New Form for your Newborn: Assessing Patient Comprehension and Preferred Formatting of Newborn Screening Result Reports.

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2019 APHL Newborn Screening and Genetic Testing Symposium
• Genetic counseling graduate student at the University of Minnesota
• Thesis project with Minnesota’s NBS program
• No conflicts of interest
Outline

- Need for research on the public’s understanding of and preference for NBS reports
- Previous research
- Our research and results
- Possible report improvements

Figure 1: Cystic Fibrosis Foundation (https://www.cff.org/What-is-CF/Testing/Newborn-Screening-for-CF/)
Background

• Need for research
  – Potential for misinterpretation of reports
  – Patients may be viewing their report
  – No published data on NBS reports and the public

• Goals of the study
  – Investigate public’s understanding and preferences for NBS reports
  – Help inform future MN NBS report templates
Previous Research on Genetic Test Reports

• Patient comprehension errors exist with genetic reports (Brewer, 2012; Ostergren, 2015)
  – Errors with identification of risk
  – Simpler format had lower error rates
  – Poorest comprehension for carrier screening results
• High health literacy levels (Brewer, 2009; Helitzer, 2009; Kasabwala, 2012)
• Patient preferences for genetic report: (Smit, 2016; Stuckey, 2015)
  – More valid information and resources
  – 100-person diagrams
  – Simple language
  – Visual appeal, reduced clutter
  – What to expect in the future/recommended next steps
• Avoid jargon or include glossaries (Haga, 2017)
• Present risk both in text and graphically
• Minimize length (Valenstein, 2008)
• Use headlines

Figure 2: 100-person diagram (http://www.visguy.com/2009/10/01/village-of-100-people-diagrams/)
Methods

• Report creation
  – Current MN NBS report and new created NBS report

• Recruitment of participants \( (n=95) \)
  – MN State Fair

• Survey
  – Demographics

• Interview
  – Comprehension
  – Preference

• Data analysis
  – Answers sorted into categories
  – T-test
Final Newborn Screening Report

(Duplicate)
LABORATORY REPORT
Submitter: Card Barode:
Address: Physician/Clinic:

Patient Information: Specimen Information:
Infant Name: Date Collected:
Date of Birth: Date Received:
MRN: Date Reported:
Mother’s Name: Copy Printed:

Page 1 of 1

SCREENING RESULTS

<table>
<thead>
<tr>
<th>Disorder/Profile</th>
<th>Value</th>
<th>Result</th>
<th>Expected Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acylcarnitine Profile*</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits</td>
<td></td>
</tr>
<tr>
<td>Amino Acid Profile*</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits</td>
<td></td>
</tr>
<tr>
<td>Biotinidase Deficiency (BTDI)</td>
<td>Within Normal Limits</td>
<td>&gt; 55 µg/dL</td>
<td></td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (21-OHP)</td>
<td>Within Normal Limits</td>
<td>Weight Dependent</td>
<td></td>
</tr>
<tr>
<td>Congenital Hypothyroidism (TSH)</td>
<td>Within Normal Limits</td>
<td>Age Dependent</td>
<td></td>
</tr>
<tr>
<td>Cystic Fibrosis (RT)</td>
<td>[RT] 65.3 ng/mL</td>
<td>One Mutation Found</td>
<td>&lt; 90th percentile</td>
</tr>
<tr>
<td>Galactosemia (GAL &amp; TGAL)</td>
<td>Within Normal Limits</td>
<td>GALT &gt; 3.2 µg/dL, TGAL &lt; 12 mg/dL</td>
<td></td>
</tr>
<tr>
<td>Hemoglobinopathies</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits = FA</td>
<td></td>
</tr>
<tr>
<td>Severe Combined Immunodeficiency (TREC)**</td>
<td>Within Normal Limits</td>
<td>TREC Present</td>
<td></td>
</tr>
<tr>
<td>X-linked Adrenoleukodystrophy (C26:0-LPC)**</td>
<td>Within Normal Limits</td>
<td>&lt;0.16 µmol/L C26:0-LPC</td>
<td></td>
</tr>
<tr>
<td>Lysosomal Disease Profile***</td>
<td>Within Normal Limits</td>
<td>Enzyme Activity Present</td>
<td></td>
</tr>
<tr>
<td>Spinal Muscular Atrophy** (zero copies of SMN1)</td>
<td>Within Normal Limits</td>
<td>SMN1 Present</td>
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</table>

Comments

CYSTIC FIBROSIS RESULT INTERPRETATION: This newborn screen is positive for cystic fibrosis. The immunoreactive trypsinogen is elevated and 1 CFTR mutation was identified. The infant may have a second CFTR mutation not identified by newborn screening. Further molecular testing may be necessary. Contact a cystic fibrosis center and arrange clinical evaluation/confirmatory sweat chloride testing. Genetic counseling may benefit the family.
- Mutations Found: CFTR Mutation #1: delta F508 (c.1521_1523delCTT), CFTR Mutation #2: Not identified
Summary: Increased Risk for Cystic Fibrosis (CF)

What was found? Elevated immunoreactive trypsinogen (IRT) and one CFTR mutation. [CFTR Mutation #1: delta F508 (c.1521_1523delCTT); CFTR Mutation #2: Not Identified]

What does this result mean? This result shows a moderately increased risk for cystic fibrosis. Infants with one CFTR mutation are most likely healthy carriers of cystic fibrosis, but there is a chance that there is a second mutation that was not identified by the newborn screen. Infants with two CFTR mutations have cystic fibrosis.

What needs to be done next? Further testing is needed to find out if the infant is a carrier of cystic fibrosis or is affected with cystic fibrosis. This is because newborn screening does not screen for every possible cystic fibrosis mutation, but instead only the most common mutations. Contact a Cystic Fibrosis Center to schedule further testing (called a ‘sweat test’) and to talk with a genetic counselor. Further testing should be done before the infant is one month old. For more information about cystic fibrosis, go to: http://www.health.state.mn.us/divs/ohh/newborn/materials/factsheets/index.html#cysticfibrosis

**Increased-Risk Screening Results:**

<table>
<thead>
<tr>
<th>Disorder/Profile</th>
<th>Expected Range</th>
<th>Value Seen</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cystic Fibrosis</td>
<td>&lt;96th Percentile</td>
<td>IRT=65.3 ng/mL, One mutation found</td>
<td>Positive (Abnormal)</td>
</tr>
</tbody>
</table>

**Low-Risk Screening Results:**

<table>
<thead>
<tr>
<th>Disorder/Profile</th>
<th>Value Seen</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acetylcarnitine Profile*</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Amino Acid Profile*</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Botulinum Deficiency (BTO)</td>
<td>&gt;55 uU/L</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (CAH)</td>
<td>Weight Dependent</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Congenital Hypothyroidism (CHTH)</td>
<td>Age Dependent</td>
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<tr>
<td>Galactosemia (GALT &amp; TGA)</td>
<td>GALT = 3.2 uU/L, TGA = &lt;12 mg/dL</td>
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<tr>
<td>Severe Combined Immunodeficiency (SCID)***</td>
<td>TREC Present</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>X-linked Adrenal Hypoplasia (CXO-ASD)**</td>
<td>&lt;1.66 umol/L</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Lysosomal Disease Profile ***</td>
<td>Enzyme Activity Present</td>
<td>Within Normal Limits</td>
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</table>

**About Newborn Screening:** Newborn screening was done on a few drops of blood taken from the infant’s heel shortly after birth. Newborn screening helps check if an infant is at risk for a panel of rare health conditions. Newborn screening results alone do not diagnose an infant with a condition; further testing is always needed. As with any screening test, false positive and false negative results are possible. An MDH genetic counselor is available for consultation regarding screening results at 651-201-3548.

**Terminology:**

A screening test is a type of health test that is designed to find out if an individual is at-risk of having a condition before there are symptoms. Newborn screening is an example of a screening test. Screening is not the same as a diagnostic test—a screening test that shows an increased risk for a condition must be followed up with further testing to actually know if the individual is affected or not.

A low-risk (normal/negative) result means that the result is within normal limits and there is no sign of the condition. This is a normal result. No further follow-up is needed.

An increased-risk (abnormal/positive) result means that the result is out-of-range or the result is abnormal. This means the infant may have the condition.

A false positive result means that the result showed an increased risk on newborn screening, but results on diagnostic testing were normal and the individual is not affected with the condition. False positives are possible on any screening test.

A true positive results means the result showed an increased risk on newborn screening and the individual is affected.

**More about cystic fibrosis (CF):** Cystic fibrosis is a lifelong condition that causes thick, sticky mucus to build up. This build-up of mucus causes trouble breathing, lung infections, and prevents the absorption of nutrients from food. Cystic fibrosis is inherited in an autosomal recessive pattern (see below). Infants with autosomal recessive conditions, like cystic fibrosis, inherit two non-working copies of the CFTR gene (e.g., two CFTR mutations) — one from their mother and one from their father. Infants that are healthy carriers for cystic fibrosis inherit one non-working copy (e.g., one CFTR mutation) of the gene. Being a carrier, so having a single non-working gene, does not cause the typical, serious symptoms that are usually associated with a condition. For more info on cystic fibrosis, please visit: http://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf.
Data Collection

- Two day period at the MN State Fair
- Eligibility criteria: English speaking, Minnesota resident, and between 18-55 years old
- Survey
  - iPad survey with demographic information
- Interview
  - Assigned one of the two sample reports ($n=46$ and 49)
  - Read paragraph on NBS and project goals
  - Viewed report
  - 14 questions: understanding of information in the report, report’s strengths and weaknesses, and improvement ideas
  - Interviewer and scribe pair asked questions and wrote responses
Demographics

• The majority of participants were:
  – Female (73.7%)
  – Married (51.6%)
  – Identified as White (87.4%)
  – Highly educated
    • 60% bachelor’s degree or further
  – Middle to upper class
    • 75.8% reported annual household income of $50,000 or higher, and
      44.2% reported annual household income $100,000 or higher

• Ages spanned from 18 to 55 years old; average age of 36.9 years
Demographics

- 54.3% had children and 45.7% did not have children
  - Number of children ranged from 0-5
- Of individuals who reported 1 or more child:
  - 43.1% reported they had received a NBS report before
  - 23.5% reported they had not received one before
  - 29.4% were not sure whether they had received one before
  - 3.9% of participants left the question blank
Comprehension: What Results Mean

- Correct answers given:
  - May have CF
  - Likely a carrier
- Incorrect answers given:
  - Has CF/sick
  - Average, healthy child
  - Predisposition for CF
  - Unsure
- Unclear
  - Needs more testing
- 2% and 13% identified the child was likely a carrier from current report and new report respectively
Comprehension: What Needs to be Done Next

- Correct answers given:
  - More testing
  - See specialist/Doctor/GC
- Incorrect answers given:
  - Redo screen
  - Treatment for CF
  - Unsure
- 14% and 4% unsure who to contact for questions the from current report and new report respectively
Comprehension: What is Confusing

- Common answers:
  - Jargon (20.4% and 26.0% from the current and new reports respectively)
  - "Mutation"
  - "Positive" test
  - IRT
  - Units
  - < and > symbols
  - Information about the mutation (14.3% and 10.9%)
  - Table: values seen and range (42.9% and 23.9%)
  - List of disorders tested for (38.8% and 26.0%)
  - If the child had CF (current report only, 16.3%)

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Preference: Length

Participants' Responses to Report Length
Current NBS Report Version

- Depends: 10.4%
- Too short: 8.3%
- Just right: 81.3%

Participants' Responses to Report Length
New Created NBS Report Version

- Depends: 4.3%
- Too long: 15.2%
- Too short: 2.2%
- Just right: 78.3%
Preference: Layout Strengths

• Generally participants liked the layout/organization

• Current report:
  – Abnormal result bolded (26.5%)
  – Table (12.2%)

• New report:
  – Positive result highlighted
  – Section headings (19.6%)
Preference: Layout Weaknesses

• Current report:
  – Color/highlight abnormal results (16.3%)
  – Contact info bigger (10.2%)
  – Other ideas: Separating positive/negative results, add condition info on the back page, add color, add bullet points to follow up, make font bigger

• New report:
  – Terminology on first page or where it was introduced (19.6%)
  – Make table bigger (13.0%)
  – Move what NBS is to top (13.0%)
  – Other ideas: Reduce number of words, make images bigger, underline/circle contact info, add bullet points to follow up, move low risk results to back page, make font bigger
Preference: What to Add

• Nothing more (8.2% and 19.6% from the current and new report respectively)
• Information on CF (40.8% and 32.6%)
• Information on all conditions tested for or links to this (26.5% and 21.7%)
• Their child’s value and the normal range (8.2% and 15.2%)
• More and clear info on what to do next (28.6% and 26.1%)
  – Contact info (14.3% and 23.9%)
• Chance child has CF (2% and 13%)
• Links (16.3%, current report only)
• Terminology section or key (10.2%, current report only)
Preference: What to Have for Normal Results

- Same type of information
  - 43.9% and 55.3% wanted all the information from the new and current report respectively
  - Only one participant from 95 participants said they didn’t want a report if results were normal
Strengths and Limitations

• Strengths:
  – Large amount of data
  – Insight into public’s experience with NBS reports
  – Response saturation

• Limitations:
  – Participants not representative of general population
    • Mostly female, white, highly educated, middle-upper class
  – Participants did not view both reports or hear others’ suggestions
  – Did not analyze comprehension or preference after medical provider discussed the report with them
  – No input from other medical professionals
Conclusions

• Errors for what results meant and what next steps were existed for both the current and new created NBS reports
• Public wanted their report for both positive and negative result outcomes
• Public preferred a report with limited jargon and explanation of terms, graphics, information on conditions (especially if positive test result), thorough next steps/contact info, use of bolding or highlighting, and both their results and the normal ranges
Creating a New Report

• Table with results
  – Normal range and child’s specific value
  – Graphic?
• Limited jargon and terminology definitions
  – Ex. Mutation, false +/-, positive screen
• Highlighted/bolded abnormal results, summary, and contact info
• Information on (or links to) disorders tested
  – Info and links for abnormal result condition
• Graphics and color
• Clear and specific next steps
  – Bullets
  – Contact info
• 1-2 pages acceptable
• Big font
About Newborn Screening:

Summary: Increased risk for Cystic Fibrosis (CF). Underlined words are defined on page 2.

What was found?

What does this result mean?

Above: Less than 1 in 10 increased risk cystic fibrosis results are expected to be true positives (affected). Over 9 in 10 positive cases are likely to be carriers.

What needs to be done next?

1.
2.
3.

Contact 555-555-5555 with questions

More about cystic fibrosis (CF):

For more info on cystic fibrosis, please visit: http://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf

Information:

- Disorder/Profile
- Value Seen
- Result

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</tr>
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<td>-55-0 mg/mL</td>
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<tr>
<td>Hereditary Glioseplementation</td>
<td>Within Normal Limits</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Severe Combined Immunodeficiency (SCID)**</td>
<td>T-Cell Positive</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>X-linked Adrenoleukodystrophy (XLALD)***</td>
<td>&lt;25.55 p/μL</td>
<td>Within Normal Limits</td>
</tr>
<tr>
<td>Tyrosine Oxidase Deficiency ***</td>
<td>Enzyme Activity Present</td>
<td>Within Normal Limits</td>
</tr>
</tbody>
</table>

All the conditions tested for on NBS are condensed into the above groupings. To see all the conditions tested for, and learn about what they are, visit https://www.health.state.mn.us/people/newbornscreening/program/2018newbornscreeningpanel.pdf
Acknowledgements

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Resources


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