Integrating genetics into primary care

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Winnipeg
CANADA
NORTH DAKOTA
International Falls
Thunder Bay
Upper Red Lake
Ely
Grand Forks
Blackduck
Bemidji
Cass Lake
Grand Rapids
Balsam Beach
Fargo
Duluth
Brainerd
WISCONSIN
Minneapolis/ St. Paul
SOUTH DAKOTA
Sioux Falls
LaCrosse
IOWA
11
59
89
72
71
1
2
38
53
61
200
10
371
169
64
35
29
90
94


BEMIDJI
PAUL
BUNYAN
1937

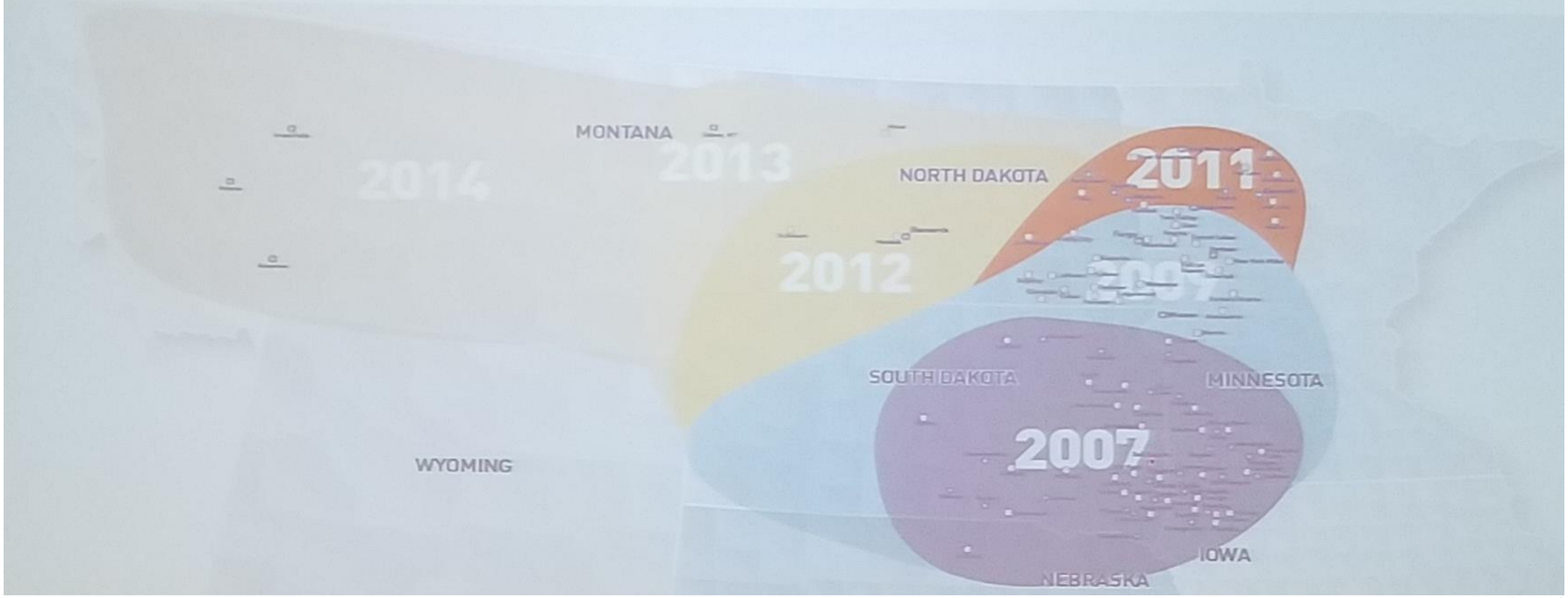
Welcome to Bemidji

- Bemidji, the First City on the Mississippi River, offers a relaxed, welcoming atmosphere with the amenities of a big city.
- Located in north central Minnesota, Bemidji is situated at the intersection of US Highway 71 and US Highway 2, approximately 230 miles northwest of the Minneapolis / St. Paul metropolitan area.
- Bemidji is the county seat of Beltrami County, an estimated **144,000** residents live within 75 miles of the community. **13,431** residents live within city limits.
- In addition, the Bemidji Regional Airport is the fourth busiest regional airport in the state, handling nearly 30,000 passengers annually. It includes daily service to MSP.
- Bemidji's local communications coop is nationally recognized as having one of the best telecommunications and broadband infrastructures in the country.

SANFORD™
HEALTH

Sanford Health

- Rural nonprofit health care system
  - Serves >2.3 million people in 132 communities
  - Distributed over 260,000 square miles in 9 states



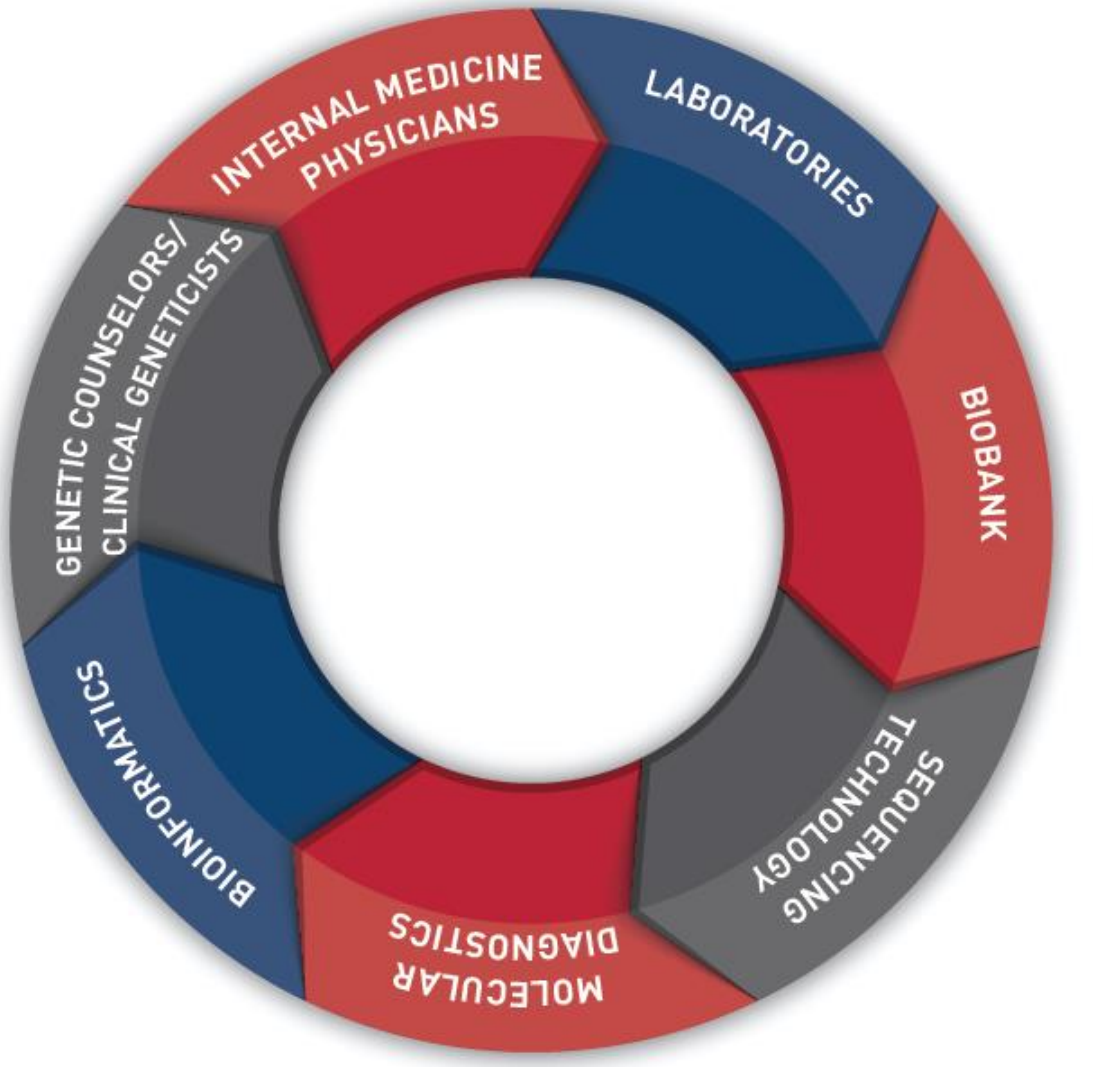
TEMP-0578

Sanford Imagenetics

- Launched in 2014, the program mission is to integrate genetics and genomic medicine into primary care for adult by:
  - Translating each patient's unique genetic profile for more precise treatment.
  - Recruiting the best and brightest in the fields of genetics and internal medicine.
  - Partnering with local universities to train the next generation of nurses, doctors and scientists in genomics medicine.
  - Accelerating scientific discovery of how DNA affects health and healing and apply that understanding to benefit patients.

What Sets Imagenetics Apart?

COMPREHENSIVE CAPABILITIES IN GENETICS AND GENOMIC MEDICINE



Not the only ones… ACS too!

- Given the lack of benefit concurrent with the increase in false-positive rates, CBE is not recommended for breast cancer screening among average risk, asymptomatic women at any age.
- Recognizing the time constraints in a typical clinic visit, clinicians should use this time instead for ascertaining family history and counseling women regarding the importance of being alert to breast changes and the potential benefits, limitations, and harms of screening mammography.

Athena Breast Health Network

- Project Goal: to improve quality of breast care by combining clinical care, quality improvement and research
- Clinical Aim: to develop a breast health program that personalizes breast cancer prevention, screening and treatment for each individual
  - Comprehensive patient data is collected as part of the clinical care process via an electronic health questionnaire
  - Patients at high risk for breast cancer are identified using multiple validated models and guidelines
  - Women with a significantly elevated risk of breast cancer are offered prevention counseling from an Athena Breast Health Specialist
  - Patients are given individualized support and referrals for tailored prevention and screening
  - Personalized risk reports and consultation summaries are provided to women and their physicians

SANFORD Edith Sanford BREAST CENTER

athena BREAST HEALTH NETWORK

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The NIH Undiagnosed Diseases Program: Insights into Rare Diseases

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Abstract

Go to:

Purpose

This report describes the NIH Undiagnosed Diseases Program (UDP), details the Program's application of genomic technology to establish diagnoses, and details the Program's success rate over its first two years.

Methods

Each accepted study participant was extensively phenotyped. A subset of participants and selected family members (29 patients and 78 unaffected family members) was subjected to an integrated set of genomic analyses including high-density SNP arrays and whole exome or genome analysis.

Results

Of 1191 medical records reviewed, 326 patients were accepted and 160 were admitted directly to the NIH Clinical Center on the UDP service. Of those, 47% were children, 55% were females, and 53% had neurological disorders. Diagnoses were reached on 39 participants (24%) on clinical, biochemical, pathological, or molecular grounds; 21 diagnoses involved rare or ultra-rare diseases. Three disorders were diagnosed based upon SNP array analysis and three others using WES and filtering of variants. Two new disorders were discovered. Analysis of the SNP-array study cohort revealed that large stretches of homozygosity were more common in affected participants relative to controls.

Conclusions

The NIH UDP addresses an unmet need, i.e., the diagnosis of patients with complex, multisystem disorders. It may serve as a model for the clinical application of emerging genomic technologies, and is providing insights into the characteristics of diseases that remain undiagnosed after extensive clinical workup.

WES

- Sanford Imagenetics will use whole exome sequencing to attempt to find a genetic cause for disease or disability in an individual.
- The integration of exome sequencing alongside traditional diagnostic testing will allow Sanford Health to further its mission to utilize translational medicine approaches to provide personalized care for each of our patients.

Getting the whole picture

- In many patients, the complex disease is multifactorial, either a known but difficult to diagnose condition or a new disease in which both genetics and environment play an important roles, and comprehensive and accurate patient characterization is critical to inform the genetic analysis that may lead to the answer.

Aim of the study

- The primary aim of this study is to identify causes of rare diseases in patients by performing comprehensive genetic, phenotypic, and exposure history characterizations of patients as a basis for understanding the underlying cause of these diseases.

Taking it a step further

- The objective of this study is to pilot test the integration of self-characterization (including self-phenotyping, and self-report of exposure (infection, environmental and lifestyle data) into the evaluation of patients with rare diseases.
- Using a self phenotyping questionnaire developed by Genome Connect (www.genomeconnect.org) as well as a newly developed self-report questionnaire for exposure data. This study will help determine if the self-characterization approach augments the diagnostic process.

Patient vs Provider

- We will also compare the self phenotyping with expert/clinician phenotyping. This will allow for better understanding of the data most useful in guiding genetic evaluation to lead to the identification of the causative gene or exposure in a patient with an undiagnosed disease, thus saving time and money in the long term.

Thank you!

"The genetic engineers gave him that birthmark as part of a sponsorship deal."