

## Abstract

The Utah Department of Health (UDOH) Newborn Screening (NBS) program is a public health program administered under Utah Statute 26-10-6 that aims to give all babies born in Utah the best chance at lifelong health. The infant's blood is collected on Guthrie filter paper cards and then punched into 3.2mm dots to be separated for different testing. Utah currently screens for over 40 disorders, but new tests are added frequently. The criteria for adding a test is that it must have an available screening method and successful treatment options. I assisted in the Mass Spectrometry (MS) laboratory where we screen for many metabolic disorders. MS has had a tremendous impact on NBS due to the ability to screen for multiple disorders at once and by keeping costs lower for families. In the MS laboratory I helped prepare samples, maintain the instruments, run the biochemical assays, and interpret results and discuss those results with the follow up team.

## Internship Activities

Since babies are born everyday, so too are samples collected everyday. When samples are received by the Department of Health, they are checked to make sure there is a sufficient amount of blood for the required tests. Then it is 'punched' into 3.2mm dots on 96 well plates and separated for each assay. In the Mass Spectrometry lab, I would quality check the blood spots sent down and then begin testing with my fellow lab techs. Once extractions were completed through various incubations, the plates were then loaded on the mass spectrometers and run under the correct method parameters. Once the plate was finished, I would review the results, along with the controls on the plate, and note any abnormal samples to inform the follow up team about. In cases where it was a close call about if it were abnormal or not, I would perform various calculations to correct for background noise and work with my supervisor and the follow up team to determine if further testing was needed.



Figure 2: SCIEX Citrine mass spectrometer.

## Acknowledgements

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Figure 1: Blood collected on a Guthrie card through a heel prick.

## Techniques

Mass Spectrometry is an analytical technique used to measure the  $m/z$  ratio of ions in order to identify and quantify molecules in simple and complex mixtures. In the newborn screening mass spectrometry lab, we utilize flow injection analysis (FIA), multiple reaction monitoring (MRM), and liquid chromatography tandem mass spectrometry (LC/MS-MS). All of these techniques helps us to specify where the instruments should look for the molecules of interest. All of the mass spectrometers used in the lab are triple quadrupole instruments which also allows for greater selectivity. In this lab I not only learned about how triple quadrupole instruments works, but also how to troubleshoot possible problems and fix minor damages.



Figure 3: Example MRM read out.