Use of family history information for neural tube defect prevention: integration into state-based recurrence prevention programs

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Estimated NTD rates in 2º and 3º relatives in selected studies, published 1965-1984

<table>
<thead>
<tr>
<th>Area</th>
<th>Rate in area’s population</th>
<th>No. of index cases</th>
<th>Rate in maternal uncles/aunts</th>
<th>Rate in paternal uncles/aunts</th>
<th>Rate in all uncles/aunts</th>
<th>Rate in maternal first cousins</th>
<th>Rate in paternal first cousins</th>
<th>Rate in all first cousins</th>
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</thead>
<tbody>
<tr>
<td>Belfast</td>
<td>0.9%</td>
<td>226</td>
<td>0.5%</td>
<td>0.6%</td>
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<tr>
<td>South Wales</td>
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<tr>
<td>Southampton</td>
<td>0.6%</td>
<td>77</td>
<td>1.0%</td>
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<td>0.3%</td>
<td>0.4%</td>
<td>0.6%</td>
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<td>London</td>
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<td>0.1%</td>
<td>0.2%</td>
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<tr>
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<td>146</td>
<td>0.0%</td>
<td>0.3%</td>
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<td>Atlanta</td>
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<td>360</td>
<td>0.7%</td>
<td>0.3%</td>
<td>0.3%</td>
<td>—</td>
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<tr>
<td>Ontario/Quebec</td>
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<td>377</td>
<td>1.0%</td>
<td>0.3%</td>
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</table>
Use of family history information for NTD prevention

- Use family history information to identify women at increased risk of having a child with an NTD
- Integrated into existing state-based recurrence prevention programs
- Target 2º female relatives (ages 18-44) or partners of 2º male relatives of an affected proband
- Retrospective: determine whether more likely to take folic acid supplements than the general population
- Prospective: intervention to promote FA use
- Evaluation: measured both by whether or not relatives take folic acid and by rates of NTD recurrence within the family

Public health need

- Only about 30% of women of childbearing age report consuming the recommended daily level of FA
- Even among those women with an affected child, a sizeable fraction do not take FA
- Familial cases account for 3.7-18% of all NTD cases
- Recurrence prevention programs have been shown to be effective in increasing FA intake and reducing recurrence
Key steps

- Protocol development and Institutional Review Board approval
- Identify 2006-2007 probands with NTDs
- Pre-contact letter sent to proband mothers
- Proband mothers contacted by telephone to obtain consent to participate in study and to determine number of eligible relatives
- Surveys sent to proband mother for distribution to relatives
- Relatives counseled about the importance of FA for prevention of NTDs and provided with standard information about FA
- Relatives re-contacted after six months to assess differences in FA use since the first contact
- Look for differences between maternal and paternal relatives, by demographics

Survey questions

- FA use
- Contraception use
- Pregnancies and outcomes
- Knowledge about FA and NTDs
- Views on having a child with an NTD
- Derived from other field-tested surveys (e.g., BRFSS, PRAMS, HealthStyles)
Survey questions

1. Do you currently take any vitamin pills or supplements?
   a. Yes
   b. No (GO TO QUESTION 5)
   c. Don’t know/Not sure (GO TO QUESTION 5)

2. Do any of the vitamin pills or supplements you take contain folic acid?
   a. Yes
   b. No (GO TO QUESTION 5)
   c. Don’t know/Not sure (GO TO QUESTION 5)

3. How often do you take this vitamin pill or supplement?
   a. Less than once a week
   b. 1-3 times a week
   c. 4-6 times a week
   d. Every day of the week

4. I take a folic acid-containing vitamin because...

Survey questions

5. I intend to take a multivitamin every day:
   Strongly agree  Strongly disagree
   1  2  3  4  5

6. It is likely that I could have a pregnancy affected by a birth defect:
   Strongly agree  Strongly disagree
   1  2  3  4  5

7. I am afraid of having a baby with serious birth defects:
   Strongly agree  Strongly disagree
   1  2  3  4  5
Survey questions

8. I will take a multivitamin every day:
   Strongly agree  1  2  3  Strongly disagree  4  5

9. It is possible that I could have a pregnancy affected by a birth defect:
   Strongly agree  1  2  3  Strongly disagree  4  5

10. The possibility of having a baby with serious birth defects scares me:
    Strongly agree  1  2  3  Strongly disagree  4  5

11. Which one of the following best describes your pregnancy plans?
    a. I am currently pregnant
    b. I am planning to get pregnant in the next year or so
    c. I am not planning a pregnancy in the next year or so, but I plan to at some time in the future
    d. I do not plan to get pregnant at any time in the future
    e. I cannot get pregnant (post-menopausal, tubal ligation, hysterectomy (GO TO QUESTION 13))

12. Of the following, which is the main method of birth control you used during the last 6 months?
    a. I didn’t use any birth control regularly
    b. Surgical (partner’s vasectomy, tubal ligation, hysterectomy, etc.)
    c. Hormone (birth control pill, Norplant, Depo-Provera)
    d. Barrier (condoms, diaphragm, cervical cap)
    e. Abstinence (not currently sexually active)
    f. Rhythm method (periodic abstinence), withdrawal
    g. Other (sponge, IUD, spermicide)

13. Did you have a neural tube defect (NTD) such as spina bifida or encephalocele at birth?
    a. Yes
    b. No
    c. Don’t know/Not sure

14. Have you had a pregnancy affected by a neural tube defect (NTD) such as spina bifida, anencephaly, or encephalocele?
    a. Yes
    b. No
    c. Don’t know/Not sure
Survey questions

You have been contacted because you have a relative with a pregnancy or child affected by a neural tube defect (NTD) such as spina bifida, anencephaly, or encephalocele.

15. Did you know that you had a relative with a pregnancy or child affected by a neural tube defect (NTD) such as spina bifida, anencephaly, or encephalocele?
   a. Yes
   b. No (GO TO QUESTION 17)

16. How were you told about your relative with a pregnancy or child affected by a neural tube defect (NTD) such as spina bifida, anencephaly, or encephalocele?
   a. The parents of the infant told me
   b. Another relative told me
   c. Someone outside the family (such as a family friend or clergyperson) told me
   d. Other (please tell us):

17. What is your relationship to the relative with a pregnancy or child affected by a neural tube defect (NTD) such as spina bifida, anencephaly, or encephalocele?
   a. She is my sister
   b. She is the sister of my husband or partner
   c. She is my brother’s wife or partner
   d. She is the wife or partner of my husband/partner’s brother
   e. She is my aunt
   f. She is my daughter
   g. She is my son’s wife or partner
   h. She is my mother
   i. She is the mother of my husband or partner

Demographics questions

18. What is your current age? ____________

19. Which of these groups would you say best represents your race?
   a. White
   b. Black or African American
   c. Asian
   d. Native Hawaiian or Other Pacific Islander
   e. American Indian or Alaska Native
   f. Multiracial (parents of different background, please indicate)

20. Are you Hispanic or Latino?
   a. Yes
   b. No
   c. Don’t know/Not sure

21. Are you Arabic or Chaldean?
   a. Yes
   b. No
   c. Don’t know/Not sure
Demographics questions

22. What is the highest grade or year of school you completed?
   a. Never attended school or only attended kindergarten
   b. Grades 1 through 8 (Elementary)
   c. Grades 9 through 11 (Some high school)
   d. Grade 12 or GED (High school graduate)
   e. College 1 year to 3 years (Some college or technical school)
   f. College 4 years or more (College graduate)

23. Are you…?
   a. Married
   b. Divorced
   c. Widowed
   d. Separated
   e. Never married
   f. A member of an unmarried couple

24. What state do you live in?
   a. Colorado
   b. Michigan
   c. Other (please specify):

Folic Acid Education Materials

Thinking About a Baby?
Healthy Mothers Have Healthier Babies
with Folic Acid

Emma’s Story