Newborn Screening News

Vol. 8 Issue 1 February 2007

Newborn Screening Report Format Change
Frequently Asked Questions

The addition of 20 new disorders to the DSHS newborn screening panel necessitated a change in the result reporting format. Some Frequently Asked Questions (FAQ's) are listed below to assist you in reading the new format.

Q. Where are results for the original disorders (PKU, CAH, etc.)?

A. Disorders are now being listed by type of disorder instead of by individual names. PKU is within the group of ***Amino Acid Disorders***, CAH and T4 are within the ***Endocrine Disorders,*** and information on Hb F / Hb A are within the H***emoglobinopathies***. ***Biotinidase Deficiency*** and ***Galactosemia*** are listed by name.
Refer to the section at the bottom of the result report for a complete list of all disorders screened.

Q. Where will abnormal values be listed?

A. On the mailed report, additional columns will be displayed with information on the abnormal analytes and recommended actions. In addition, you will be contacted by telephone or FAX by the Newborn Screening Case Management staff if any of the newborn screening results are abnormal.

Q. Do I have to read all of the information at the bottom of the report?

A. The first block of information contains important messages for providers which should be reviewed periodically for updates. The last paragraph on the report lists all 27 disorders in the DSHS newborn screening panel. This list will not change unless another test is added.

Note: You will be notified of any abnormal NBS result by telephone and FAX prior to receiving the mailed report. At that time, Newborn Screening Case Management nurses will provide specific instructions on necessary actions.

Q. What are analytes?

A. An analyte is the specific enzyme, chemical, or molecule that is tested for and measured to determine if a child has a NBS disorder. Many of the analytes are listed by name. For example, phenylalanine is the analyte measured which indicates PKU and thyroxine (T4) is the analyte measured which indicates hypothyroidism. However, Fatty Acid and Organic Acid disorder analytes are listed by abbreviations of the acylcarnitine markers or ratios that identify the disorder (i.e. C8, C6, C10:1, C10, C8/C2 are the analytes that are measured and may indicate the MCAD disorder).

Q. Are we responsible for ordering the plasma acylcarnitine profile, urine acylglycine, and urine organic acid tests, or will the specialist order them? If we have to order the tests, what laboratory do you suggest we use?

A. The suggested confirmatory tests are listed so that if you wish to order the tests before the specialist is consulted, you can do so. Some people prefer to consult with the specialist first and have the specialist order the tests. The laboratory used will depend on insurance requirements and local availability.

Q. Why do some abnormal results only require a repeat screen and others require multiple laboratory tests?

A. The requested or recommended actions are based on levels of the abnormal analytes. The majority of abnormal analyte results only indicate the need for a repeat screen; however some results are within a critical range and require immediate action to prevent or control serious health problems.

Q. When an abnormal result note states "Possible metabolic disorder", what does this mean?

A. Sometimes the screen results do not provide enough information to determine the exact disorder that may be indicated. Typically, the results do not indicate a critical situation so the action needed is to provide a repeat screen as soon as possible.

Q. Why does the 'Screening Result' indicate "Abnormal" but some analytes are listed as "Normal" in the 'Analyte Result' column?

A. Multiple analytes are used to determine a patient's risk for some disorders. Not all the analytes have to be abnormal to indicate a problem.

Q. Why do the result notes indicate a possible MCAD and recommend a specialist for further tests, and also a General Elevations(s) with a request for a repeat newborn screen? Which directions am I supposed to follow?

A. Depending upon the specific analytes that are elevated, multiple disorders may be indicated. Therefore multiple screening notes may be printed. NBS case management staff will contact you to provide guidance on necessary actions and follow-up.

Q. I received a 2 page abnormal result report but the second page was blank. Is there information missing?

A. We want to make sure the section with general messages for providers accompanies each result report. If the message cannot fit on the first page, a second report page is automatically generated to provide the information.

Q. Some screening result notes mention TPN and General Elevation(s). What do those terms mean?

A. TPN stands for total parenteral nutrition – the babies nutrition (amino acids, sugar and lipids) is given intravenously. General Elevation(s) refers to abnormal analyte(s) that are not recognized as a specific pattern associated with one of the screened disorders.

Q. The result for hemoglobinopathies states "awaiting DNA studies". Do I have to order the study?

A. As part of the NBS program, DNA testing is automatically performed on specimens that test positive for specific hemoglobinopathies. Specimens that will have the DNA testing performed will have the "Awaiting DNA studies" note on the NBS report. The DNA report will be mailed separately when the testing is completed.

Q. Are there any other changes associated with expansion of which we need to be aware?

A. NBS specimen collection cards have also been updated. The first page of the new card is a perforated information sheet that should be torn off and given to parents to take to the first well-child visit. In addition, there are new demographics fields including: time of birth and time of specimen collection; information about the physician that will follow the child after hospital discharge and the physician's NPI number.

Note: The new collection cards are slightly larger than previous years. If you order NBS shipping envelopes from DSHS, please request the larger size.

For more information please visit
http://www.dshs.state.tx.us/lab/newbornscreening.shtm
Or call 1-888-963-7111 ext. 7333
Email: newborn@dshs.state.tx.us

TEXAS Department of State Health Services
Publication Number 05-10897