Revamping Provider Educational Materials for Newborn Screening Referrals

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Public Health Laboratories
Washington State Department of Health
New Conditions Grant (UC9MC30369)

• Method development
• Educational outreach campaign
• Long-term follow-up
Health Promotion and Communication Section

• Helped revamp the brochure
  – audience testing
  – graphic design
  – (poster #129)

• Could we use them to help us with other stuff?
Methods

• Qualitative interviews to improve X-ALD general overview
• HPCS: what else to you send to PCPs?
REFERRAL

Washington State
(360) 710-5405

This baby was referred by [provider] to the [baby's name] newborn screen as [mother's name].

Patient's Name:

Mother's Name:

NBS Referral Date:

These results indicate that this child has a presumptive disorder. Please collect blood for very long chain fatty acids and amino acids and submit to Seattle Children's Hospital.

From: Caroline N

Subject: Referral

Remarks: [Include any additional relevant information here]

This referral order is due within 24 hours.

IMPORTANT

All results should be reviewed by the provider.

NEWBORN

PRESUMPTIVE PHENOTYPE

For long chain fatty acids and amino acids, collect blood for very long chain fatty acids and amino acids and submit to Seattle Children's Hospital.

Signatures:

Follow-up will be sent to [provider] at [email]. Please contact provider if you have any questions or require further information.

Send specimens to: SEA 4680

X-linked Adrenoleukodystrophy (X-ALD)

General Overview

- **What is X-ALD?**
  - X-ALD is a rare, X-linked disorder that affects the central nervous system.

- **How many forms are there?**
  - There are three major forms of X-ALD: adrenocortical (the form seen in newborns), childhood cerebral (the most severe), and adult form.

- **What does it mean if a baby has an abnormal X-ALD screen?**
  - An abnormal screen may suggest a baby may have X-ALD. Additional testing may be recommended by doctors who take care of children with X-ALD.

- **How does one find out if a baby has an abnormal X-ALD screen?**
  - A blood test can detect the buildup of very long-chain fatty acids. Additional tests may be recommended by doctors who take care of children with X-ALD.

- **What are the effects of having X-ALD if it is not treated?**
  - Untreated X-ALD can cause cognitive decline, behavioral problems, and loss of motor skills.

- **What is the treatment for X-ALD?**
  - Treatment options depend on the type of X-ALD and may include medications or surgery.

For more information: [http://www.crooks.org/XALD.html](http://www.crooks.org/XALD.html) and [http://www.babyfirsttest.org/search?ALD](http://www.babyfirsttest.org/search?ALD)
X-ALD General Overview

- Written with dual purpose
  - Educate the PCP
  - Pass along page to families
- FAQ format – plain talk

Q. How many forms of X-ALD are there?
A. There are three main forms of X-ALD: an adrenal form (the early form), a childhood cerebral form (the most severe) and a mild form that shows up in adulthood (the late form). X-ALD can present with different symptoms, even within the same family.
Methods

• Provided HPCS with contact info for PCPs of recent X-ALD referrals

• Round one of phone calls soliciting participation [1 interview]
  – “I’ve left a lot of messages and even had a couple “wrong numbers”
    or had to be routed through an automated phone system a few times. I have a new appreciation for the work you do for sure!”

• Round two of phone calls

• One-page fax request to remaining 15 PCPs [3 interviews]
## Results

- 8-page report from interviewer

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<thead>
<tr>
<th>Key Question</th>
<th>Short Answer</th>
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<tr>
<td><strong>What are the general communication preferences of the primary audience?</strong></td>
<td>Providers want to receive the information directly in the electronic medical record, but acknowledge this is probably not practical and think that fax and telephone follow up continues to be the best means of communication about newborn screening results.</td>
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<td><strong>What is a typical flow of information in the PCPs office when a fact sheet is received?</strong></td>
<td>Providers rely on the pre-fax phone call. The fax, when received, is picked up by a nurse or medical assistant and passed directly to the provider in paper form. Providers direct follow up care then usually pass the hard copies off to medical records staff who add notations to the medical record.</td>
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<td><strong>To what extent do PCPs find the fact sheets useful?</strong></td>
<td>Providers generally do not use or rely on the fact sheets at all. They appreciate that the information is there should they need it, but it is not a primary source of information for them. The primary concern of providers is ordering the follow up testing or care and referring the patient to a specialist if necessary.</td>
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<td><strong>To what extent is information received from the NBS program shared with parents?</strong></td>
<td>Primary providers rarely or never share the fact sheet with the parents and most would be reluctant to do so. They would prefer to refer the family to a specialist who can answer questions. Providers fear offering too much information without feeling confident in their ability to answer follow up questions. They are also concerned about overwhelming the family with too much information that they think is complicated.</td>
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Recommendations

- Revise instructions for follow-up in fax
- Ask PCPs to include a copy of the packet in the baby’s electronic medical record
- Reorganize the content into headings rather than a FAQ format
NEWBORN SCREENING FOLLOW-UP RECOMMENDATIONS FOR

Baby Name
DOB 1/1/2018
Mother: Mom Name

This baby had a positive newborn screen for X-Linked adrenoleukodystrophy (X-ALD). Summary of next steps:

☐ Notify the parents about results;
☐ Order diagnostic tests to rule out or confirm X-ALD;
☐ Include a copy of these documents in the patient’s medical record;
☐ Complete and return Referral Notification Form.

There are three main forms of X-ALD:

1. Adrenal form (the early form)
2. Childhood cerebral form (the most severe)
3. Adult form that shows up as adulthood (the late form)

SYMPTOMS
If the adrenal or cerebral forms of X-ALD are not treated, the buildups of very long chain fatty acids can affect the adrenal glands and brain, leading to behavioral problems, muscle weakness, hearing loss, blindness, and potentially death. X-ALD can present with different symptoms even within the same family.

TREATMENT
The type of treatment depends on the form of X-ALD. Treatments are given to treat insulin-refractory hypoglycemia. A ketone test strip can help identify ketosis in patients with the cerebral form. Doctors at Seattle Children’s and other centers can help patients with X-ALD and can talk about tests and treatments.

NEXT STEPS
The next step is to get a blood test to detect the buildups of very long chain fatty acids. If the blood test is abnormal, a visit to a biochemical geneticist may be needed. They may recommend additional tests or treatment. The tests cannot predict the form of X-ALD that will develop.

RESOURCES
For more information about X-ALD, please see the Disorders section of our website:
www.rudd.org/disorders. Information can also be found at https://www.xald.org.
More Feedback

- CF NBS QI - DNA testing
  - Need updated CF referral packets – DNA results
  - CF Center – Tacoma
    - PCP review group
    - 6-page report
  - Feedback
    - Online resources?
    - SwCl scheduling?
    - Meantime?
Cost

Qualitative Interviewing
• HPCS qualitative interviewing = $1,863 (two staff)
• CF partners network = $0

Re-formatting PCP materials
• Staff time
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