Introduction
Argininosuccinic aciduria (ASA, OMIM 276900) also known as argininosuccinic aciduria is an inborn error of metabolism caused by deficiency of the enzyme argininosuccinate lyase. The clinical presentation of ASA is variable and ranges from neonatal to adult phenotypes. The condition is identified by the accumulation of argininosuccinic acid in the urine and characterized in 1959 and 1960.

The condition, eventually identified as ASA, was first described in 1958 by R.G. Westall. 

Westall.

In the neonatal onset form of ASA, ammonia accumulates in the blood and, babies in the USA and is included on the newborn screening panel in all 50 states.

The urea cycle disorder, argininosuccinic aciduria (ASA), occurs in 1/70,000 live births. The condition is identified by the accumulation of argininosuccinic acid in the urine and characterized in 1959 and 1960.

The condition, eventually identified as ASA, was first described in 1958 by R.G. Westall.

Other laboratories have used a variety of different internal standards, including Arg IS = L-Arginine (5-$^{13}$C$_{4}$N$_{4}$) shown on the left. The corresponding $m/z$ 140

Note that the mass difference between CIT and ASA.

Known Sample Tests
Samples from previously identified and diagnosed patients with classic ASA (CT) and asymptomatic babies (WI) in the Connecticut State Labs were analyzed using the new validated method. Three known samples included one conditional ASA (ASA-1) sample, two unaffected family members and one known normal control sample. The new validated method includes stable labeled argininosuccinic acid and $^{13}$C$_{15}$N$_{15}$ successfully differentiates between CT and WI.

Blind Samples Tests
Eight dialyzed and dialyzed samples from patients with conditional diagnosis of argininosuccinic aciduria (ASA) and 5 patients with CT (T) as well as samples from normal newborns tested by Wisconsin State Lab as blind samples to test the new validated method of Connecticut State Labs. The next validated method, using Cambridge Isotope Laboratories stable labeled argininosuccinic acid enabled differentiation among samples measuring high concentration of argininosuccinic acid.

Conclusion
The new validated method, argininosuccinic acid enables differentiation among samples measuring high concentration of argininosuccinic acid.

Additional Information
Please see the accompanying handout for additional information.

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