Laboratory Test Stewardship in Evaluation of Mass Spectrometry Assay for Congenital Adrenal Hyperplasia Screening in Pediatric Hospital

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**Background**

Congenital adrenal hyperplasia (CAH) is a debilitating disease requiring early diagnosis to prevent long-term disability and even death. CAH results from deficiency of enzyme or enzymes involved in production of steroid hormones. Hence, screening for CAH requires quantitation of several steroid hormones. There is no cure for CAH but early intervention can dramatically improve health and development of affected patients. Newborn screening utilizes an immunoassay that measures 17-hydroxyprogesterone and its elevation suggests 21-hydroxylase (21-OH) deficiency, the most common form of CAH. Follow-up requires measurement of steroid hormones. Liquid chromatography -mass spectrometry (LC/MS/MS) quantitation is often used because of its sensitivity.

At our institution, we offer in-house testing for 11- and 21-hydroxylase deficiency and a send-out option for a comprehensive CAH profile. Send-out CAH screening test has a turn-around time of 5-6 days while in-house testing has a faster turn-around time of 2 days. Our institution also has developed an automated system, EQL, which alerts pathologists/clinical chemists about specific physician test orders. In tandem with LC-MS/MS analysis, we recently employed EQL alerts when CAH send-out test is ordered. After review of the patient chart and discussion with the ordering physician, the test is either approved for send-out, modified for in-house testing or rejected.

We screened 140 tests in 3 months since utilizing automated alerts. We found 19% of the tests were modified to in-house testing and 1% was rejected. These changes resulted in approximately $25,000 of savings by utilizing in-house MS CAH screening. Our results showed that clinical application of MS in tandem with Laboratory Utilization strategies can improve turn-around time, result into savings and quicker patient management, especially for children that present with symptoms in the ER or NICU.

**Laboratory Test Stewardship Strategy**

<table>
<thead>
<tr>
<th>Order Request</th>
<th>E-mail alerts</th>
<th>Specimen Management Pathologists/Clinical Chemists</th>
<th>Decision</th>
</tr>
</thead>
<tbody>
<tr>
<td>Physicians/ Nurses</td>
<td></td>
<td></td>
<td>Send-out, In-house, Reject</td>
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**Results**

- Decreased turn-around time from 5 to 2 days
- 140 CAH Screening order in 3 months
- $25,000 in savings

**Summary**

- Utilizing in-house mass spectrometry for CAH screening resulted in:
  1. Decreased turn-around time from 5 to 2 days
  2. Monitoring test utilization for 3 the first three months resulted in $25,000 in savings

Most modern clinical laboratory utilizes mass spectrometry. Adding appropriate clinical tests in mass spectrometry can improve turn-around time that allows clinicians to render therapy earlier compared to when tests are sent-out.

We used automated alert system, EQL, to improve test utilization of in-house MS CAH screening assay. Using this automated alert system enabled pathologists/clinical chemists to review test orders promptly and avoid delay of testing.

The send-out test requires only 0.5 mL of plasma while our in-house test require’s 1.5 mL. Furthermore, our in-house screening measured only 5 steroids while the reference lab for our send-out tests measures 9 steroids. Improving our existing MS CAH assay to match the send-out test will be clinically and financially valuable.

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**References**

White PC. Nat Rev Endocrinol 2009 Sep; 5(9):490-8

Minutti CZ et al. J Clin Endocrinol Metab. 2004 Aug; 89(8): 3687-93