



## Collaboration Education and Test Translation Program

The CETT (Collaboration, Education and Test Translation) Program promotes the translation of rare disease genetic tests from research to clinical laboratories. This is achieved through collaborations among clinicians, laboratories, researchers and patient advocates.

### Researchers

By establishing a relationship with a Clinical Laboratory Improvement Amendments (CLIA)-approved laboratory and patient advocate group, researchers can:

- Focus resources on research questions, not on clinical care and service.
- Provide clinical results to their research participants, patients, families and referring clinicians in an approved regulatory manner.
- Identify potential new subjects from clinical testing for research participation with documented clinical findings.
- Fulfill their commitment to the research community by bringing research efforts to clinical practice.
- Ensure that the knowledge base obtained from the collection and storage of clinical data helps move research forward on the rare disease.

### Laboratory Directors

By establishing relationships with researchers and patient advocates, laboratories can:

- Provide quality testing and improve access to control materials.
- Obtain expert help on test result interpretation.
- Obtain expert help to develop appropriate educational materials for the public about the genetic test and appropriate referrals of individuals and families for genetic testing.
- Expand awareness of test availability to the clinical and patient communities.
- Collect and store genetic test information to increase the ability to interpret future test results.
- Work with patient advocates to increase knowledge about testing and understanding the research and clinical data.

### Patient Advocates

By establishing relationships with researchers and Clinical Laboratory Improvement Amendments (CLIA)-certified laboratories, patient advocates can:

- Provide their members and their families a quality diagnostic test for the rare condition.
- Increase access to all testing needs — diagnosis, carrier, prenatal and pre-implantation.
- Provide appropriate resources and educational materials to increase understanding of the rare disease and the genetic test.
- Develop a database of information on the genetic test that would help with future interpretation of the genetic test and clinical expression of the rare disease.



The CETT Program is an initiative developed and sponsored by the NIH Office of Rare Diseases.



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**The CETT team includes:**

- Giovanna Spinella, M.D., CETT Program Director, NIH ORD
- Andy Faucett, M.S., C.G.C., CETT Program Coordinator, Emory U
- Suzanne Hart, Ph.D., CETT Program Scientific Advisor, NIH NHGRI
- Roberta Pagon, M.D., CETT Program Review Board Coordinator, U Washington
- Lisa Forman, Ph.D., CETT Program Database Coordinator, NIH NLM NCBI
- William Gahl, M.D., Ph.D., CETT Program Biochemical Advisor, NIH NHGRI
- Kate Reed, M.S., C.G.C., CETT Program Education Coordinator, NCHPEG

*NIH- National Institutes of Health; ORD – Office of Rare Disease; Emory U – Emory University School of Medicine; NHGRI – National Human Genome Research Institute; U Washington – University of Washington; NLM – National Library of Medicine; NCBI – National Center for Biotechnology Information; NCHPEG – National Coalition for Health Professional Education in Genetics*

People with rare inherited diseases and their families need the reliable information that comes through quality genetic testing. While rapid progress is being made in discovering the genetic basis of disease, test development is not keeping pace. The CETT Program's Mission is:

- To promote the development of new genetic tests for rare diseases.
- To facilitate the translation of genetic tests from research laboratories to clinical practices.
- To establish collaborations and provide education about each rare genetic disease, related genetic research and the clinical impact of testing.
- To support the collection and storage of genetic test result information in publicly accessible databases to leverage the information into new research and new treatment possibilities.

❖ **Examples CETT tests:** CETT Program-reviewed tests include: Cornelia de Lange syndrome; Joubert Syndrome; Cherubism; X-linked Chondrodysplasia Punctata; Kallman Syndrome; Progressive Familial Intrahepatic Cholestasis; Russell Silver; MPS VI; Niemann Pick A/B; X-Linked Periventricular nodular heterotopia;; Primary Ciliary Dyskinesia; Infantile Neuroaxonal dystrophy; MADD; Arginase; Allan Herndon Dudley; 9q34 deletion; Epimerase GALE; PXE; Familial Focal Segmental Glomerulosclerosis; Arrhythmogenic Right Ventricular Cardiomyopathy; X-linked Recessive Brachytelephalangi Chondrodysplasia Punctata; Bilateral Frontoparietal Polymicrogyria; Autosomal Recessive Agammaglobulinemia; Urea Cycle Disorders – CPS1 & ASL, DMD micro-array, and AR primary microcephaly.

❖ **Participating laboratories include:** University of Chicago, Prevention Genetics, Toronto Sick Children, Gene DX, Emory, Harvard, UNC, Oregon HS, University of Colorado at Denver, UCLA and Correlagen.

## Frequently Asked Questions

**How do we get started?** We strongly encourage applicants to contact Andy Faucett, CETT Program Coordinator, at [info@CETTProgram.org](mailto:info@CETTProgram.org) or 404-727-4510.

**What if the advocate group and researcher are committed but don't have a laboratory?**

The CETT Program will work with you to find a laboratory. The six laboratories in the National Laboratory Network (NLN) for Rare Disease Genetic Testing are experienced clinical laboratories that are ready to work with CETT Program applicants and become active participants in a Collaborative Group, and the CETT Program has identified other laboratories that also are ready to be involved.

**What if the collaborative has a laboratory and a researcher group, but there is not an advocate group?**

The CETT Program has affiliations with the Genetic Alliance (GA) and the National Organization for Rare Disorders (NORD) to help CETT Program applicants. The CETT Program also has brought together a group of experienced patient advocates to act as mentors for each collaborative group.

**Who is responsible for collecting the clinical and genotype information?** Each Collaborative Group develops a collection and data storage plan that works best for the applicants. Contact the National Center for Biotechnology Information (NCBI) ([info@ncbi.nlm.nih.gov](mailto:info@ncbi.nlm.nih.gov)) early for assistance in this area. For each test the information that should be collected and that can be collected efficiently may be different. The CETT Program has developed a secure online data entry system as one option.

**How should the budget be developed?** The CETT Program Team uses a working budget of approximately \$1,000 per amplicon (one or more exons) as a guide for sequencing based tests. In addition, funds are available to support the development of educational materials. These figures include overhead and indirect costs. For other test methods, please contact The CETT Program. It is expected that the laboratory (institution) and/or the patient advocate group will contribute to the cost of test translation.

**Is there help available to assist with the development of educational materials?** The CETT Program Education Coordinator is available to help with developing educational materials at all stages. Examples of effective educational materials are available on the CETT Program website and from the CETT Program Education Coordinator.

**For more information, go to the [CETT Program Web site at www.cettprogram.org](http://www.cettprogram.org) or e-mail [info@cettprogram.org](mailto:info@cettprogram.org)**