



## Branched Chain Amino Acids in DBS LC-MS/MS Analysis Kit

Maple syrup urine disease (MSUD) is an innate metabolic problem caused by a deficiency of the branched-chain ketoacid dehydrogenase (BCKDH) complex. MSUD is characterized by the accumulation of the branched-chain amino acids (BCAA) leucine, isoleucine, alloisoleucine, valine in urine, blood and spinal fluid and is a rare autosomal disease. For quantitation of branched amino acids in DBS samples LC-MS/MS based methods are useful for rapid confirmatory diagnosis when MSUD patients are diagnosed by clinical findings or family history. Detection and quantification of Allo-isoleucine in DBS samples of potential MSUD cases, in conjunction with the determination of the leucine/phenylalanine ratio, would be a useful approach to shorten the time during the confirmatory diagnostic process.

### Highlights of the Analysis Kit

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Simultaneous quantitation of leucine, isoleucine, allo-isoleucine and valine



Total analysis time 10.0 min.



Just a few pipetting steps for sample preparation; “**extract and shoot**”



Safeguarded by its own stable labelled isotope as internal standard



Long life span of HPLC column

**Parameters**

Leucine, Isoleucine, Allo-isoleucine and Valine

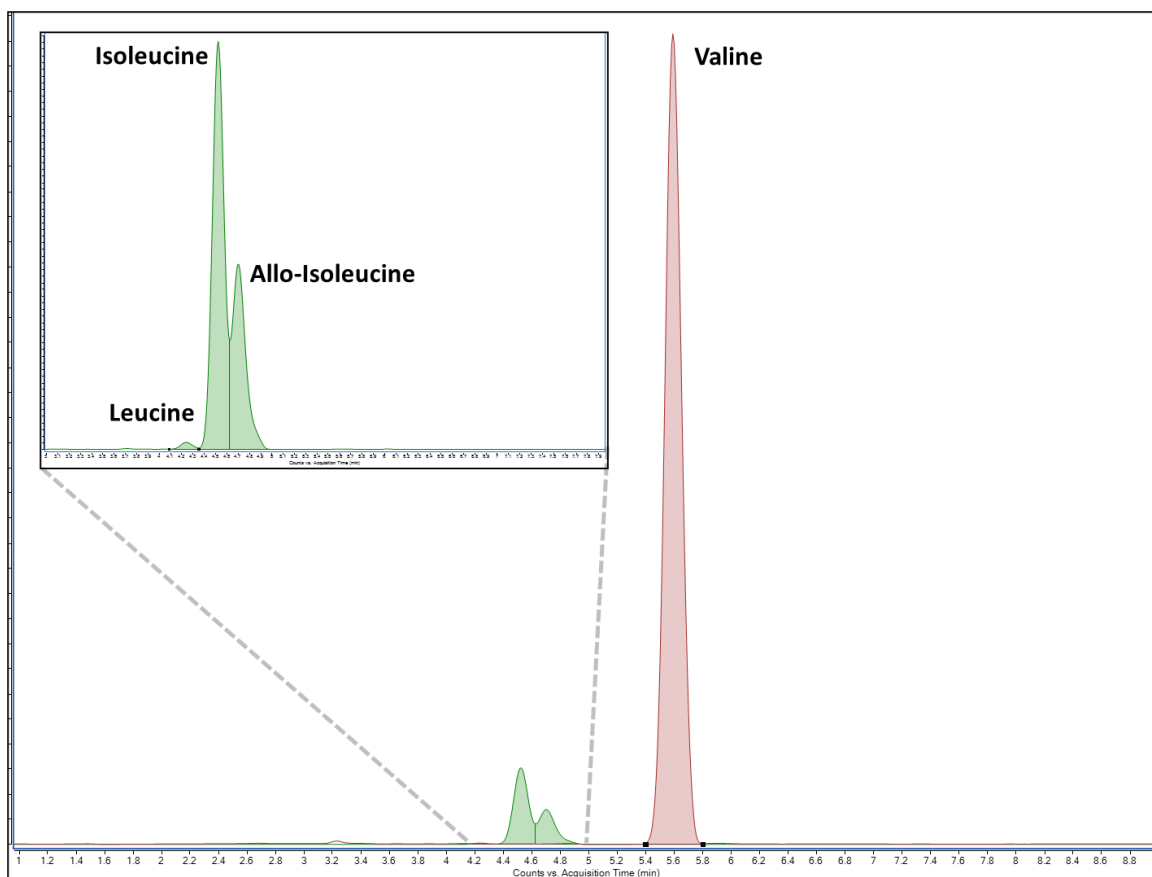
**Sample Type**

Dried Blood Spot (DBS)

**Sample Preparation**

1	Punch 3 mm of blood spot into a conical microtube
2	Add 200 µL of extraction reagent (containing internal standard) and shake for 15 min. at room temperature
3	Pipette extract into a HPLC vial prior to injection

**Example Chromatogram**



Total ion chromatogram of the analyses

## Method Performance

All validation results were obtained using Agilent 6465 TQ (Ultivo) system

Analytes	LOQ (mg/l)	Linearity (mg/l)	Recovery (%)	Repeatability (%CV)
Leucine	7.35	33.5 – 679.4	98	0.46
Isoleucine	4.91	14.3 – 616.5	96	0.48
Allo-isoleucine	0.37	0.8 – 623.3	99	2.02
Valine	4.99	41.1 – 564.1	117	0.26



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