

# A Collaborative Risk Assessment and Educational Tool for Health Professionals and the Public

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October 6, 2010

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Genetic Alliance

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# A COLLABORATIVE RISK ASSESSMENT AND EDUCATIONAL TOOL FOR HEALTH PROFESSIONALS AND THE PUBLIC



Emily Edelman, Bruce Lin, Brian Drohan  
Genetic Alliance Webinar



October 6, 2010

# Outline

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- Background
- Recent Progress
- Next Steps
- Demonstration

# Family History for Prenatal Providers

3

- Three-year cooperative agreement with HRSA (6/09 – 5/12)
  - NCHPEG
  - March of Dimes
  - Harvard Partners
  - Genetic Alliance
  
- Produce a family history and genetic screening tool for primary-care providers delivering prenatal care
  - Collects medical and FHx information
  - Clinical decision support & education
  - Point-of-care



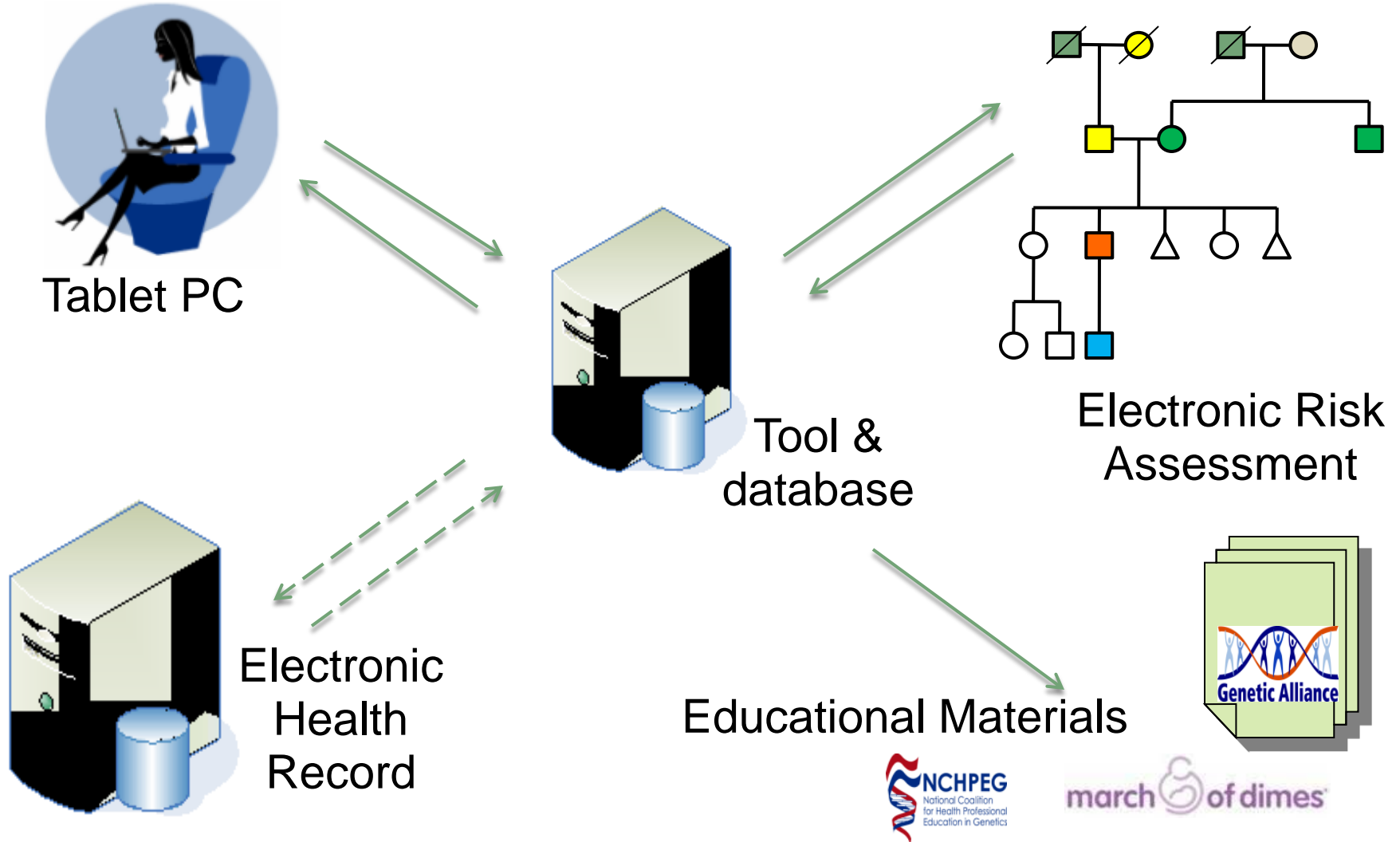
# Purpose

4

- Improve identification of women at risk for genetic disease
  - Tool is the centerpiece for provider education
  - Relevant patient education materials
  
- Provide “just in time” recommendations and support for ordering genetic tests, and management of genetic risk factors
  
- Save time for the clinician
  
- Streamline the intake process in the clinic
  
- Reduce the risk of clinician and institution liability
  
- Empower and educate patients

# “Prenatal Screening Assistant,” a.k.a. “Pregnancy and family history questionnaire”

5



# Core Elements of Tool

6

1. Patient demographics
2. Patient medical history
3. Pregnancy history
4. Newborn screening awareness & education
5. FOP demographics
6. Family History
  - Select common and rare disorders based on ACOG and ACMG recommendations
  - Genetic testing

# How did we come up with the questions on the tool?

7

- Detailed review of existing prenatal intake forms
- Literature review
- Professional society guidelines
  - ACOG, ACMG
- Expert opinion and consensus

## **In Summary**

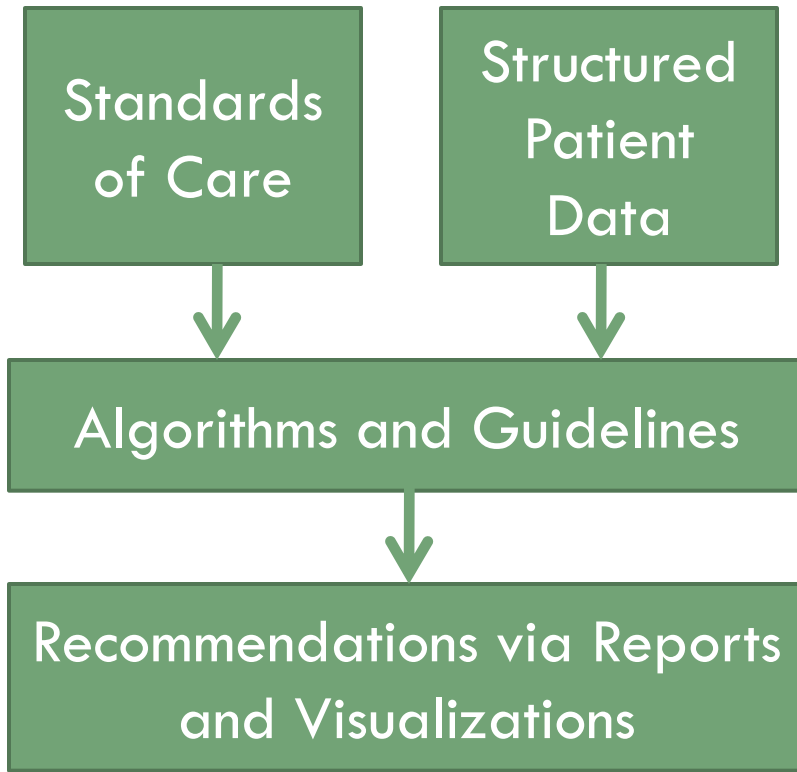
- Same as what a person is asked at an average visit
  - More emphasis on family history
  - A few unfamiliar terms and questions



# Clinical Decision Support

8

## □ **Specific responses trigger considerations for provider**



- **Ethnicity based risk**
- **Family Hx of syndrome / manifestation**
- **Personal Hx of syndrome / manifestation**
- **Pregnancy risk factors / exposures**
- **Previous genetic testing**

# Recent Progress

9

- 1<sup>st</sup> Advisory-Committee Meeting: October 2, 2009
- Beta version of tool is complete
- Formative evaluation underway
  - 15 women recruited from the general public
  - 20 prenatal providers from different disciplines and potential clinical sites
  - Results in mid-November

# Next Steps

10

- Complete formative evaluation
- 2<sup>nd</sup> Advisory-Committee Meeting: November 15 and 16, 2010
- Planned clinical implementation in March 2011
- Targeting clinical sites in Maine, Maryland, and California
- Provider education materials (Holly Tutko)

# DEMONSTRATION

# Pregnancy and Family History Questionnaire

## About You

Please select what best describes your race

We ask this question because it helps us assess your risk for certain diseases.

Click all that apply

- African American or Black
- Asian or Pacific Islander
- Caribbean or West Indian
- Caucasian or White
- Native American
- Prefer not to answer
- Not Sure

Back





Next

## Pregnancy and Family History Questionnaire

### Your Medical History

Do you have any or have you ever had any of the following diseases or conditions?

Click all that apply

- |                          |  |   |
|--------------------------|--|---|
| <input type="checkbox"/> | Diabetes   |  |
| <input type="checkbox"/> | Heart disease (sometimes called atherosclerosis) |   |
| <input type="checkbox"/> | Hypertension (high blood pressure)               |  |
| <input type="checkbox"/> | Blood clots or DVT (deep vein thrombosis)        |  |
| <input type="checkbox"/> | Pulmonary embolism (blood clot in the lung)      |  |

Back








Next

## Pregnancy and Family History Questionnaire

### Your Family History

Has anyone in your family ever had any of these conditions?

Click all that apply

<input type="checkbox"/>	Autism	
<input type="checkbox"/>	Sudden unexpected death in a child or adult	
<input type="checkbox"/>	Cystic fibrosis	
<input type="checkbox"/>	Fragile X syndrome	
<input type="checkbox"/>	Hemophilia or von Willebrand disease	
<input type="checkbox"/>	Sickle cell disease or sickle cell trait	
<input type="checkbox"/>	Spinal muscular atrophy	
<input type="checkbox"/>	Thalassemia	
<input type="checkbox"/>	Developmental delay or mental retardation	

Back

Next

## Pregnancy and Family History Questionnaire

### Family History

Have you, the father of the pregnancy, or anyone in either of your families had genetic testing?

 Yes Yes No Not sure



## Prenatal Survey Summary

v9-5 @work woman	DOB: 01/11/1975	Age: 35	Gender: F
Unit #: 99909101001	Provider:		09/13/2010

### CURRENT PREGNANCY INFORMATION

LMP	8/1/2010
LMP Confidence	No
Best Due Date (self report)	5/12/2011

### CONSIDERATIONS

#### For the Patient:

Syndrome/Condition	Action	Follow Up Care	Reason
Thalassemia	Check CBC, MCV	If MCV < 80, consider ordering hemoglobin electrophoresis and serum ferritin (to exclude iron deficiency anemia).	The patient reports an ethnicity of Caribbean or West Indian.
Sickle cell disease	Check CBC, MCV	If MCV < 80, consider ordering hemoglobin electrophoresis and serum ferritin	The patient reports an ethnicity of Caribbean or West Indian.

# DISCUSSION

# Partners

18

## NCHPEG

- Joan Scott
- Emily Edelman

## Harvard Partners/ NWH/MGH

- Kevin Hughes
- Brian Drohan
- John Sharko
- Adam Wolfberg

## Genetic Alliance

- James O'Leary
- Vaughn Edelson

## HRSA

- Alaina Harris
- Michele Puryear

## March of Dimes

- Siobhan Dolan
- Bruce Lin



# THANK YOU!

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