

Chapter 31

What's on the Horizon: Research and Other Prospects

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Introduction

From coping with a rare disease diagnosis to navigating medical management and treatment, individuals with Telomere Biology Disorders (TBDs) and their family members may feel that they are facing a constant uphill battle. However, hope can be found knowing the advancements that have been made and that are on the horizon in the field of TBDs.

Science is moving faster than ever before, and medical knowledge has been expanding exponentially. It was estimated that the time it took to double medical knowledge in

1950 was 50 years. In 2020, medical knowledge was estimated to double in just 0.2 years – a mere 73 days [1].

This rapid progression of science is clearly evident in the field of TBDs. From when classic Dyskeratosis Congenita (DC) was first described in its most primitive form in the early 1900s to the present, countless advancements and discoveries have been made in the field of Telomere Biology Disorders: the elucidation of the underlying defects in telomere maintenance, the development of CLIA-certified flow FISH telomere length testing, the identification of TBD-related genes, the improved treatment of TBD symptoms and presentations, the initiation of TBD-specific clinical trials, and much more.

Essential to the field has been a better understanding of the range and spectrum of how TBDs present with symptoms in different individuals. Even at the start of the 21st century, TBDs were largely viewed as a pediatric bone marrow failure syndrome. However, it is now understood that individuals impacted by TBDs present with a range of physical presentations, with some individuals having no apparent physical symptoms of the disease and others displaying complex and serious problems in multiple parts of the body.

Around the globe, numerous clinicians, researchers, health professionals, and advocates are working towards the same goal: to better diagnose, manage, and treat TBDs.

Ultimately, each discovery and advancement is a piece of the puzzle. As the pieces come together, the future for people impacted by TBDs gets brighter.

Combining their perspectives, the authors contributing to this chapter have listed progress that individuals with TBDs, families, caregivers, clinicians, and scientists can expect in the coming years as our field continues to rapidly evolve.

For individuals, families, and caregivers impacted by TBDs, one can expect that

- The time to proper and accurate diagnosis will decrease
- The number of people diagnosed with TBDs will increase
- The track record for successful hematopoietic cell transplantation, lung transplantation, and liver transplantation at a wider number of medical institutions will improve
- Treatment options will increase and will be safer and more effective
- Improvements to treatments and continuing scientific advancements will not only lengthen lives but also increase quality of life
- TBD-specific prospective trials will increase; ongoing studies and patient registries will shine light on long term outcomes and disease progression
- Clinical trials evaluating novel therapeutic options that prevent end organ failure, supersede organ transplantation, and aim to target multiple body systems may begin
- Physicians and other health professionals will have increasing awareness of TBDs, leading to more clinical centers offering the specialized and multidisciplinary management and care that individuals with TBDs require
- Centers of Excellence will be developed, offering comprehensive multidisciplinary clinical management and care options to affected individuals and their families
- Access to community outreach will continue to increase, providing you education and guidance so you can become your (or your loved one's) best advocate and maintaining that *you are never alone*
- Awareness of how mental health impacts you and your family in the journey will increase
- Research will continue to be funded, and we will keep community needs at the forefront of clinical trials

For clinicians caring for individuals with TBDs, one can expect that

- TBD-specific experience across clinical disciplines and age ranges will continue to grow and appear in the published literature for reference
- Colleagues with specific expertise in TBDs will be increasingly identifiable and accessible for consultation and team care
- TBD-focused guidelines and clinical studies will define optimal preventive and interventional practices
- More children with TBDs will survive into adulthood, and more families with multiple generations at risk will be identified, requiring collaboration of adult and pediatric practitioners for transitional care and cross-referral

For scientists working to advance TBD science, one can expect that

- TBD-related genes and pathways will continue to be defined, yielding more opportunities to understand basic human telomere biology, and to define disease mechanisms and potential therapeutic targets
- TBDs will emerge as an ideal proving ground for the translation of cutting-edge scientific advances, including gene therapy, CRISPR/Cas9, and RNA medicine
- The forthcoming Team Telomere TBD research roadmaps and roundtables will identify critical unanswered questions in the field with the largest potential impact on patients and focus the research community's attention on developing effective treatments and cures
- Collaborative efforts between affected individuals, patient advocacy organizations, clinicians, and researchers – with patients and families at the heart of the discussions and work – will lead to exponential progress in basic and translational research

Team Telomere's vision is to see a world where every person impacted by Telomere Biology Disorders – including the affected individuals, caregivers, researchers, and clinicians – has accessible care, community, and resources, with the goal of positively

changing the course of this disease, driving toward improved treatments and ultimately one day a cure.

To see this vision to fulfillment, our mission is to provide information and support services to families worldwide affected by Telomere Biology Disorders, including Dyskeratosis Congenita, to encourage the medical community's research in finding causes and effective treatments, and to facilitate improved diagnosis by educating medical providers.

Our wholehearted commitment is to our community. We commit to working to find the path to accessible diagnosis and treatment. We commit to learning how to diversify science to be inclusive of all. We commit to working shoulder to shoulder with every stakeholder until a cure can not only be found but also become accessible to everyone affected by Telomere Biology Disorders. Our wish is that this promise brings hope to you and yours.

References

1. Densen P. Challenges and opportunities facing medical education. *Trans Am Clin Climatol Assoc.* 2011;122:48-58.