1. Title of Project:
Using the “Information Rx” Process to Refer Parents of Newborns with Metabolic Conditions Identified by Screening to the Genetics Home Reference Web Site for Health Information

2. Name of Institution:
University of Utah, Department of Biomedical Informatics

3. Location of Institution:
26 South 2000 East
HSEB Suite 5700
School of Medicine
Salt Lake City, UT 84112-5750

4. Name, Mailing and E-Mail Addresses, Voice and Fax Numbers, of Person Submitting Report:
Denise E. Beaudoin, mailing address as above, email address denise.beaudoin@hsc.utah.edu, phone 801-581-4080, FAX 801-581-4297

5. Number and Inclusive Dates of Quarterly Report:
Quarterly Report #1, covering the period from 10/01/07 through 01/15/08

6. Submission Date:
January 15, 2008
NARRATIVE DESCRIPTION

1. Executive summary. Please provide a brief, one-paragraph narrative summarizing major accomplishments made during the quarter.

This is the first reporting period for this project. Progress has been made in several key areas. The NLM approved the revised project budget and an agreement between the NN/LM MidContinental Region, University of Utah and the Department of Biomedical Informatics was signed. The study was reviewed by the University of Utah’s Institutional Review Board and determined to be exempt from federal regulations governing human research, allowing the study to move forward. The study surveys (initial and follow-up) were developed. A prototype of the online follow-up survey was created by NLM systems development staff. Planning for survey pre-testing is underway.

2. Description of Progress toward the Project's Major Objectives
   a. Administrative/Planning Activities:
      i. A study application was submitted to the University of Utah IRB for review on October 25, 2007. The study was deemed exempt on November 11, 2007. (Please see attached IRB notification.)
      ii. The Department of Biomedical Informatics was notified on November 19, 2007, that the revised budget of $67,945 had been approved by the NLM.
      iii. An agreement between the NN/LM MidContinental Region/U of U and the Department of Biomedical Informatics was signed on December 11, 2007.
   b. Collaborations/Partnerships:
      i. Dr. Beaudoin has contacted Ms. Gina Pola-Money, Director of Utah Family Voices, for assistance with recruitment of parents to pre-test study surveys.
   c. Publicity/Marketing Activities: Not applicable.
   d. Product/Resource Development Activities: Not applicable.
   e. Site Visits: None at this time.
   f. Outreach activities:
      i. Dr. Joyce Mitchell provided information about the Genetics Home Reference resource to Eccles Health Sciences librarians on January 14, 2008. Dr. Beaudoin summarized the “Info Rx” evaluation project following Dr. Mitchell’s presentation. Enrolled parents requiring access to the Internet or assistance in viewing the GHR site will be referred to reference staff at the Eccles Library or to local public libraries as appropriate.
      g. Web site development activities: Not applicable.
      h. Exhibits: Not applicable.
   g. Loansome Doc/Document Delivery Activities: Not applicable.
   h. Evaluation Activities: Not applicable at this time.
   i. Problems/Corrective Actions (including significant changes made in implementation of the project): No significant changes have been made in project implementation.
6. Lessons Learned/Significant Feedback: Not applicable at this time.

7. Projected Activities for Next Quarter:
   (a) Pre-testing of both surveys will be conducted with a small number of parents. Two rounds of pre-testing are planned. Revisions to the surveys will be made based on feedback from parents and comments from project partners.

   (b) The revised surveys will be resubmitted to the IRB for review (per protocol).

   (c) Following IRB review, the surveys will be finalized and either printed (initial paper-and-pencil survey) or made available online (follow-up survey) via the “mock” GHR site developed by NLM.

   (d) Dr. Beaudoin will meet with study co-investigator Nicola Longo, MD, and staff at the Metabolic Service Clinic to review the logistics of study implementation and begin recruitment of parents into the study.

8. Reports of Training/Demonstration Sessions and/or Exhibit Reports: Not applicable.

APPENDIX

Include copies of: communications, materials produced, evaluation tools/instruments used or developed, press releases, advertisements, articles for newsletters, etc.

Please see attached study description and surveys (original versions).
Thank you for submitting your request for approval of this study. The IRB has administratively reviewed your application and a designated IRB member has determined that your study is exempt, under 45 CFR 46.101(b), Category 2, from the Federal regulations governing human research. Before implementing any changes in the study, you must submit an amendment application to the IRB and secure either approval or a determination of exemption.

It is the policy of the University of Utah that all human subject research which is exempt under this section will be conducted in accordance with (1) the Belmont report (http://ohrp.osophs.dhhs.gov/humansubjects/guidance/belmont.htm), (2) this institution's administrative procedures to ensure valid claims of exemption, and (3) orderly accounting for such activities.

This determination of exemption only applies to the research study as submitted to the IRB. Since this determination is not an approval, it does not expire or need renewal. Remember that all research involving human subjects must be approved or exempted by the IRB before the research is conducted.

If you have questions about this, please contact our office at 581-3655 and we will be happy to assist you. Thank you again for submitting your proposal.
Study Description

Title: Using the “Information Rx” Process to Refer Parents of Newborns with Metabolic Conditions Identified by Screening to the Genetics Home Reference Web Site for Health Information

Principal Investigator: Joyce A. Mitchell, PhD
Department of Biomedical Informatics
26 South 2000 East
HSEB Suite 5700, School of Medicine
University of Utah
Salt Lake City, UT 84112-5750

Purpose: To learn about parents’ reactions to getting an “information prescription” that directs them to a Web site where they may retrieve accurate and consumer-friendly health information about their child’s metabolic condition, and their assessment of the information found on this site.

Criteria for eligibility:
- Parent/caregiver of a child who has been diagnosed by newborn screening with at least one of 22 specified metabolic condition(s)
- Child’s diagnosis was made by Nicola Longo, MD, PhD at the University of Utah Metabolic Clinic during the enrollment phase of the study

Potential benefits of participation:
- Increased understanding about your child’s medical condition
- Improved communication with your child’s doctor

Time commitment involved:
- Completion of paper-and-pencil survey at the clinic (5 minutes)
- Exploration of GHR Web site on your own (variable time)
- Completion of online survey six weeks after the clinic visit (online survey should not take more than 30 minutes to complete)

Contact: Denise E. Beaudoin, MD, MSPH, MS, Department of Biomedical Informatics, University of Utah, phone: 801-581-4080
List of Specified Metabolic Conditions:

Argininosuccinic acidemia (ASA)
Beta ketothiolase deficiency (BKT)
Biotinidase deficiency (BIOT)
Carnitine uptake defect (CUD)
Citrullinemia (CIT)
Classical galactosemia (GALT)
Glutaric acidemia type 1 (GA I)
Homocystinuria (due to CBS deficiency) (HCY)
3-hydroxy 3-methyl glutaric aciduria (HMG)
Isovaleric acidemia (IVA)
Long-chain 3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
Maple syrup disease (MSUD)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
Methylmalonic acidemia (Cbl A,B)
Methylmalonic acidemia (mutase deficiency) (MUT)
Multiple carboxylase deficiency (MCD)
Phenylketonuria (PKU)
Propionic acidemia (PROP)
Trifunctional protein deficiency (TFP)
Tyrosinemia type I (TYR I)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
Using the “Information Rx” Process to Refer Parents of Newborns with Metabolic Conditions Identified by Screening to the Genetics Home Reference Web Site for Health Information

Initial Patient Survey

1. How frequently do you look up sources of medical information? (Check one please)
   - Very frequently
   - Somewhat frequently
   - Neither frequently or infrequently
   - Somewhat infrequently
   - Very infrequently

2. How frequently do you discuss the medical information that you look up with your doctor? (Check one please)
   - Very frequently
   - Somewhat frequently
   - Neither frequently or infrequently
   - Somewhat infrequently
   - Very infrequently

3. Do you find it easy or difficult to understand the medical issues that your doctor discusses with you? (Check one please)
   - Very easy
   - Somewhat easy
   - Neither easy or difficult
   - Somewhat difficult
   - Very difficult

4. Do you find it easy or difficult to read health information on a computer compared to a book or pamphlet? (Check one please)
   - Very easy
   - Somewhat easy
   - Neither easy or difficult
   - Somewhat difficult
   - Very difficult

5. Where is the location of the nearest computer you can use? (Check one please)
   - At home
   - At the office
   - At a school
   - At a public library
   - At a hospital library
   - At another family member’s location
   - Other: _____________________________ (Please fill in!)

6. On average, about how many hours a day (both at home and work) do you check for email and use the Internet for all other purposes? (Check one please)
   - 0 to 1 hour
   - 2 to 4 hours
   - 5 to 6 hours
   - More than 6 hours
7. Had you ever visited the Genetics Home Reference Web site prior to receiving the “Information Prescription” from your doctor today? (Check one please)
   - Yes
   - No
   - Don’t know

8. If yes, approximately how many times have you visited the Genetics Home Reference Web site? (Check one please)
   - Once
   - Twice
   - Three to five times
   - More than five times
   - I have visited the site before but don’t know how many times
   - I have never visited the Genetics Home Reference Web site

9. How did you first learn about the Genetics Home Reference Web site? (Check one please)
   - From my doctor at today’s clinic visit
   - From another doctor
   - From the staff in my doctor’s office
   - From an ad
   - From friends and family
   - From an Internet search
   - From a local librarian
   - From either a newspaper, or a local radio/television station
   - Other: ____________________________________________ (Please fill in!)

10. I think receiving a prescription from my doctor to visit the Genetics Home Reference Web site for more information is a good idea. (Please tell us if you agree or disagree with this statement. Check one please)
    - Strongly agree
    - Somewhat agree
    - Neither agree or disagree
    - Somewhat disagree
    - Strongly disagree

11. Did you receive enough information today from your doctor to feel comfortable about using the Genetics Home Reference Web site? (Check one please)
    - Yes
    - No
    - Don’t know

12. What is your role in this child’s medical care?
    - Parent
    - Guardian/caretaker
    - Other: ____________________________________________ (please explain)
Please provide the following demographic information about yourself:

13. Name____________________________________________________

14. Date of birth: ______/_______/________
   (month)         (day)          (year)

15. Gender: (Please check the appropriate box)
   ☐ Female
   ☐ Male

16. Street address: ______________________________________________

17. City:_______________________________________________________

18. State: _________

19. Zip code: ________________

20. Telephone number: (___) – (_______) ___

21. E-mail address (if available):_______________________________________________

22. Name of your child’s physician _____________________________________________

23. Which of the following best describes the highest level of education you have
    completed?  (Please check the appropriate box)
   ☐ Did not complete high school
   ☐ High school graduate or equivalent (GED)
   ☐ Some college or vocational school
   ☐ College graduate
   ☐ Some postgraduate school
   ☐ Graduate/professional degree

24. Which of the following best describes your race/ethnicity? (Please check the appropriate box)
   ☐ American Indian or Alaskan Native
   ☐ Asian or Pacific Islander
   ☐ Black, not of Hispanic origin
   ☐ Hispanic
   ☐ White, not of Hispanic origin

Assigned ID number____________

Thank you very much for completing this survey. We will contact you in approximately
six weeks to ask you to complete an online survey about the Genetics Home Reference
Web site. Your feedback is very important to us as it will be used to improve the site.
Using the “Information Rx” Process to Refer Parents of Newborns with Metabolic Conditions Identified by Screening to the Genetics Home Reference Web site for Health Information

Patient Follow-up Survey

ID#_____

1. Approximately how many times have you visited the Genetics Home Reference Web site since you received the ‘Information Prescription’ from your doctor? (Check one please)
   - Only once (when my child was first diagnosed with a metabolic condition)
   - Twice
   - Three to five times
   - More than five times
   - I have visited the site but don’t know how many times
   - I have never visited the Genetics Home Reference Web site

   If you have never visited the Genetics Home Reference Web site, please go directly to question 26.

2. For which condition(s) did your child receive a confirmed diagnosis? (Please indicate below)
   - Argininosuccinic acidemia (ASA)
   - Beta ketothiolase deficiency (BKT)
   - Biotinidase deficiency (BIOT)
   - Carnitine uptake defect (CUD)
   - Citrullinemia (CIT)
   - Classical galactosemia (GALT)
   - Glutaric acidemia type 1 (GA I)
   - Homocystinuria (due to CBS deficiency) (HCY)
   - 3-hydroxy 3-methyl glutaric aciduria (HMG)
   - Isovaleric acidemia (IVA)
   - Long-chain 3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
   - Maple syrup disease (MSUD)
   - Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
   - 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
   - Methylmalonic acidemia (Cbl A,B)
   - Methylmalonic acidemia (mutase deficiency) (MUT)
   - Multiple carboxylase deficiency (MCD)
   - Phenylketonuria (PKU)
   - Propionic acidemia (PROP)
   - Trifunctional protein deficiency (TFP)
   - Tyrosinemia type I (TYR I)
   - Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
   - Other: ______________________________________________________
   - Don’t know
   - My child had a positive screening test but was not ultimately diagnosed with one of the above conditions (false positive test)

3. When was your child first diagnosed with the above condition(s)?
   ______/______/______
   (day) (month) (year)
4. Overall, was the information within the Genetics Home Reference Web site easy or difficult to understand? (Check one please)
   - Very easy
   - Somewhat easy
   - Neither easy nor difficult
   - Somewhat difficult
   - Very difficult

5. Overall, how helpful was the information on the Genetics Home Reference Web site relating to your child’s condition(s)? (Check one please)
   - Very helpful
   - Somewhat helpful
   - Neither helpful nor unhelpful
   - Somewhat unhelpful
   - Very unhelpful

6. How did you use or do you plan to use the health information found on the Genetics Home Reference Web site? (Please check all that apply)
   - Discussed, or will discuss, with my doctor
   - Discussed, or will discuss, with family or friends
   - Has improved my understanding of an illness or health condition
   - Has influenced, or may influence, future health decisions for my child
   - Have contacted, or will contact, a support group in my area
   - Looked for, or will consider looking for, more health information
   - Other ________________________________ (please fill in!)

7. I trust the information on the Genetics Home Reference Web site because my doctor prescribed it. (Please tell us if you agree or disagree with this statement. Check one please)
   - Strongly agree
   - Somewhat agree
   - Neither agree nor disagree
   - Somewhat disagree
   - Strongly disagree

8. A high-quality source of health information helps me talk to my doctor.
   (Please tell us if you agree or disagree with this sentence. Check one please)
   - Strongly agree
   - Somewhat agree
   - Neither agree nor disagree
   - Somewhat disagree
   - Strongly disagree

9. The health information that I find on the Genetics Home Reference Web site will help me make better health decisions for my child.
   (Please tell us if you agree or disagree with this sentence. Check one please)
   - Strongly agree
   - Somewhat agree
   - Neither agree nor disagree
   - Somewhat disagree
   - Strongly disagree
10. The information I received on the Genetics Home Reference Web site added to what doctors told me about my child’s condition. (check one please)
   - Strongly agree
   - Somewhat agree
   - Neither agree nor disagree
   - Somewhat disagree
   - Strongly disagree

11. Overall, how satisfied were you with the health information you found on the Genetics Home Reference Web site? (Check one please)
   - Very satisfied
   - Somewhat satisfied
   - Neither satisfied nor dissatisfied
   - Somewhat dissatisfied
   - Very dissatisfied

12. Was it easy or difficult to find the information you were seeking? (Check one please)
   - Very easy
   - Easy
   - Neither easy nor difficult
   - Difficult
   - Very difficult

13. Did you notice any missing information about your child’s metabolic condition in the Genetics Home Reference Web site?
   - Yes
   - No

14. If yes, in which general area of the Genetic Home Reference Web site did you notice that information was missing? (please check all that apply):
   - An overall explanation of the condition
   - The genes that are related to or cause the condition
   - How persons inherit the condition
   - Where to find more information about treatment for the condition
   - Where to find additional information about the condition
   - Other names people use for the condition
   - A gateway to getting specific questions answered about the condition
   - Glossary definitions that help with understanding the condition
   - Information about diagnosis
   - All the above
   - None of the above

15. Please let us know briefly what information was missing when you searched for information on the Genetics Home Reference Web site. (please write your comments below)

____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
16. Did you use these Internet sources to fill in the missing information? (please check all that apply)

- WebMD.com
- MedlinePlus.gov
- MSN Health
- Genetic Alliance (http://geneticalliance.org/)
- Ask the Geneticist (http://www.askthegen.org/)
- Understanding Genetics: A guide for patients and professionals (http://geneticalliance.org/ws_display.asp?filter=pubs.understanding.genetics.download)
- Madison's Foundation (http://www.madisonsfoundation.org/)
- March of Dimes (http://www.marchofdimes.com/pnhec/4439.asp)
- None of the above

17. When you used the Genetics Home Reference Web site, how frequently did you visit:

**Pages that focus on a single genetic condition?** (check one please)

- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use

**Pages that focus on a particular gene?** (check one please)

- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use

**Pages that focus on a particular chromosome?** (check one please)

- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use

The ‘Help Me Understand Genetics Handbook’ (background information about genetics, including inheritance, genetic counseling)? (check one please)

- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use
Definitions of glossary terms? (check one please)
- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use

Resource lists (that link to other online resources)? (check one please)
- Very frequently
- Frequently
- Neither frequently nor infrequently
- Infrequently
- Very infrequently
- Did not use

18. Which of these areas was most helpful to you within the Genetics Home Reference Web site? (check one please)
- “Genetic Conditions” (information about a single genetic condition)
- “Genes” (information about a particular gene)
- “Chromosomes” (information about a particular chromosome)
- “Handbook”
- “Glossary”
- “Resources”
- None of the above

19. Within the Genetics Home Reference Web site, were the resource lists and links more helpful to you than…

Information about a single genetic condition?
- Yes
- No

Information about a particular gene?
- Yes
- No

Information about a particular chromosome?
- Yes
- No

The ‘Help Me Understand Genetics Handbook’?
- Yes
- No

The glossary terms?
- Yes
- No
20. Among the links to resources that provide other genetic information and organizations outside of the Genetics Home Reference Web site, which site was most helpful to you? (check one please)

- WebMD.com
- MedlinePlus.gov
- MSN Health
- Genetic Alliance (http://geneticalliance.org/)
- Ask the Geneticist (http://www.askthegeen.org/)
- Understanding Genetics: A guide for patients and professionals (http://geneticalliance.org/ws_display.asp?filter=pubs.understanding.genetics.download)
- Madison’s Foundation (http://www.madisonsfoundation.org/)
- March of Dimes (http://www.marchofdimes.com/pnhec/4439.asp)
- None of the above

21. The following resources for genetic information were more helpful to me than the Genetics Home Reference Web site:

WebMD.com (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

MedlinePlus.gov (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

MSN Health (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

Genetic Alliance (http://geneticalliance.org/) (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use
Ask the Geneticist (http://www.askthegen.org/) (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

Madison’s Foundation (http://www.madisonsfoundation.org/) (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

March of Dimes (http://www.marchofdimes.com/pnhec/4439.asp) (check one please)
- Strongly agree
- Somewhat agree
- Neither agree nor disagree
- Somewhat disagree
- Strongly disagree
- Did not use

22. How likely are you to use the Genetics Home Reference Web site again? (Check one please)
- Very likely
- Likely
- Neither likely nor unlikely
- Unlikely
- Very unlikely

23. How likely are you to recommend the Genetics Home Reference Web site to others? (Check one please)
- Very likely
- Likely
- Neither likely nor unlikely
- Unlikely
- Very unlikely

24. Please let us know what area, or link, you found the most helpful when you used the Genetics Home Reference Web site. (please write your comments below)

____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
25. Please feel free to add other comments about the Genetics Home Reference Web site:
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________
____________________________________________________________________________

26. If you have NEVER visited the Genetics Home Reference Web site, please tell us the reason(s). (Please check all that apply)

☐ I do not have access to a computer and the Internet
☐ I do not use the Internet because it is too complicated
☐ It's difficult for me, at times, to understand written health information
☐ English is not my first language
☐ I prefer another Internet source for health information rather than the Genetics Home Reference Web site
☐ It's upsetting to read about an illness that affects my child
☐ I already know enough about the medical issues my doctor asked me to look up
☐ What the doctors tell me is sufficient
☐ It's just not my nature to read about medical issues
☐ I forgot
☐ I have not had time
☐ Other: __________________________________ (Please fill in)

27. Would it increase your interest in using the Genetics Home Reference Web site to know that many local libraries will:
* provide free access to the Internet for patients who do not have their own computers,
* help patients locate health information using the Genetics Home Reference Web site,
* demonstrate the Genetics Home Reference Web site to you?
(Check one please)

☐ Very much
☐ Somewhat
☐ Not at all

28. Do you use any of the following health information resources? (Please check all that apply)

☐ Health care providers
☐ Friends and/or family members
☐ Other Web sites (please list):____________________________________________________
☐ Medical journals
☐ Print media (newspapers/magazines)
☐ Radio/television programs
☐ Other (please list):______________________________________________________________

29. Did you search on any of these Internet services to find health information about your child’s condition?

Yahoo
☐ Yes
☐ No

Google
☐ Yes
☐ No
30. Compared to all other health information sources you use at this time (regardless if they are
or are not located on the Internet) how frequently will you use the Genetics Home Reference
Web site in the future? (check one please)
   □ Very frequently
   □ Frequently
   □ Neither frequently nor infrequently
   □ Infrequently
   □ Very infrequently
   □ Never

Thank you very much for taking the time to complete this survey about the Genetics
Home Reference Web site.