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A Strategy for Screening for Birth Defects and Genetic Disorders

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Primary Care Physicians and Genetics

- "we recommend that attempts to educate PCPs in genetics…be linked to evaluation and outcomes, considering the impact on practice as well as knowledge acquired." Greendale and Pyeritz, 2001
- 39% of surveyed ACOG members considered genetics as ***least important*** among a list of priority issues Wilkins-Haug, 2000

Primary Care Physicians and Genetics

- "to utilize genetic medicine optimally requires an educated health professional workforce." Guttmacher, Jenkins, and Uhlmann, 2001
- barriers to PCP provision of genetic services are inadequate knowledge of basic genetics, lack of updated family histories, lack of confidence, and lack of referral guidelines Suther and Goodson, 2003

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Prenatal assessment/ genetic evaluation

Origins of ***First*** **PAGE**

- 1996 - SPRANS grant funds development of *ProgramME*
  - **P**rimary **O**B **G**enetic **R**isk **A**ssessment and **M**anagement in **M**ain**E**
    - Linda Bradley, Ph.D.
  - Subsequently utilized in a variety of other venues

Genetic History Questionnaire (GHQ)

- Designed to be completed by patients or administered by office staff
- 15 element questionnaire
- Each question is a *screening test* to identify risk for a major fetal disorder or condition for which a course of action is available

Genetic Risk Screening Office Guide

- Designed to function as both a clinical practice and educational tool
- 15 tabbed sections correspond to the questions on the GHQ
- A brief *secondary questionnaire* designed to elicit key information about family history, previous testing, etc.

Guidelines for assessing risk

General recommendations for follow-up, including when referral may be appropriate

Information on screening/diagnostic testing

March of Dimes Fact Sheets for patients

Community resources and local/national support groups

Brief description of the disorder with references

Related ACOG information

ProgramME Distribution

- Distributed in 1996 to 212 Maine PCPS
  - OB
  - FP
  - CNM
  - NP
  - PHN

Evaluation of Satisfaction

28 question evaluation form

(55% of practices responded)

- 85% of respondents used GHQ with all new prenatal patients
  - 91% at 1st prenatal visit
- 7 minutes to complete
  - 73% had patient complete GHQ
- 4.4 (out of 5) rating for satisfaction

Evaluation

- *ProgramME did not* affect
  - Number, type, or outcome of calls to 2 genetics centers
- *ProgramME did* affect referral indication
  - Family history ↑ from 13.2% to 27.4% ($p<0.01$)
  - Maternal indication ↑ 9.9% to 19.5% ($p=0.03$)

Development of *First* PAGE

- Revision and distribution of materials was funded by a grant from the March of Dimes Mission Investment Opportunity Program
  - Survey original *ProgramME* users
  - Content review and update
  - Modify format

2002 Survey of *ProgramME* Users

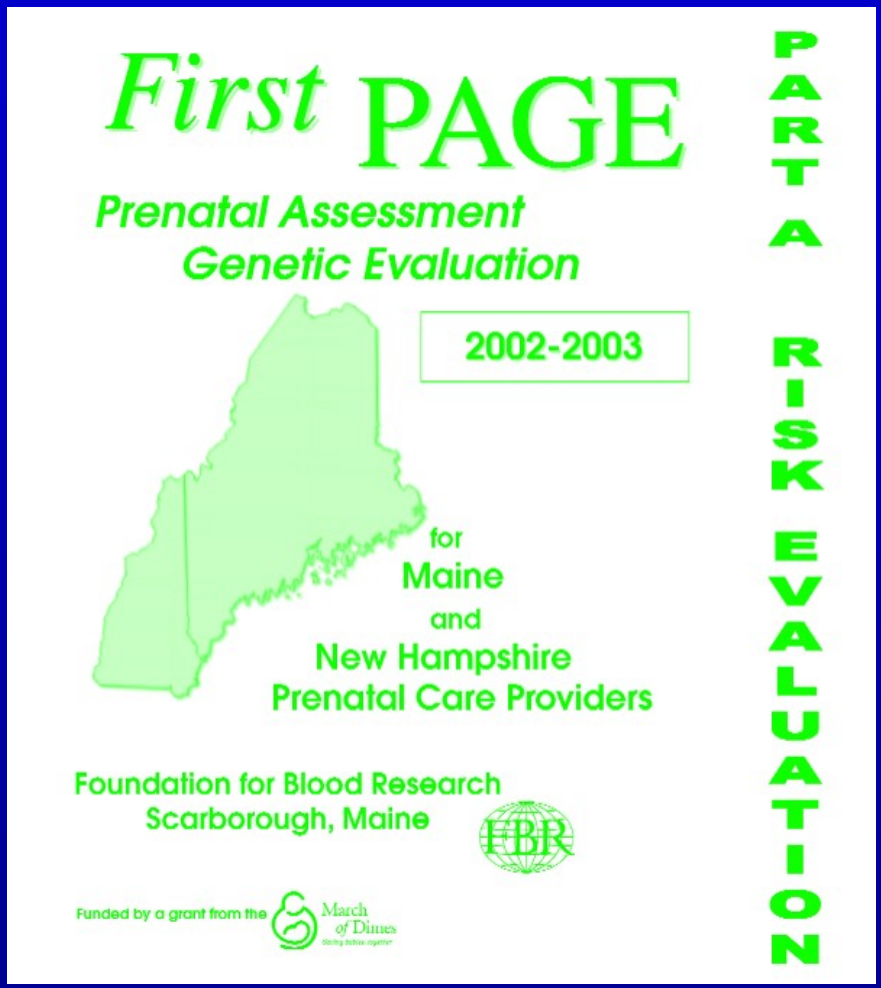
189 distributed, 54 (29%) returned representing over 150 practitioners

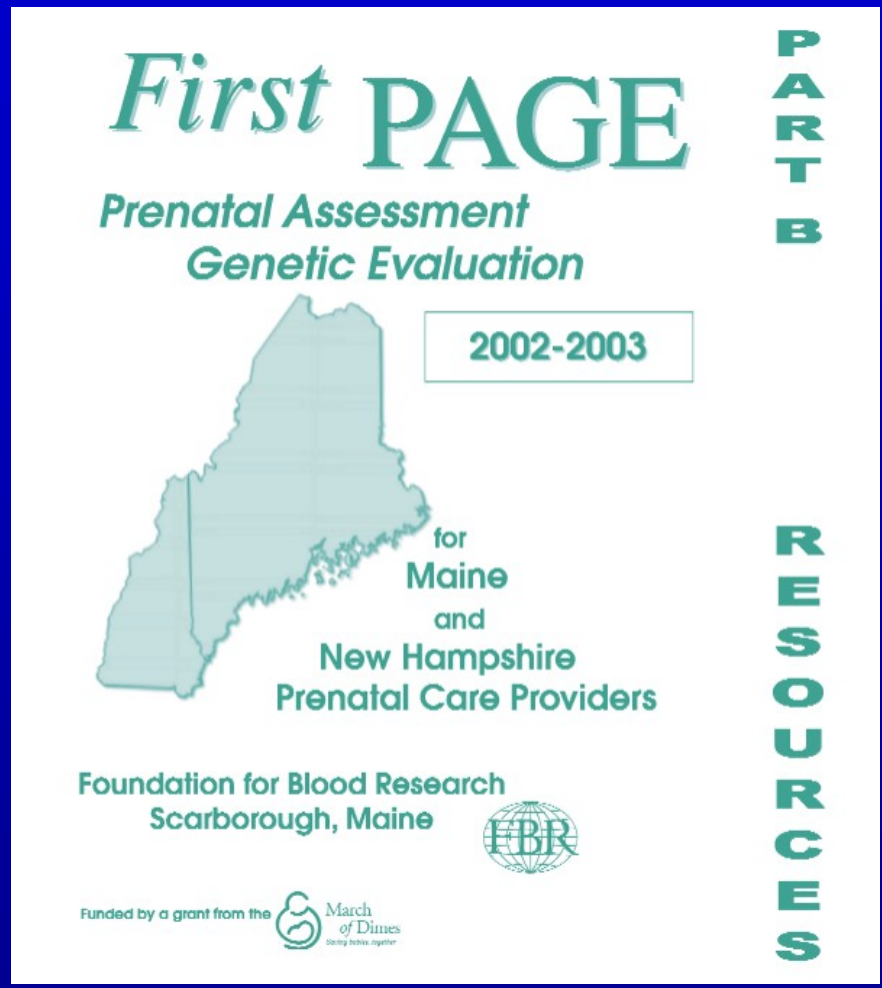
- 48% continued to use *ProgramME* with all new prenatal patients

Content Review and Update

- All 15 sections extensively revised, including content, professional references, and patient resources

Format Modification





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First PAGE Distribution

- 193 modules distributed to 100 practices served by 294 Maine prenatal care providers in 2002
- 172 modules delivered to 108 practices in NH

First PAGE Evaluation 2003

- 42/145 (29%) surveys returned
  147 practitioners represented
- 79% had used *ProgramME*
- 83.3% planned to use *First* PAGE with a all new patients

First PAGE Evaluation (cont.)

- 71.4% -- makes them more confident discussing genetics issues
- 69% -- simplifies risk assessment
- 59.5% -- learned more about genetics
- 73.8% -- helps address genetic risk earlier

Conclusions: *First* PAGE

- is a simple, acceptable mechanism for PCPs to screen for risk
- updates family history, provides referral guidelines, and instills confidence
- educates PCPs about genetics
- assists PCPs in discussing genetics issues with patients
- probably will have no impact on the number of calls or referrals for genetics
- probably will improve the referral process

Applications to Pediatric Conditions?

- Different time frames
  - 9 months vs. 18 years
- Different range of conditions
  - 15 vs. many more
  - Symptoms-free vs. symptoms
- Similar type of physician audience
  - Maintain health in well populations
    - FPs etc
    - OBs vs Peds

Lessons

- Keep it simple
  - Practical, concise, focused
- Involve patients
  - Completing questionnaire, providing family history, discussing level of interest
- Involve specialists as peers
  - Respect PCP turf
- Keep it current
  - Useful, accessible, reliable

Credits for ***First*** **PAGE**

Funded by a grant from the March of Dimes

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