Genetic Services Branch

Heartland RC Meeting
April 24, 2014

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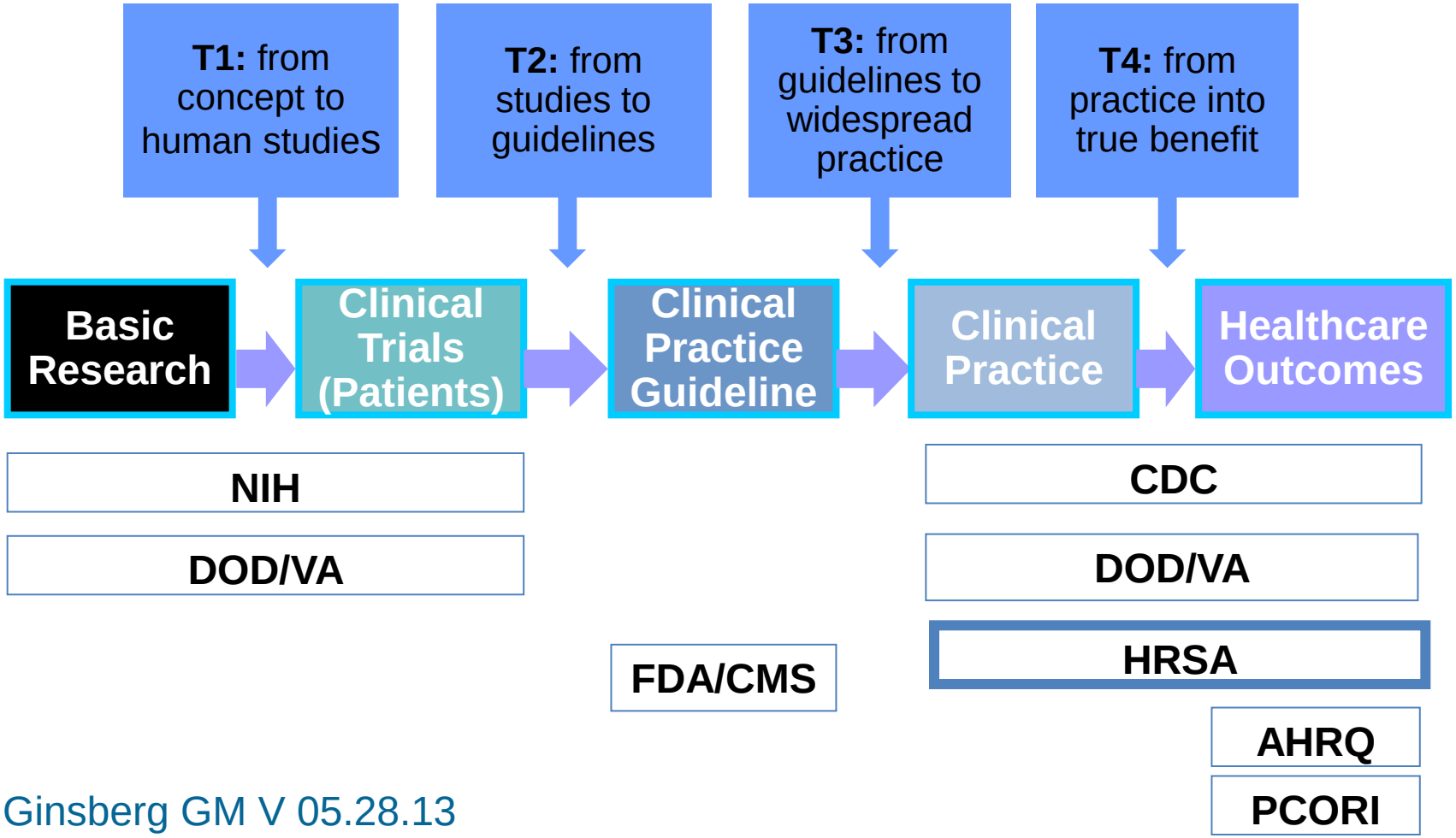
Chief, Genetics Services Branch
Division of Services for Children with Special Healthcare Needs
Maternal and Child Health Bureau
Health Resources and Services Administration
Department of Health and Human Services

Translating Genomics to Health

U.S. Department of Health and Human Services
HRSA
Health Resources and Services Administration

Roadmap of Federal Agencies Along the Translational Pathway



Ginsberg GM V 05.28.13

Health Resources and Services Administration (HRSA)

- America's healthcare safety net - HRSA is the primary Federal agency for improving access to health care services for people who are uninsured, isolated, or medically vulnerable.

Maternal and Child Health Bureau (MCHB)

- Provides leadership to improve the physical and mental health, safety and well-being of the maternal and child health population which includes all of the nation's women, infants, children, adolescents, and their families, including fathers and *children with special health care needs*.

Genetic Services Branch

Vision

Genetic science is intrinsic to the understanding of health and well-being throughout the life course of an individual.

Mission

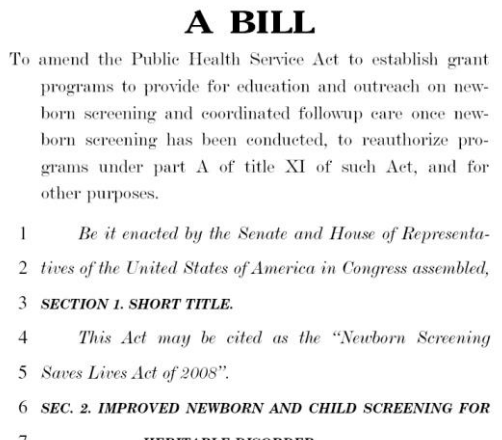
Provide national leadership to improve, expand, strengthen, and evaluate access to a system of genetic services and the quality of those services for children, youth, and adults across their life course.

Legislative Authority

- **Title V of the Social Security Act, Section 502(a)**: Authorizing funds for Special Projects of Regional and National Significance (SPRANS) for
  - *genetic disease testing, counseling, and information development and dissemination programs*
  - *comprehensive hemophilia diagnostic treatment centers without regard to age*
  - *the screening of newborns for sickle cell anemia, and other genetic disorders and follow-up services.*

Legislative Authority

- Title XXVI of Children's Health Act of 2000 "Screening for Heritable Disorders" enacted three sections of Public Health Service Act
  - Grant programs to improve ability of States to provide newborn screening and to evaluate effectiveness of screening, counseling or health care services
  - Established Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)
- Amended by Newborn Screening Saves Lives Act of 2008
  - Clearinghouse of newborn screening
  - Program on laboratory quality
  - Interagency Coordinating Committee
  - National contingency plan for newborn screening
  - The Hunter Kelly newborn screening research program



A BILL

To amend the Public Health Service Act to establish grant programs to provide for education and outreach on newborn screening and coordinated followup care once newborn screening has been conducted, to reauthorize programs under part A of title XI of such Act, and for other purposes.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

This Act may be cited as the "Newborn Screening Saves Lives Act of 2008".

SEC. 2. IMPROVED NEWBORN AND CHILD SCREENING FOR

Legislative Authority

Sickle Cell Disease Treatment Demonstration Program

- Section 712(c) of the American Jobs Creation Act of 2004, Public Law 108-357
- Build a model of service deliver to improve sickle cell care though;
  - Coordination of services
  - Genetic counseling and testing
  - Bundling of technical services
  - Training of health professional

HRSA's Inherited Blood Disorders Programs: Comprehensive Care Models

- The National Hemophilia Program
- Thalassemia Program
- Sickle Cell Newborn Screening Program

- Sickle Cell Treatment Demonstration Programs

Sickle Cell Newborn Screening Program

Sickle Cell Disease for Newborn Screening Program (6)

- Established in 2002
- Community-based networks partner with State Title V and state newborn screening programs, comprehensive sickle cell treatment centers, and other stakeholders to provide support to infants
- Work with the SCD National Coordinating Center to implement models of follow-up care for individuals with sickle cell diseases and trait

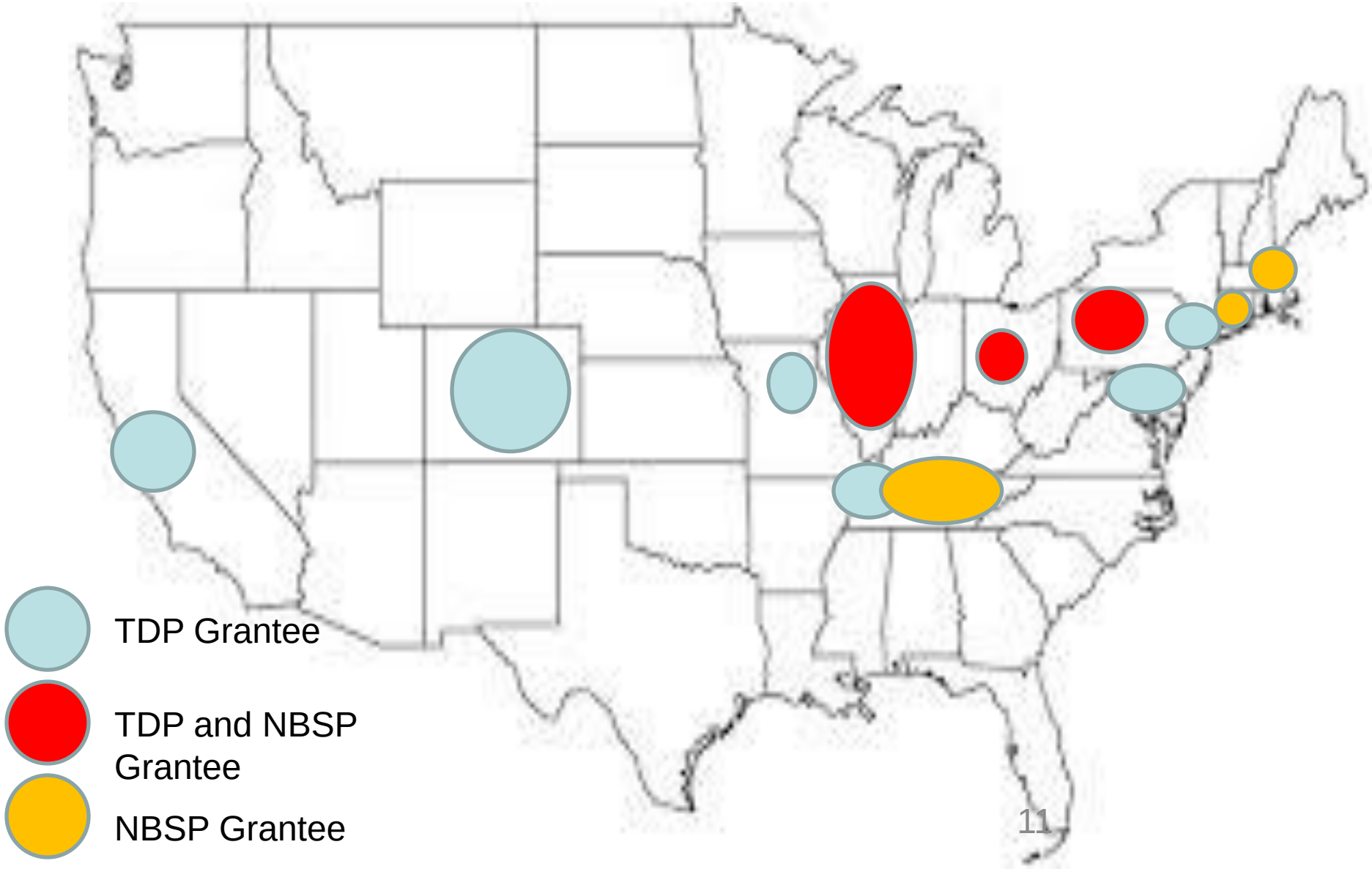
SCD National Coordinating Center

- Coordinates and supports grantee networks through technical assistance and information exchange.

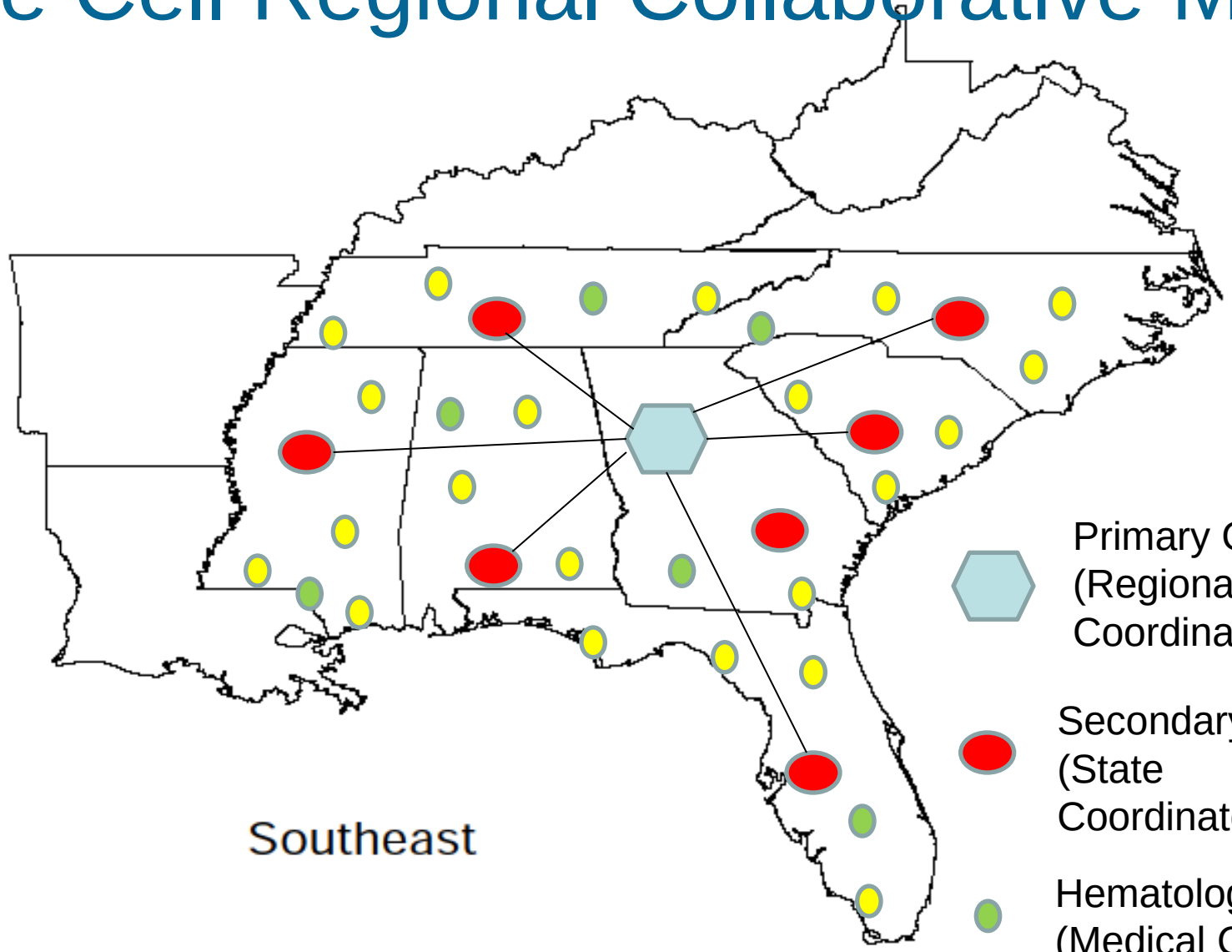
Sickle Cell Treatment Demonstration Program (8)

- Established in 2004 to improve:
  - access to disease-modifying therapies
  - patient and provider education
  - continuity and coordination of service delivery
- Grantee networks:
  - support the provision of coordinated, comprehensive, culturally competent and family-centered care
  - work collaboratively with non-federal partners
  - participate in quality improvement initiatives

Hemoglobinopathy Program



Sickle Cell Regional Collaborative Model



Heritable Disorders Program: GSB Newborn Screening Activities

- Staff the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC)
- Establish grants
  - Support state newborn screening programs
  - Increase awareness and provide education for broad range of stakeholders
  - Increase access to and coordinate services

DACHDNC

- Provides recommendations and advice to the Secretary
- Make systematic, evidence-based recommendations about conditions to add to the Recommend Uniform Screening Panel (RUSP)
  - Screening and diagnostic test
  - Availability of treatment
  - Harms and benefits of screening
  - Public health impact
- Consider ways to ensure that all states attain the capacity for screening and for short and long-term follow-up
- Quality assurance, oversight, evaluation, and education

DACHDNC – committee activities

- Make systematic, evidence-based recommendations about conditions to add to the Recommend Uniform Screening Panel (RUSP)
  - Screening and diagnostic test
  - Availability of treatment
  - Harms and benefits of screening
  - Public health impact
- Quality assurance, oversight, evaluation, and education
  - Timeliness of sample collection, transit, and reporting

New

New

Grant Programs to Support State NBS

- Critical Congenital Heart Disease (CCHD) Demonstration Program
- Long-term Follow-up Program: Demonstrate a plan for assessing the feasibility of establishing LTFU in a primary care setting

Grant Programs to Support State NBS

New

- **Clearinghouse of Newborn Screening Information**

  Aim: Increase awareness of newborn screening

New

- **Newborn Screening Data Repository & Technical Assistance Center**

  Aim: Measure performance of newborn screening

Regional Genetics Service Collaboratives (RCs)

Provide a regional infrastructure of public health genetics to improve, expand, and evaluate access to a system of genetic services and the quality of those services to improve health outcomes for children, youth and adults across their lifespan.

1. **NEGC: New England Genetics Collaborative**
2. **NYMAC: New York-Mid-Atlantic Consortium**
3. **SERC: Southeast Regional Collaborative**
4. **Region 4: Region 4 Genetics Collaborative**
5. **Heartland: Heartland Regional Genetics and Newborn Screening Collaborative**
6. **MSGRCC: Mountain States Genetics Regional Collaborative**
7. **WSGSC: Western States Genetic Services Collaborative**

Programs to Support Integration of Genomics into Clinical Practice

- Prenatal and Pediatric Family History Project
- Genetics and Primary Care Institute

Pregnancy & Pediatric Health Profile: A Screening and Risk Assessment Tool

- Helps the busy primary care provider translate family history data for clinical care
- Engages the patient as an active participant
- Provides a personalized clinical encounter with
  - → Clinical decision support
  - → Provider and patient materials
- Freeware

NCHPEG National Coalition for Health Professional Education in Genetics

Genetic Alliance

march of dimes

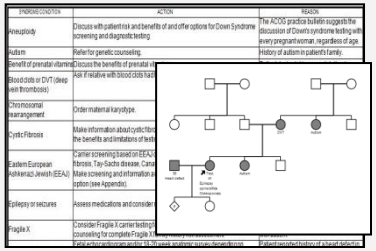
PARTNERS HEALTHCARE

U.S. Department of Health and Human Services HRSA Health Resources and Services Administration

How it Works

Waiting Room or Exam Room

Patient & Provider Meet



Patient completes e-form on Tablet, returns Tablet to front desk

Clinician interface & database

Clinician prints and reviews report and ed. materials

Clinician discusses recs with pt.

Pt receives targeted educational materials

Clinician documents encounter, uploads report into paper or e-record, orders tests/referrals

Clinician makes updates to input data as needed

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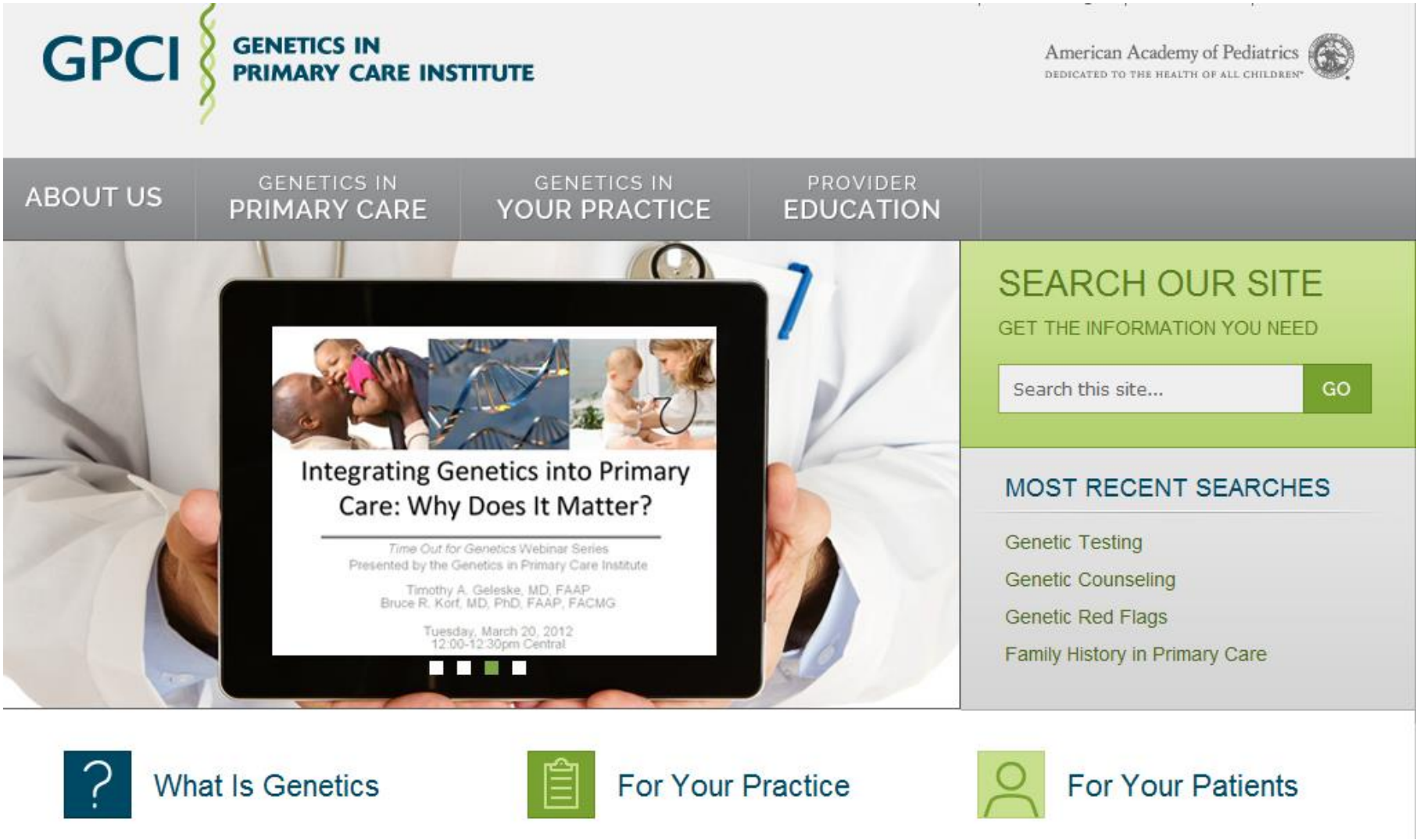
GPCI
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American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN
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GENETICS IN PRIMARY CARE
GENETICS IN YOUR PRACTICE
PROVIDER EDUCATION
Integrating Genetics into Primary Care: Why Does It Matter?
Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute
Timothy A. Geleske, MD, FAAP
Bruce R. Korf, MD, PhD, FAAP, FACMG
Tuesday, March 20, 2012
12:00-12:30pm Central
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