Facilitating the Iterative Design of Informatics Tools to Advance the Science of Autism

David R Kaufman\textsuperscript{a}, Patrick Cronin\textsuperscript{a}, Leon Rozenblit\textsuperscript{b}, David Voccola\textsuperscript{b}, Amanda Horton\textsuperscript{b}, Alisabeth Shine\textsuperscript{a}, And Stephen B Johnson\textsuperscript{b}

\textsuperscript{a} Department of Biomedical Informatics, Columbia University, New York

\textsuperscript{b} Prometheus Research, LLC, New Haven, CT, USA
SFARI Base

• Simons Foundation Autism Research Initiative (SFARI) established
  – a permanent repository of phenotypic and genetic data set from 2,700 families
  – each of which has exactly one child affected with ASD

• SFARI Base, a web-based platform developed in collaboration with Prometheus Research LLC
  – Provides access to scientific data
  – Information management and analytic tools to advance the science of autism
Data Dig

- Scientific investigators
- Research coordinators
- Data managers
- Autism data curators
Select Columns

Choose a group to specify the list of columns that appear below.

Light grey bars indicate Variable Tags. Clicking a light grey bar will narrow the list of columns that appear below.

Grey numbers indicate how many variables fall under each Tag category

Dark grey bars indicate Variables. Clicking a dark grey bar will cause the corresponding variable to return in your query result.
Mental Model of Query Process

Males age 4 to 12 years of age with significant language delays as measured by different variables
Objectives

• Usability evaluation/iterative design
• Characterize high yield problems
• Identify tractable changes to Data Dig
  – Greatly enhance the user experience
• Provide resources for users to formulate queries with relative ease
• Comprehensible feedback/output of search
• Consistency, navigation, feedback and enhanced functionality
Methods

A. Cognitive walkthrough
B. Heuristic evaluation
C. Usability testing
D. Web-based survey of scientists
E. Participant design study
F. Observations of autism researchers in-situ
Analysis

• Cognitive Walkthrough
  – Two analysts
  – Nielsen’s Usability Heuristics (3 raters)
  – Ennis and Sutcliffe Cognitive Model of IR
    • Domain and device knowledge

• Usability Testing
  – Recorded 3 users with Morae
  – 10 queries of varying complexity drawn from journal articles on autism
  – Identified issues from recording
Cognitive Walkthrough

• Task-analytic method for understanding task complexity
  – Goals /Subgoals for each task
  – Action/step
  – Necessary knowledge
  – Feedback (ie, what’s visible on the display)
  – Potential problems/cognitive demands

• Task complexity
  – number of actions
  – number of screen transitions
  – time needed to complete a task and
  – required chunks of knowledge
Usability Testing

• Task: Formulate queries to answer problems of varying complexity
  1. How many probands in the database?
  2. Autism Diagnostic Interview (ADI-R) total score?
  3. Is there data on the proband’s birth?
     Head circumference and weight at birth
     Proband born vaginally or by a C-section?

• Think-aloud and video-capture using Morae
Moraе Capture of Data Dig

**Step 1** Choose a group to explore, navigate and filter measure variables, and click to select what you want in your query.

Group: All-Item-Level-Variables

All-Item-Level-Variables -> Pregnancy

- Birth-Order 6
- Demographics 2
- Diet 30
- Illness 272
- Medical 1145
Results Summary

- Users able to answer most queries with some help from the moderator
- Numerous difficulties learning how to master the different elements of the system
- More than 50 usability problems
  - relatively minor to more serious ones impeded effective and efficient use of the tool to answer queries
- Problems with greatest frequency:
  - Difficulty understanding meanings of variables
  - filter categories correctly, use the Boolean filter
  - correctly interpret the feedback provided by the system
- Difficulty forming a mental model of the underlying database
  - Precluded them from making informed navigation choices while formulating queries
Feedback: There is No Visual Cue to Mark Selected Variables in Step 1

• Description
  – Selected variables lack cue

• User Data
  – Variables were missed
  – Unclear on selection

• Potential Solution
  1. Checkbox
  2. Disappearing Variables
  3. Shading
Feedback: There is No Visual Cue to Mark Selected Variables in Step 1

Step 1 Choose a group to explore, navigate and filter measure variables, and click to select what you want in your query.

Group: Core-Descriptive-Variables

Core-Descriptive-Variables ➔ Comorbidity

Select the variables you want to include in your query.

Step 2 Optionally, add filter criteria.
Limited Functionality: Inability to Select Multiple Variables

- **Description**
  - Only select variables individually

- **User Data**
  - All users desired capability

- **Potential Solution**
  1. “Select All” button
  2. Highlight & Drag
Limited Functionality: Inability to Select Multiple Variables

Step 1. Choose a group to explore, navigate and filter measure variables, and click to select what you want in your query.

Group: All-Variables-by-Instrument

Step 2. Optionally, add filter criteria.

Step 3. View query results.
Limited Functionality: Boolean Filter Issues

• Description
  – Functionality is lacking

• User Data
  – Ignored Boolean filter entirely
  – 1 of 3 limited use

• Potential Solution
  1. Improve functionality
  2. Increase constraints
  3. Eliminate & Replace with checkbox
Summary

• Documented a range of usability problems
• Data Dig presented challenges to new users (lacking either device or domain knowledge)
  – Difficulty formulating a mental model
• Identified tractable changes and offered solutions when possible
  – Continuous iteration with marked improvement
Conclusions

• Clinical research informatics provides new tools to advance clinical science and practice
• Lack a stable interaction paradigm for enabling scientific researchers and other users to access and analyze large data sets
  – Complexity and learning curves
• Significant challenges as well as opportunities for human-computer interaction researchers to contribute to the advancement of effective and enabling tools
  – Advance the science of autism and other disorders