EARLY DETECTION AND DIAGNOSIS COMMITTEE

Purpose:
The Early Detection & Diagnosis (ED&D) Committee advances the TANGO2 Research Foundation's mission by promoting earlier recognition, accurate diagnosis, and timely access to care for individuals with TANGO2 Deficiency Disorder (TDD). The committee works collaboratively with families, clinicians, researchers, laboratories, advocacy organizations, and other stakeholders to reduce the diagnostic odyssey and improve outcomes through education, awareness, research, and strategic partnerships.

Committee Composition:
The committee consists of approximately **5–10 volunteer and staff members** representing diverse expertise and perspectives, which may include:

- Research Director (Committee co-lead)
- Foundation staff and Board members
- Parents and caregivers
- Adults living with TDD (when appropriate)
- Clinical and laboratory professionals
- Genetic counselors
- Researchers
- Rare disease advocacy leaders
- Public health or newborn screening professionals
- Other individuals with expertise or interest in early diagnosis and rare disease

Committee Roles and Responsibilities:
The committee serves in an advisory and collaborative capacity to support T2RF's early detection and diagnosis initiatives by:

Strategic Planning

- Develop and periodically update the Foundation's Early Detection & Diagnosis strategy.
- Establish annual priorities, goals, and measurable objectives.
- Recommend new initiatives that improve timely diagnosis of TDD.

Education & Awareness

- Develop educational resources for healthcare professionals, families, laboratories, and the public.
- Increase awareness of the signs and symptoms of TDD to promote earlier recognition.
- Support educational campaigns aligned with Rare Disease Day, Genetic Counselor Awareness Day, and other key awareness opportunities.
- Review educational materials to ensure scientific accuracy and accessibility.

Clinical & Diagnostic Initiatives

- Identify opportunities to improve diagnostic pathways and reduce barriers to diagnosis.
- Provide input on clinical practice resources and diagnostic guidance.
- Explore opportunities related to genomic testing, newborn screening, and other emerging diagnostic technologies.

- Monitor advances in rare disease diagnostics that may be relevant to TDD.

Partnerships & Collaboration

- Foster collaborations with clinicians, researchers, diagnostic laboratories, newborn screening programs, professional societies, and other rare disease organizations.
- Identify opportunities to participate in collaborative initiatives that advance early diagnosis.

Research & Data

- Advise on research priorities related to diagnosis, natural history, biomarkers, and implementation science.
- Review emerging evidence and recommend opportunities for T2RF involvement.
- Help identify knowledge gaps affecting diagnosis and referral.

Evaluation & Reporting

- Monitor progress toward committee goals.
- Report recommendations and activities to Foundation leadership.
- Evaluate the impact of committee initiatives and recommend improvements.

Committee Members Expectations:

- Attend and actively participate in committee meetings (minimum 80% attendance annually).
- Review meeting materials in advance and provide thoughtful feedback.
- Participate in committee projects and working groups as appropriate.
- Serve as ambassadors for the Foundation and its mission.
- Share expertise, experiences, and professional perspectives respectfully.
- Identify opportunities for collaboration, education, and outreach.
- Maintain confidentiality when discussing sensitive information.
- Disclose potential conflicts of interest.

Time Commitment:

- 2-4 hours per month (includes meeting, committee activities/tasks and outreach events)
- Appointment terms are annual, and volunteer members can be reappointed

For more information or to join the committee, please contact Deena Chisholm at deena.chisholm@tango2research.org