National Coordinating Center
for the Regional Genetic Service Collaboratives

NCC:
What's happening at the Coordinating Center

Heartland
October 2016

The NCC is funded by *U22MC24100*, awarded as a cooperative agreement between the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, and the American College of Medical Genetics and Genomics.

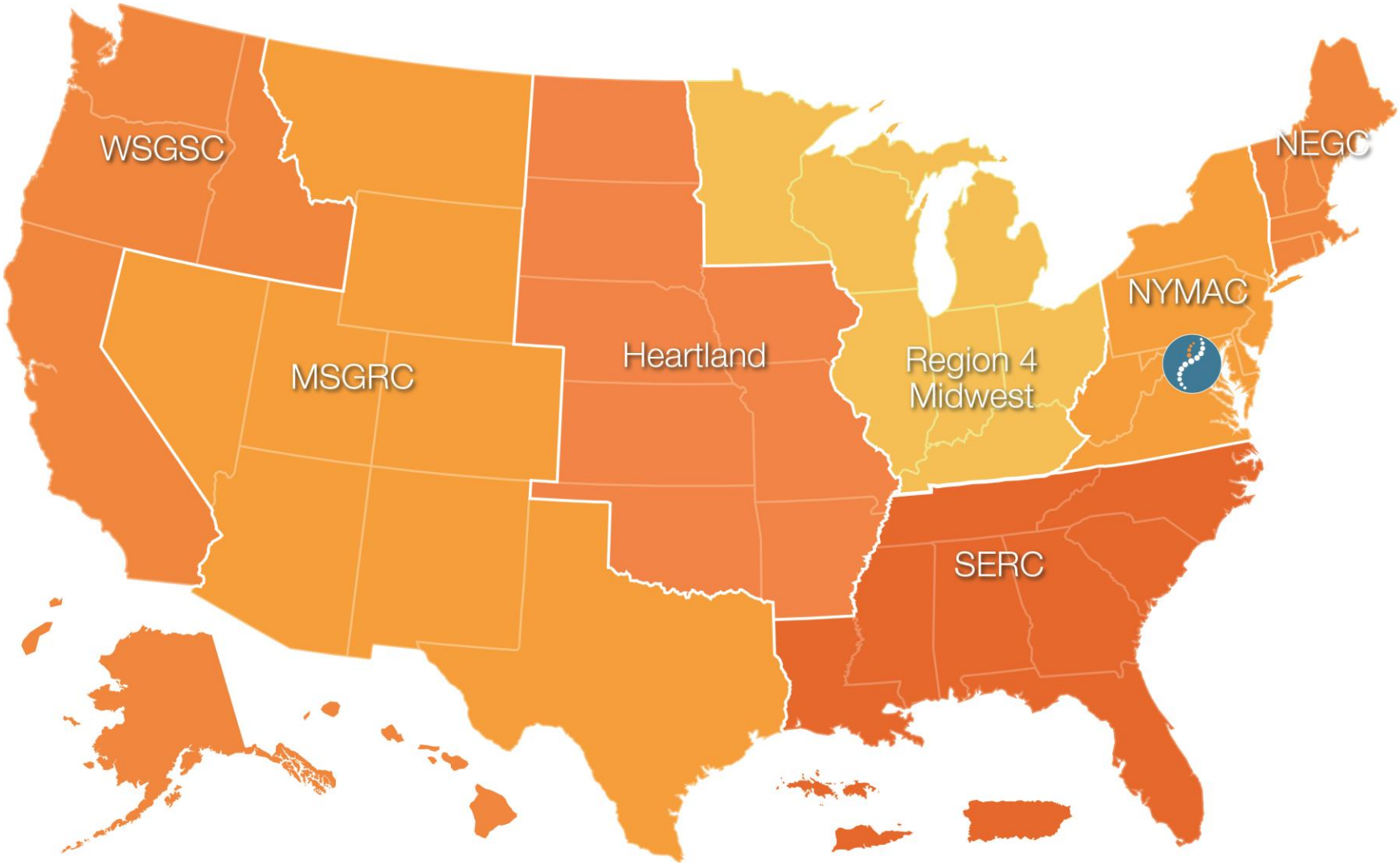
What I will cover today

- Overview of the NCC/RC System
- NCC Goals
- Workgroup and NCC Project Updates
- NGECN updates

What I won't cover:

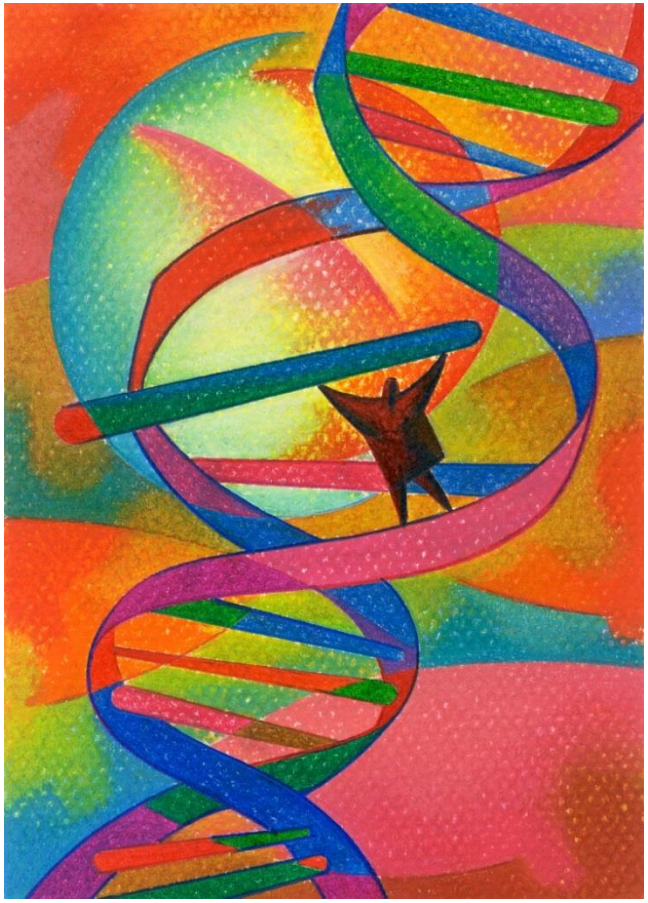
- The current FOAs that are out for the open competitions for the RGNs, the NGEFSC, and the NCC (HRSA 17-082, 17-083, 17-084)

NCC/RC System Today



NCC June 1, 2015 –May 31, 2017 (NCC Funding Period)

- **3 Goals of NCC:**
  - Develop a Framework for regional genetic support service centers models (RSSM) that reflect the needs of diverse regions;
  - Provide an infrastructure that strengthens communication and collaboration between the RCs, offers technical and clinical expertise as needed, promotes and disseminates outcomes of RC activities of national significance, facilitates partnerships with federal and non-federal entities, and evaluates the impact of RC activities; and
  - Implement a national genetics education and consumer network (NGECN) that provides access to genomic information and resources that cover the lifespan for consumers and the public.



Goal 1: RSSM Project.

Some of you may recall the listening sessions from our last meeting, they served as the foundation for the Regional Support Service Model work that the NCC conducted between August 2015 and March 2016.

Methodology: RSSM WG and AC

- August 2015--Convened a workgroup (met twice monthly) and advisory committee (met monthly)
- Considered existing recommended regional models (via listening sessions, literature review, and workgroup member suggestions)
- Reviewed national surveys to understand identified gaps
- Established priorities based on national surveys, RC input, and wg and ac.
- Develop recommendations for components of models to ensure access to genetic services for underserved populations.
- Draft recommendations out for comment
- Revisions based on comments
- Submission to HRSA, March 31, 2016

Recommendations: 8 Models

- RSSM Model Workgroup considered and developed a list of pros/cons for the following models:
  - Regional Genetic Service Resource Network (e.g. current infrastructure)
  - Regional Clinical Support Centers (workforce capacity)
  - Regional Genetics Education and TA centers (education focus, similar to ECHO)
  - Regional Patient Engagement Centers (Consumer-focused)
  - Public Health Model (address public health concerns, state public health partnership focus)
  - Quality Improvement Model (e.g. Pathways, PCORI-related)
  - Regional Clinical Support Network (e.g. hemophilia networks)
  - Genetic Service Data Centers (data, find out where patients are and when they get services, carefully selected information)

Key Message in Final Recommendations

In the executive summary, the brief addresses the following overarching principles:

1. Consumer engagement is a key element in the formation and functioning of future genetic service resource centers. Beyond participation in wgs and various projects, the level of consumer engagement should be within center top leadership; and
2. A significant level of uniformity between genetic service resource centers is essential if meaningful national outcome measures are to be identified. However, this required uniformity should not preclude innovation at the regional level, within the goals of the GSB.

Final Recommendations

- Recommended Genetic Services Support Model: Hybrid model (models 1, 2, and 3)—Regional Genetic Service Resource Network, Regional Clinical Support Center, and Regional Genetics Education and Technical Assistance Centers.
  - This model would have a primary focus on promotion of efficient practice within genetic centers (technical assistance [TA] for telegenetics, TA for authorization of genetic testing and other genetic services, TA for genetic counselor licensure and billing; other activities as identified through interaction with genetics providers); and aggressive and targeted support of non-genetics providers through promotion of other tools to enhance the level of care that can be delivered in conjunction with, but outside of, the genetics center itself.

RSSM Project Products (so far)

- RSSM brief available at www.nccrcg.org
- Working on a publication, aiming for January 2017 journal submission.

Other products from this process:

- Listening sessions: over 250 participants across all genetics stakeholder groups
- 2 national needs assessments, genetics provider (included public health and primary care providers) and consumers—more than 2,000 respondents over both; 925 to the provider assessment, 1231 to the consumer assessment; and
- Publications and national meeting presentations from the needs assessments and the RSSM process

NCC Workgroup Updates

- Evaluation Workgroup:
  - Year 4 data collected and brief available on the NCC website.

- Care Coordination
  - Excellent Care Coordination Whitepaper published on NCC website April 2016
  - Group has developed a driver diagram to help define a list of possible outcomes measures for genetic services care coordination, and explore external funding or article development.

NCC Workgroup Updates (2)

- Telegenetics:
  - Small subgroup is developing better outcome measures and will be sharing those in the coming months;
  - The workgroup encourages all RCs to have projects or develop projects in the coming months around telemedicine and genetics.
- Healthcare Access and Financing
  - Small groups (consumers, coverage of genetic services, genetic services in integrated delivery systems) are meeting and conducting small projects as solutions/further exploration of the issues identified at the June 2015 in-person meeting.
  - Panel 1- definitions and paying for services. Panel 2- Medicaid policies. Panel 3- effectively communicate for coverage –each project substantial, working to scale to what is achievable between now and May 31, 2017.

NCC Workgroup Updates (3)

- ACT Sheets:
  - Finalizing LSDs ACT sheets (revised) and (new) algorithms.
  - Finalizing secondary findings ACT Sheets
  - Working with a larger ACMG committee on Non-invasive prenatal screening ACT Sheets (as part of the guidelines process that committee is using)
  - Developing a pcp education ACT Sheet with GA.
- Long-Term Follow-up (LTFU)
  - NCC/RC NBSTRN LPDR Public health Questions and Data Elements Project
  - Collaborating with APHL

NCC Other Activities

- Advocate Leader program 2017
  - Exploring possibility of a leader training partnership with Family Voices in addition to advocate leader “regular” program with genetic counselor student mentors.
  - RCs will support 1-2 participants for a total of 10-14 participants. Heartland is supporting second year genetic counseling student mentors.

- Community Conversation 2017: Phoenix!!!
  - Topic: Reaching Minority Populations: Genetics in the Frontiers
  - Usually held Tuesday evening 7-9 pm
  - Open to the public, not just meeting attendees
  - Open forum that brings a topic of interest within this region to the ACMG meeting attendees

NGECN Updates
Year 1 & 2

By end of Year 1

- Monograph on patient navigators for individuals w/ complex care needs
  - Models, considerations (payment/reimbursement, evaluation, etc.), recommendations for the field
- Journal article on national needs of consumers related to information, referral, and diagnosis (~May 2016)
- Report on cultural considerations and strategies for genetic services outreach to high risk/insular populations
  - Amish, African-American, Ashkenazi Jews, Southeast Asian, Persian (Iranian) Jews

Continuing Activities

- GA Family Advocates Listserv
  - Quarterly calls, relevant events/updates
  - Sign up at: geneticalliance.org/about/mailinglist
- Technical Assistance for RC Consumer Groups
  - **E.G.** How to involve more consumers remotely?
- Promote tools and resources developed by RCs

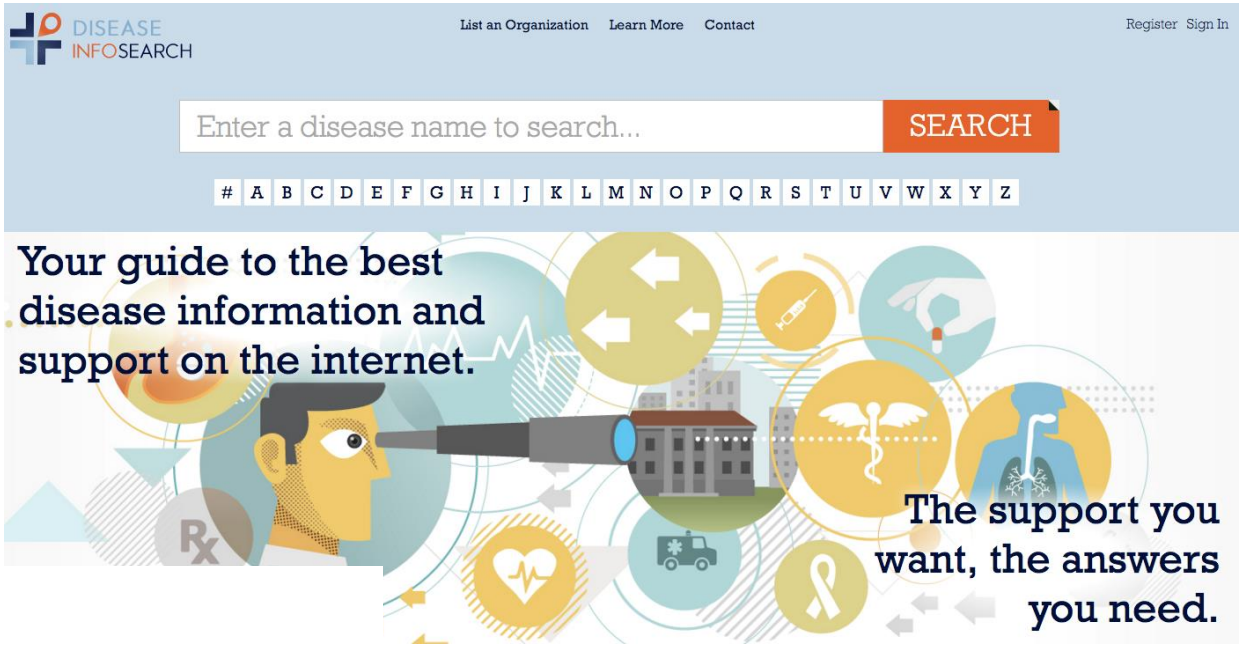
Year 2 Activities

- Services portal in DiseaseInfoSearch.org
  - What information is most helpful to individuals to make decisions about (where/from whom) specialty care?
- Outreach to Federally Qualified Health Centers, other healthcare institutions, EHRs
  - Delivering patient-centered materials on genetics and health for information at point-of-care
  - Embedding accessible information in patient portals, EHRs

Coming Soon!

New and Improved! DiseaseInfoSearch.org

Overview	Trusted Sites Resources
Find Support	23 Support Groups
News & Events	30 News Feeds Events
Clinical Trials	Open Studies
Publications	19 Editorial Articles 1.04K Research Articles 172 Review Articles
Participate	How do you compare to others with this disease?



Fabry Disease

Anderson-Fabry Disease | Ceramide Trihexosidosis

Overview

ADD UPDATE

Type of Disease: Not available

Fabry disease is an inherited disorder that results from the buildup of a particular type of fat in the body's cells and affects many parts of the body. Signs and symptoms may include episodes of pain, particularly in the hands and feet (acroparesthesias); clusters of small, dark red spots on the skin called angiokeratomas; a decreased ability to sweat (hypohidrosis); cloudiness of the front part of the eye (corneal opacity); and hearing loss. Potentially severe complications can include progressive kidney damage, heart attack, and stroke. Milder forms of the disorder may appear later in life and affect only the heart or kidneys. Fabry disease is caused by mutations in the GLA gene and is inherited in an X-linked manner. Treatment may include enzyme replacement therapy (ERT); pain medications, ACE inhibitors; and chronic hemodialysis or renal transplantation for end stage renal disease. Source: Genetic and Rare Diseases Information Center (GARD), supported by ORDR-NCATS and NHGRI.

Thank you!

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