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A Guide: Positive & Borderline Newborn Screen Results

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Learning objectives

- **Objective 1:**
  - Define positive and borderline (versus unsatisfactory) newborn screening (NBS) results
- **Objective 2:**
  - Understand how to frame the initial conversation about positive and borderline results to meet provider and family needs
- **Objective 3:**
  - Identify appropriate steps to take following positive and borderline results

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What is a positive newborn screen result?

- **Result interpretation:** positive, abnormal, out-of-range
- **Result meaning:** the child is at high(er) risk for having one of the conditions on the screening panel
- **Potential interferences/influences** (https://www.aphl.org/programs/newborn_screening/Pages/NBS%20Interference%20List.aspx):
  - Transfusion
  - TPN
  - Low birth weight/prematurity
  - Specimen taken too early/too late
  - Heat/humidity during specimen transport
- **Key talking points when communicating results:**
  - Screening is **NOT** diagnostic testing, so more testing needs to be done to figure out if the child is truly affected
  - Provide an action plan (e.g., referral information, timeline, etc.) and informational resources to family after consulting with local specialists

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What is a borderline newborn screen result?

- **Result interpretation:** borderline, inconclusive, equivocal
- **Result meaning:** the result value was between normal and abnormal values (i.e. just outside the normal range); further testing is needed in order to clarify
- **Potential interferences/influences:**
  - Transfusion
  - TPN
  - Low birth weight/prematurity
  - Specimen taken too early/too late
  - Heat/humidity during specimen transport
- **Key talking points when communicating results:**
  - The result fell between the normal and abnormal range
  - Further testing needs to be completed in order to clarify the risk to the child. Each program will provide their recommendations in these cases; often this will be a repeat specimen.

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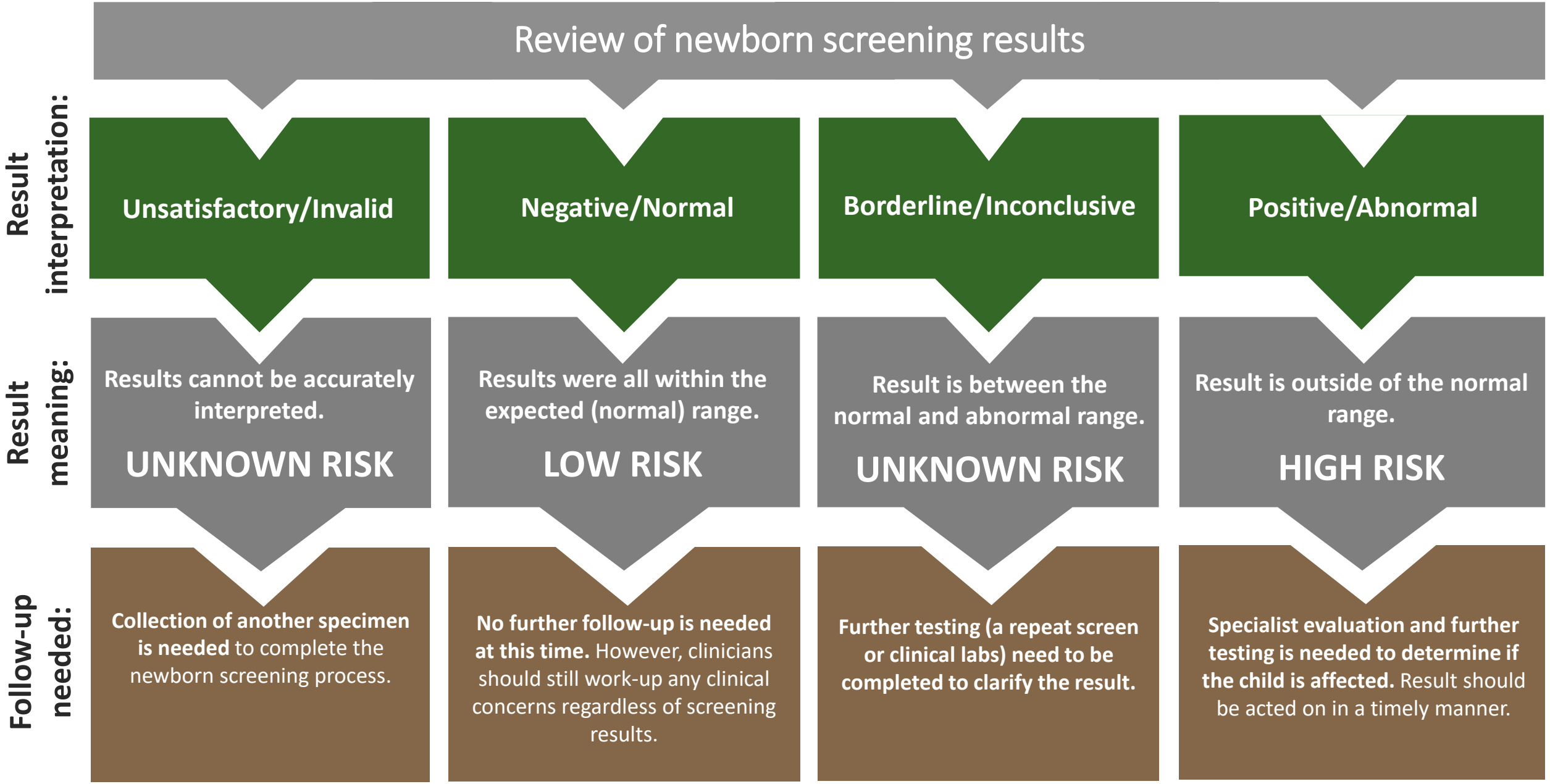
Unsatisfactory ≠ Borderline

Unsatisfactory result

- The specimen was deemed invalid for accurate screening; results cannot be accurately interpreted
- Another specimen needs to be collected as soon as possible


Review of newborn screening results
Result interpretation:
Unsatisfactory/Invalid
Negative/Normal
Borderline/Inconclusive
Positive/Abnormal
Result meaning:
Results cannot be accurately interpreted.
UNKNOWN RISK
Results were all within the expected (normal) range.
LOW RISK
Result is between the normal and abnormal range.
UNKNOWN RISK
Result is outside of the normal range.
HIGH RISK
Follow-up needed:
Collection of another specimen is needed to complete the newborn screening process.
No further follow-up is needed at this time. However, clinicians should still work-up any clinical concerns regardless of screening results.
Further testing (a repeat screen or clinical labs) need to be completed to clarify the result.
Specialist evaluation and further testing is needed to determine if the child is affected. Result should be acted on in a timely manner.

How to provide high-anxiety news

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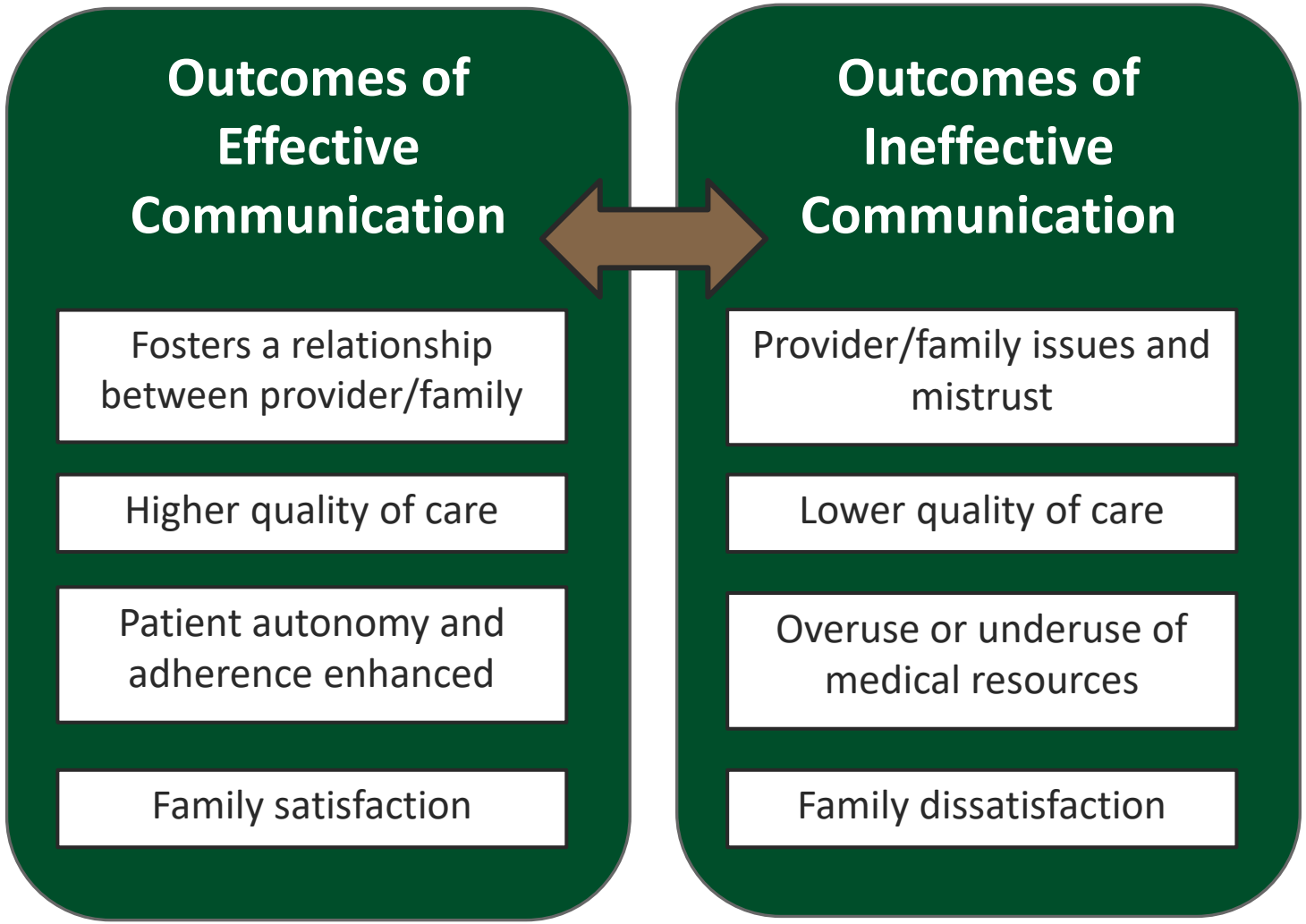
How did you feel when you were told about your child's newborn screening results?

"I felt that my world had ended and I had failed my daughter by not giving her what she needed. That people would see her as "less than"."

"I understood that she would not have a life that would be easy after they listed all of the symptoms she would have."

"I was completely overwhelmed. We experienced anxiety, fear, and overall a profound sadness that our beautiful baby boy had an unexpected, life-altering diagnosis."



Adapted from Rocque, R. 2015. PLoS ONE 10(10): e0139577

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Advisory Committee on Heritable Disorders in Newborns and Children: *SCREEN* Communication Guide

- Because this type of communication is not a routine activity for the primary care provider, this communication guide may be used to help frame the discussion with families to:
  - Improve understanding of the screening result
  - Increase adherence to follow-up recommendations
  - Enhance the family's overall experience with NBS
- Families who have had positive newborn screening results have suggested that the key points of the *SCREEN* communication guide are important in helping families cope with the uncertainty of a positive NBS result and understand the next steps needed to gain certainty

Your words become part of the family's story forever!

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Advisory Committee on Heritable Disorders in Newborns and Children: Communication Guide

- **S**hare the newborn screening result and disorder-specific information with the family
- **C**omprehension: assess the family's understanding/recall of newborn screening
- **R**eiterate what screening is and is not
- **E**ngage with the family and provide information at their desired level and pace
- **E**xplore the family's emotions
- **N**ext steps: discuss a shared plan and provide resources

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Share the newborn screening result and disorder-specific information with the family

- Inform family that you will be discussing findings from the newborn screen
- Help the family understand that a positive NBS result is serious, but that you are there to help guide them through the next steps regardless of your level of knowledge of the condition

Disorder-specific information

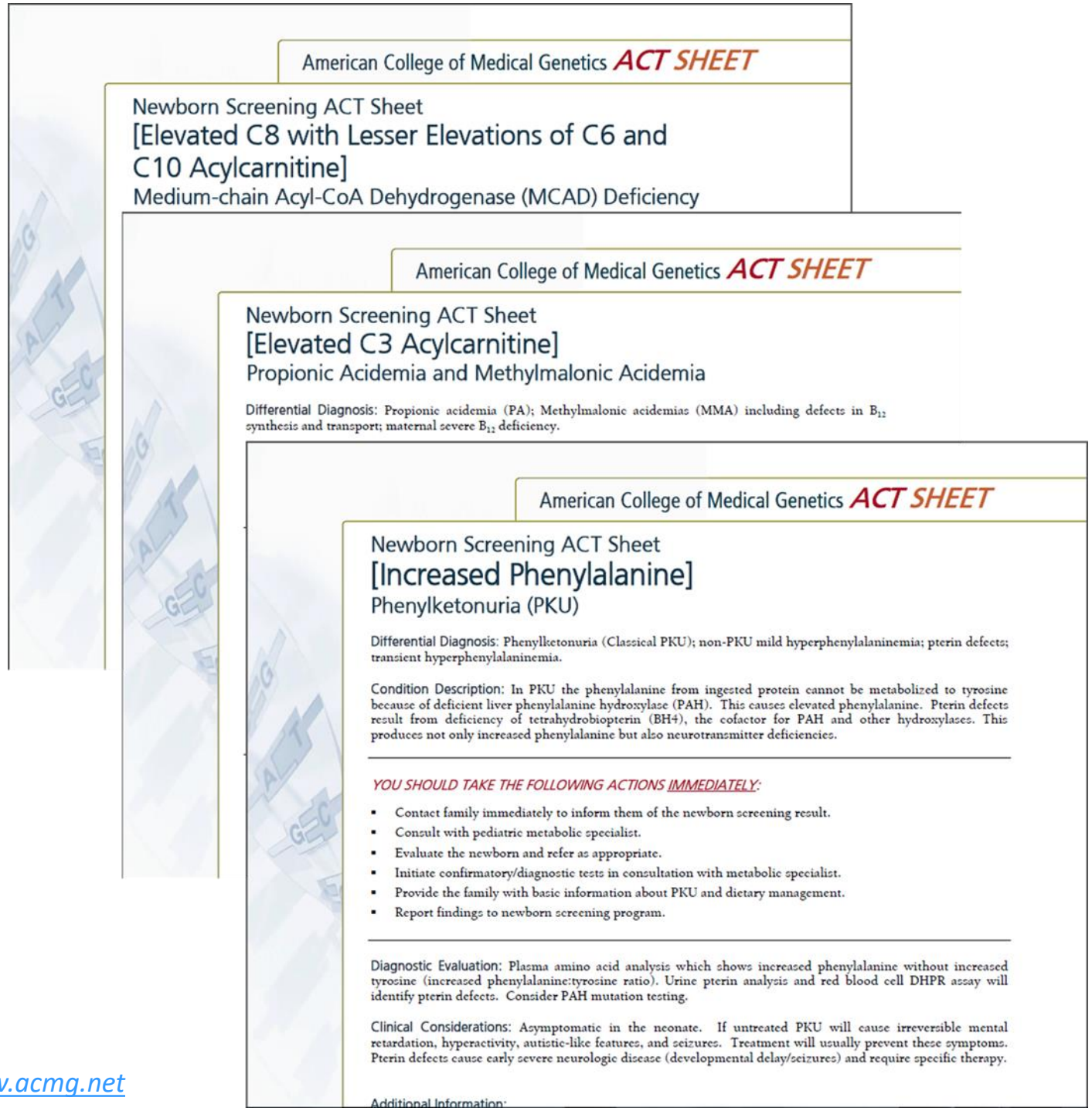
Providers find this to be most challenging component of receiving and communicating presumptive positive NBS results*

And they want information in words that they can use with the families

** According to MN Provider Survey completed as part of NewSTEPS New Disorders Grant*

ACT Sheets are available

- American College of Medical Genetics and Genomics (ACMG) developed ACTion (ACT) sheets, that:
  - Describe short term actions a health professional should follow
  - Determine the appropriate steps in follow-up
  - Include a diagnostic algorithm
- ACT Sheets freely available at www.ACMG.net



www.acmg.net

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sCREEN

Comprehension: assess the family's understanding/recall of newborn screening

- Assess if the family recalls and understands the process of NBS

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Resources for families

Recommended resources:

- Basic facts about newborn screening and what to expect from the newborn screening process
- State-specific newborn screening program information for parents and health professionals, including conditions on state panels, state program contacts, and support resources
- Condition-specific information, including causes, treatment, and family experiences
- An interactive blog that provides an open space for users to learn from one another and explore opportunities in education and policy (*Baby's First Test Only*)

HRSA Newborn Screening Information Center (https://newbornscreening.hrsa.gov/)

Baby's First Test (www.babysfirsttest.org)

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Reiterate what screening is and is not

- Remind the family about the purpose of NBS; it is a risk assessment and not a diagnostic test, so it is important that timely follow-up confirmatory testing be done

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SCR**E**EN

Engage with the family and provide information at their desired level and pace

- Offer to provide the family additional result-specific information
  - May be provided by the state NBS program (ask for these)
  - HRSA Newborn Screening Information Center & Baby's First Test
- Discuss information using non-medical terms, at the family's pace and desired level of detail

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Explore the family's emotions

- Explore with the family how they might use their support system or other support resources now and as they go through the diagnostic process
- Remember there is a wide spectrum of how families may cope with this result (anxiety to denial)
- Tailor your discussion to help the family hear and retain the information discussed

Table 2. Examples of empathic, exploratory, and validating responses

Empathic statements	Exploratory questions	Validating responses
"I can see how upsetting this is to you."	"How do you mean?"	"I can understand how you felt that way."
"I can tell you weren't expecting to hear this."	"Tell me more about it."	"I guess anyone might have that same reaction."
"I know this is not good news for you."	"Could you explain what you mean?"	"You were perfectly correct to think that way."
"I'm sorry to have to tell you this."	"You said it frightened you?"	"Yes, your understanding of the reason for the tests is very good."
"This is very difficult for me also."	"Could you tell me what you're worried about?"	"It appears that you've thought things through very well."
"I was also hoping for a better result."	"Now, you said you were concerned about your children. Tell me more."	"Many other patients have had a similar experience."

Adapted from Baile, W. et al. SPIKES—A Six-Step Protocol for Delivering Bad News: Application to the Patient with Cancer. The Oncologist 2000;5:302-311

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Next steps: discuss a shared plan and provide resources

- Discuss with the family a shared plan that is concrete, specific, and includes the following:
  - Where, when, and with whom is the next appointment
  - What testing will be considered and/or done
  - What should they watch for in their child while they wait?
  - Who can they contact if they have additional questions or concerns?
- Assess the family's understanding of the visit and information provided using teach-back methods, and provide valid websites for them to get more information.

So… now what?

Next steps for PCPs

- Determine clinical status of baby
- Consult with specialist if necessary
- Initiate timely confirmatory/diagnostic testing
- Utilize ACMG ACT sheets/algorithm
- Educate family about signs, symptoms, and when urgent treatment may be required
- Report findings to newborn screen program

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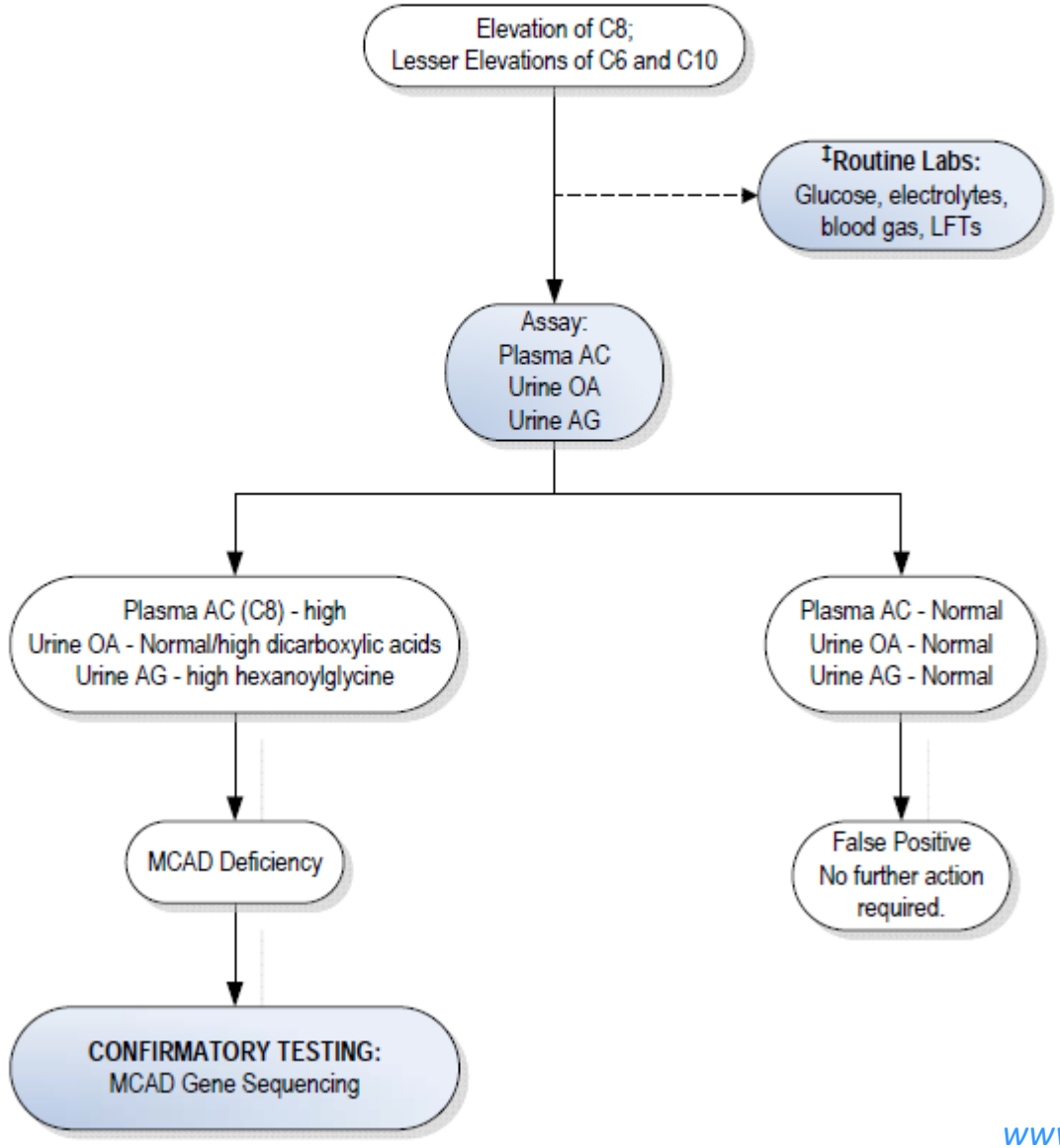

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Perspective and Questions