for, families.
- Help the patient and family members to imagine the potential next illness manifestations.
- Help families adjust to living each day and focusing on activities apart from the illness as crucial components of day-to-day coping.
- Facilitate relationships and communication with DC/TBD professionals.
- Seek out specialists at times of decision-making.
- Establish relationships with counselors so they will be available at times of crisis.
- Join the Telomere Biology Disorder patient registry through RARE-X: <https://teamtelomere.org/resources/rare-x/>. Patient registries for rare diseases can be used to recruit patients for clinical trials, develop therapeutics, and better understand the patient population. Team Telomere is developing a secure patient registry to help identify DC/TBD patients around the globe and support research scientists and physicians in advancing knowledge of telomere disorders. Participation is optional but encouraged and can be done anonymously.

In rare, complex illnesses, such as DC/TBD, meetings with the treatment team can serve to educate and inform family members, garnering both practical and emotional support for the individuals facing DC/TBD. There is value in an ongoing dialogue with extended family members to discuss what is happening medically, and to create mutual support, as living with DC/TBD is truly a family endeavor.

Holding a broader family meeting (nuclear family members, and at times extended family members) with medical staff affords an opportunity for a wide range of issues and questions to be explored. Such meetings will exponentially increase family members' knowledge about DC/TBD, potentially increasing support that family members are able to provide to one another.

Traveling for Treatment

Because of the rare nature of DC/TBD, individuals often have to travel for evaluation and treatment with DC/TBD specialists in distant locations. Such medical travel is disruptive, challenging to organize, and can have enormous financial impact. Guidance from DC/TBD individuals and advocates who have traveled for treatment includes:

- **Researching and identifying centers:** Team Telomere maintains a network of physicians and clinicians at centers of excellence around the world who have specific expertise in DC/TBD, including genetic counselors, social workers, and specialists in organ and bone marrow transplantation in DC/TBD patients. Do not be afraid to ask your current medical care provider for referrals to other medical care providers. Several websites such as UNOS.org and bethematch.org provide statistics about the number and type of solid organ and bone marrow transplants being done at centers around the world. Refer to Chapter 30, Finding Clinical Trials for more information about researching clinical trials.
- **Contacting centers:** Although it can seem daunting at first, do not be afraid to reach out directly or via your medical care provider to any center in the world where you think your loved one might find treatment or support. Keep emailing and calling as necessary, and do not get discouraged. Be friendly, patient, and persistent. Ask about the option of a virtual or online consultation to minimize travel when possible.
- **Creating a resume:** As DC/TBD individuals go through their medical journey, they may accumulate hundreds of pages of test results and medical notes that can become challenging for even the most dedicated medical care providers to wade through. When communicating with new medical care providers, it can be helpful to summarize the key points of the patient into a brief “resume” or patient profile that can be shared in PDF or printed format. Key information could include: patient name, photo (head shot), contact information, gender, age, height and weight, blood type, known allergies, summary of current physical condition,

family status, medical history in bullet points, relevant family history, description of treatments to date, list of centers and physicians (with contact information) where care has been provided, most recent lab results, and treatment goal. Be sure to include a link to the Team Telomere Clinical Guidelines! The 5-page health summary templates in the Appendix of Chapter 26, Transitioning from Pediatric to Adult Medical Care, can be useful for this.

- **Talking to other families:** Through Team Telomere’s family chats, Facebook groups, and other forums, reach out to other patients and their family members. Compare notes about your experiences.
- **Working with your health insurance provider:** Talk to your insurance provider about what is covered if you travel outside your local area. Some health insurance providers can connect you with a specialized caseworker who can help you navigate and manage the costs of organ or bone marrow transplantation.
- **Reaching out for help from your network:** While some members of the family are traveling to distant medical centers, other family members may be left at home to continue their lives. Reach out to your support network to find help for those remaining at home, for example, asking a grandparent, aunt/uncle, or trusted friend to come stay with children left at home while parents are traveling with another child. As much as possible, keep in regular touch with family members at home, e.g., through text messaging, Zoom, FaceTime, and other electronic communications.
- **Reaching out for help at the center:** Large medical centers around the world often provide support services for patients who are traveling long distances to those centers for care. The range of services varies widely from center to center but can include such offerings as reduced-price, long-term housing for patients and caregivers, meal vouchers, free shower access, travel to and from the local airport, and translation services for international patients.

Conclusion

DC/TBD remains a difficult diagnosis to deliver and a complicated illness to live with. However, the connections within an affected family may run deeper than those of their healthy peers, as their experiences teach them a great deal about life. Parents of and those with DC/TBD sometimes describe having a greater appreciation for the things they do with their children, learning how to experience each day to its fullest and enjoying and valuing life. Patients, partners, and parents talk about being stronger than they realized and their ability to endure. The resilience exhibited by many of those affected with DC/TBD and their family members is truly remarkable; it reflects their ability to cope with this unique illness day-to-day. Given the complexity of family dynamics, the genetic components of DC/TBD, and the various roles and responsibilities that exist for affected individuals and family members, the support of Team Telomere and the commitment of professionals around the world united in advancing research and the treatment of DC/TBD, must be recognized as major components in enhancing the emotional well-being of those living with DC/TBD.

It is important for individuals to have the chance to tell their DC story; sharing one's own narrative leads to a personal place of healing.

Every time we talk to another DC patient or family, we learn. Even if our story is not exactly the same – we can all learn from each other's experiences. It's one of the most important parts of our community. It helped me to hear similar stories and feelings, especially in the beginning days.

This is our first time telling our story and everyone was able to understand and/or relate. I've never experienced that before. The value of telling our story is to bring awareness of this rare disease to help a new family who may be struggling. [It is about] empowering others, processing my emotions, forming connections... I love to tell our story. I find it empowering to be able to educate others and raise awareness... so others can understand or try to understand that we are in a battle with a disease that doesn't have a cure.

Talk, ask questions, take one step, the rest will follow. You might wobble and fall, but someone here will pick you up and help you take another step again.

—Compiled advice from members of the TBD community

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