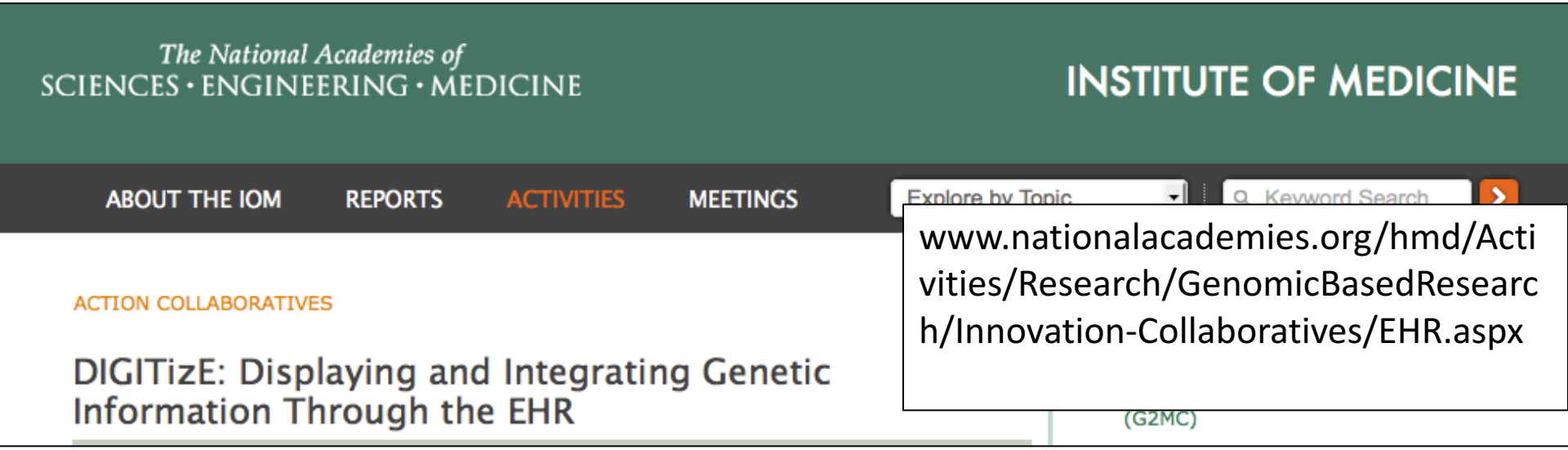
# Displaying and Integrating Genetic Information Through the EHR Action Collaborative

DIGITizE AC

# What is DIGITizE?

- Displaying and Integrating Genetic Information Through the EHR Action Collaborative



# Who is DIGITizE?

- Government
- Providers
- Laboratories
- Vendors
- Patients Representatives
- Standards Organizations

# Membership

- Sandy Aronson, Partners HealthCare
- J.D. Nolen, Cerner
- Mark Adams, Good Start Genetics
- Gil Alterovitz, Harvard Medical School
- Brian Anderson, athenahealth
- Jane Atkinson, NIDCR
- Larry Babb, Partners HealthCare
- Dixie Baker, Martin, Blanck and Associates
- Gillian Bell, Mission Health
- Adam Berger, HHS
- Colleen Campbell, University of Iowa
- Chris Chute, Johns Hopkins University
- Chris Coffin, Invitae
- Mauricio de Castro, Department of Defense
- Carol Edgington, McKesson
- Laurel Estabrooks, Soft Computer Corporation
- Robert Freimuth, Mayo Clinic
- Birgit Funke, Partners HealthCare
- Geoff Ginsburg, Duke University
- Jennifer Hall, University of Minnesota
- Stephanie Hallam, Good Start Genetics
- Heather Halvorson, U.S. Air Force
- Erin Hauenstein, Northrop Grumman
- Jonathan Hirsch, Syapse
- Gillian Hooker, NextGxDx
- Stan Huff, Intermountain Healthcare
- Kristen Janes, Kaiser Permanente
- Joan Kapusnik-Uner, First Databank
- Andrew Kasarskis, Mount Sinai
- Anthony Kerlavage, NCI
- Deborah Lange-Kuitse, McKesson
- Debra Leonard, University of Vermont
- Steve Lincoln, Invitae
- Ira Lubin, CDC
- Elaine Lyon, ARUP Laboratories
- Perry Mar, Partners eCare
- John Mattison, Kaiser Permanente
- Jennifer McKay, Avera
- Larry Meyer, VA
- Blackford Middleton, Vanderbilt
- Doug Moeller, McKesson
- Sean Mooney, University of Washington
- Massimo Morra, Personalis
- Scott Moss, Epic
- James O'Leary, Genetic Alliance
- Brian Pech, Kaiser Permanente
- Teji Rakhra-Burris, Duke University
- Priyadarshini Ravindran, Allscripts
- Mary Relling, St. Jude Children's Research Hospital
- George Robinson, First Databank
- Roberto Rocha, Partners eCare
- Wendy Rubinstein, NCBI
- Hoda Sayed-Friel, Meditech
- Megan Schmidt, Sunquest Information Systems
- Jud Schneider, NextGxDx
- Sam Shekar, Northrop Grumman
- Brian Shirts, University of Washington
- Brad Strock, Epic
- Jeff Struewing, NHGRI
- Jessie Tenenbaum, Duke University
- Charles Tuchinda, First Databank
- Christopher Vitale, Partners eCare
- Deepak Voora, Duke University
- Michael Watson, ACMG
- Scott Weiss, Partners HealthCare
- Jon White, ONC
- Bob Wildin, NHGRI
- Ken Wiley, NHGRI
- Kirk Wilhelmsen, UNC-Chapel Hill
- Marc Williams, Geisinger
- Grant Wood, Intermountain Healthcare

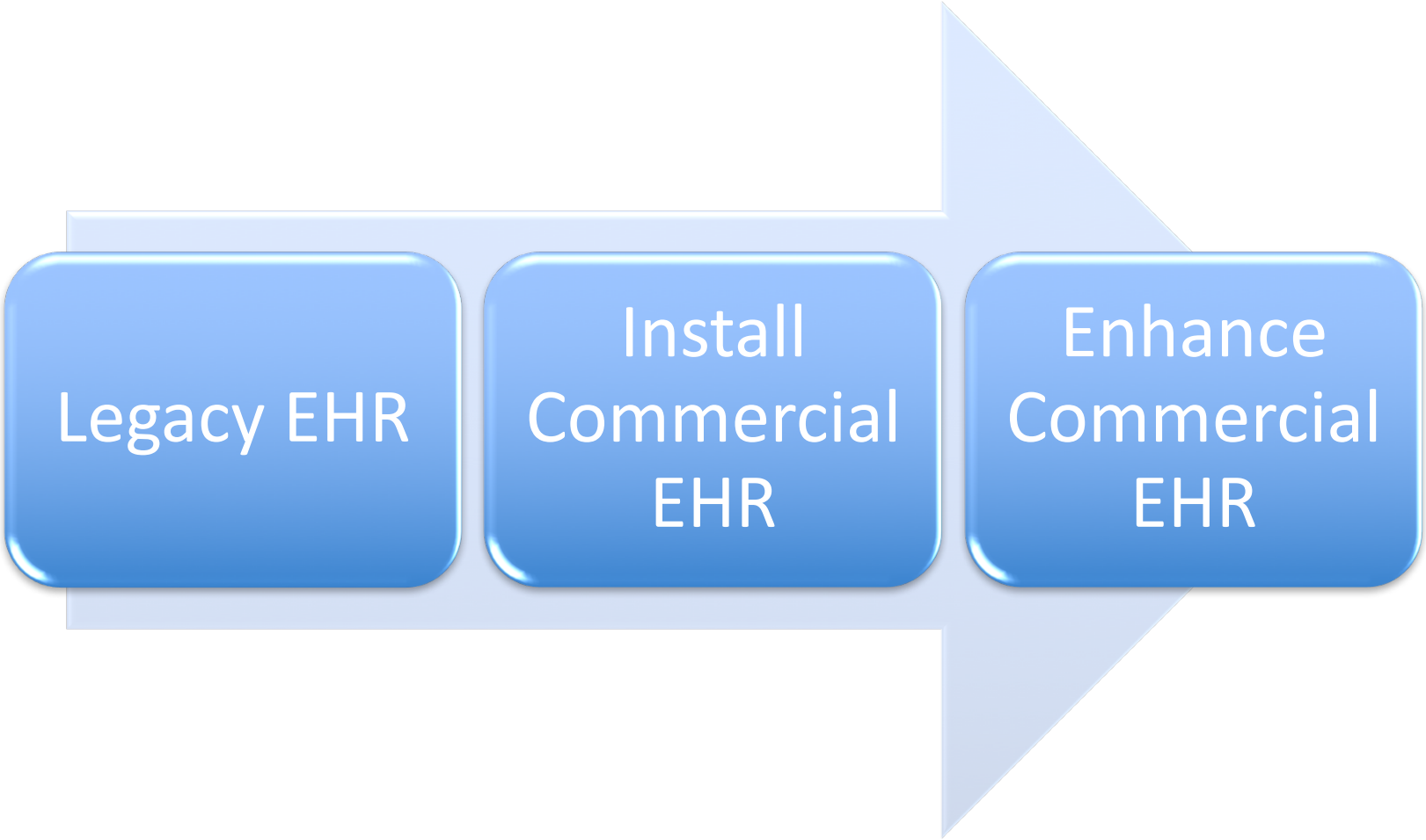
# DIGITizE's Purpose

Facilitate Development and Rollout of Genetic Aware Electronic Clinical Decision Support

# DIGITizE's Focus Areas

- Develop Implementation Guides
- Enable Collaborations to Piloting Guides

# Common Hospital IT Phases



# Developing Guides

# First Implementation Guide

- Pre and post test alerts for abacavir and azathioprine
- Required data movement from lab to provider
- Recommended provider CDS logic

# Establishing Connectivity and Pharmacogenor Decision Suppo Protect Patients HLA-B*57:01 ar Variants

An Implementation Guide

12/1/2015
Displaying and Integrating Genetic Information T
(DIGITizE AC)

Version 1.0

When applying this observation to an HL7v2 message, 50956-2:*HLA-B**57:01 represents the observation code that would populate the OBX-3 field of a single OBX segment returned to the ordering provider system from the testing laboratory system. The observation result value associated with this same OBX segment is found in the field OBX-5 which would contain one of two ordinal values, positive or negative.

> The SNOMED-CT codes for Positive and Negative are 10828004 and 260385009, respectively. SNOMED codes are strongly recommended for accurately and precisely representing the Positive or Negative result.

The reporting laboratory is to use this LOINC code to affirm that they tested for the specific *HLA-B**57:01 allele and one of the two SNOMED codes to convey they have observed that the patient's specimen either contains it or does not contain it.
If an OBX segment containing this LOINC code is not returned by a testing laboratory and stored in a manner that is available to the CDS algorithm then it cannot be assumed that the patient has been tested for the presence or absence of the *HLA-B**57:01 allele, which is directly related to the hypersensitivity of abacavir.
Here's a partial OBX segment example of the key elements discussed above:

For a positive finding of the *HLA-B*57:01* allele...
**OBX|1|...|50956-2^HLA-B*57:01^LN ||10828004^Positive^SCT~LA6576-8^Positive^LN-ANS |...**

And, for a negative finding of the *HLA-B*57:01* allele...
**OBX|1|...|50956-2^HLA-B*57:01^LN ||260385009^Negative^SCT~LA6577-6^Negative^LN-ANS |...**

> NOTE: The examples above includes the equivalent SNOMED-CT (SCT) and LOINC Answer (LN-ANS) codes in each OBX-5 field. While SNOMED-CT is the preferred standard, it does not prevent the ability to provide the equivalent answer code from the LOINC Answer list. This guide does specify a requirement to use the SNOMED-CT values so that the receiving system can fulfill the CDS rules associated with this test result.

# CSER – DIGITizE Collaboration

- Building Lynch Syndrome Guide
- Goal is to provide clinicians with proactive alerts to prompt screening colonoscopies for patients with Lynch Syndrome
- Key step is prompting clinicians to consider placing Lynch Syndrome on the problem list in the event a positive genetic result

# Key Learning

- Many individuals from many different backgrounds will invest their time to help
- But dedicated effort for project management, analysis and coordination still required
- Limited number of individuals who can play these roles

# Pilots

# Status

- Multiple organizations working towards pilots
- Significant interest in remaining engaged and sharing war stories through the process
- Still the realm of hospital IT – vendors including EHR vendors play a supporting role

# Lessons Learned

- Competition of Clinical IT Resources is Extreme
- Multi-phase approval processes
- Multiple levels of complexity in implementation

# Common Hospital IT Phases