

In this issue, we discuss a question we were asked by health practitioners on the difference between MS/MS and GC/MS for newborn screening (NBS).

MS/MS vs. GC/MS

Q: We recently came across an organization that is offering NBS services using GC/MS. Given that every comprehensive NBS program worldwide uses MS/MS, do you know the reasons why MS/MS is more popular and what are its advantages over GC/MS?

A: The technologies are complementary and at a high level accomplish the same end result but the processes are different.

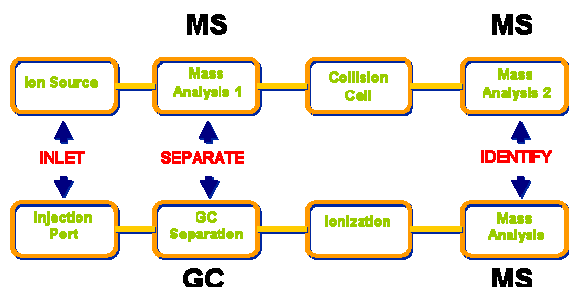


Diagram from "The Impact of Tandem Mass Spectrometry Newborn Screening on the Presentation of Metabolic Disorders" by Joseph Muenzer, M.D., Ph.D., Department of Pediatrics, UNC, NC, USA

#		MS/MS	GC/MS
3	Sample	Dried Blood Spots (DBS). Universal standard for screening.	Urine. Variability in concentration. Good for confirmation but NOT for screening.

Interpretation – This is What Matters!

There are many factors that can affect a result, ranging from collection and storage of the dried blood spot (pre-analytical) to interpretation (post-analytical). Ultimately, the clinical utility of a screen comes down to the ability to consistently provide an accurate result.

We have five documented cases where a sample analyzed by NeoGen Labs was also sent to other laboratories by the hospital or a physician. In all five of these cases, our results correlated with the final clinical outcome. In four cases, the competing labs reported the result to be positive or "may have" a disorder. Our result for the same four cases was negative, saving parents unnecessary anguish. In the fifth case, we detected a disorder which one of the other laboratories called normal, saving a newborn's life.

We believe we have the best interpretation technology available in the world, developed from a database of over 3 Million babies of all ethnicities.

Newsletters

Past newsletters can be accessed at, http://www.neogenlabs.com/dr_res.shtml

We have covered various IEMs, cases and other related issues. You can access missed issues here. The April 2008 issue is particularly important since it discusses reporting by NeoGen Labs.

November 2008 Statistics

- 1 Case of UCD
- 1 Case of LCHAD
- 1 Case of MSUD
- 1 Case of SCAD
- 1 Case of G6PD

Screening Panels

- **First Step** (Over 50 IEMs for Rs. 3975)
- **First Step MS/MS** (45 IEMs, includes Fatty Acid Oxidation Disorders, Amino Acid Disorders, and Organic Acid Disorder panels for Rs. 3250)
- **First Step Bio** (5 IEMs which include CH, CAH, G6PD, GALT and Cystic Fibrosis for Rs.1500).

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#		MS/MS	GC/MS
1	Sensitivity and Specificity for routine screening	Very high specificity, sensitivity and accuracy (supported by > 200 published scientific papers). No false negatives. False positive rate < 0.1% (using technology licensed from Perkin Elmer, Bridgeville, PA, USA, formerly Pediatrics Screening).	Unknown – has never been used in a routine newborn screening program. High probability of false negatives and false positives when used for screening (urine can be concentrated or dilute). However, if index of suspicion is high for a specific disorder then urine is the preferred sample for confirmation.
2	Operational Ease	MS/MS requires minimum clean up and maintenance. No columns required for separation.	GC columns can become fouled with non-volatile material extracted from the samples affecting results.