# Collaboration Education and Test Translation Program

*www.cettprogram.org*

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NIH ORD CETT Program

# CETT Program Objectives

- To promote the development of new genetic tests for rare diseases.
- To facilitate the translation of genetic tests from research laboratories to clinical practices.

# CETT Program Objectives

- To establish collaborations and provide education about each rare genetic disease; related genetic research & 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.

# Guiding Philosophy

All parties benefit when:

- Quality of testing for rare disorders meets or **exceeds** existing standards
- Clinical laboratories, researchers, clinicians, and disease specific advocacy groups collaborate
- High-quality educational materials explain what the test can and cannot tell you and how best to use the test

# CETT Program Staff

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|---|---|
| ORD Program Director: | Giovanna Spinella, MD |
| Project Coordinator: | Andrew Faucett, MS |
| Scientific Advisor: | Suzanne Hart, PhD |
| Review Board Coordinator: | Roberta Pagon, MD |
| Education Coordinator: | Kate Reed, MPH, MS |
| NCBI Liaison: | Lisa Forman, PhD |
| Biochemical Advisor: | William Gahl, MD, PhD |

# The Process

1. Collaborative Group formed
   - CLIA certified laboratory
   - Researcher
   - Clinical consultant
   - Advocacy
   - (Genetic counselor found helpful)
2. CETTrack
   - Online application process
3. Contact with Education & Data staff
4. Intent to submit – 1st Friday
5. Submit – 2nd Friday (accepted monthly)
6. Staff Review – modified, returned or forwarded
   - Collaboration
   - Scientific merit
   - Lab qualifications
7. Review Board
   - 6 Member Team plus RB Coordinator
8. Evaluation with Suggestions & Requirements
9. Modified Testing Plan
10. Funding and Test Development

# CETT Test Update

- Success Summary May 2008
  - 42 Rare Diseases
  - 37 genes
  - 33 applications reviewed
  - 31 approved (1 partial)
  - 5 new CG working in CETTrack
- Test Development Summary
  - April 2008 – 25 tests available
  - Over 1,100 tests performed

# Modifications Since Inception

- Review Board added biochemical expertise (15 to 18)
- Application Revised
  - Two goals – Review information & questions to help Collaborative Group consider rare disease testing issues
- CETTrack – online collaboration building and application system
- Advocate Mentors
- Educational Materials Coordinator
  - From 3 formats to 2
- Funding – added support for researcher and collaboration building

# Tools Created & Shared

- Laboratory Guidelines
- Model Test Reports
- Educational Materials Guidelines
- Clinical and Mutation Data Collection System

# Research Collaborator Quotes

- R. H.- Arrhythmogenic Right Ventricular Cardiomyopathy
- *The collaboration has been extremely beneficial…For other researchers considering test translation, the involvement of a clinician, basic scientist interested in gene expression, research geneticist, clinical laboratory geneticist and a patient support group have all been beneficial.*
- I. K. – Cornelia de Lange
- *The diagnostic lab has also helped us in testing some families who needed more urgent screening…and where the families did not have resources to pay for the test. Communication…that has been mutually beneficial…Families in which a mutation has not been identified…are often referred to us for further research testing (this is ideal, as we do not need to burn through resources testing for genes available clinically on a research basis in the lab, but we are very interested in those families…)…It lifts a huge burden of responsibility off the investigators back and should be wholeheartedly pursued..*

# Lab Collaborator Quotes

- S. R. – Infantile Neuronal Axonal Dystrophy
- *We are completely in sync with the CETT philosophy and endorse CETT as a wonderful model for test translation… We also appreciate that CETT reviews our methods…makes helpful comments… improve the clinical testing that we offer.*
- P. R. - ARVC
- *The iterative approach for improving the applications seems to work very well….*
- K. C. – Primary Ciliary Dyskinesia
- *Working with patient advocates is extremely useful…*
- M. D. – CdLS,& Robinow
- *We have found working with the researchers and patient advocates very helpful. We now participate in the national patient advocacy meetings.*
- L. C. – Kallman, PXE, & X-linked chrondrodysplasia punctata
- *Yes, particularly when we find a VOUS. The input from the researchers has been extremely helpful in the interpretation of these variants..*

# Advocacy Collaborator Quotes

- . P. W. – NBIA Disorders – Infantile Neuronal Axonal Dystrophy
- *We have found the educational materials very beneficial.*
- M. M. – Primary Ciliary Dyskinesia
- *…we have been able to greatly increase patient awareness not just of the genetic test itself…this gave us the opportunity to provide practical education in basic genetics…it was our involvement in this program that made this vital education pertinent to our group and I think it has been key to helping the patient population understand the genetic nature of their disease….In 1991 I asked about …research… I was told it would never happen…My daughter's diagnosis was delayed by seven years…she had already sustained permanent damage to her lungs. We have people in our group who are not diagnosed until adulthood... The genetic test…will do more to encourage early diagnosis than our other educational efforts combined….*
- C. WV. – Niemann Pick
- *The most compelling outcome of participation in the CETT program is that prenatal diagnosis by molecular methods has become readily available to most families who chose it.*