

# Development of an automated method for the measurement of leucocyte cystine by LCMS

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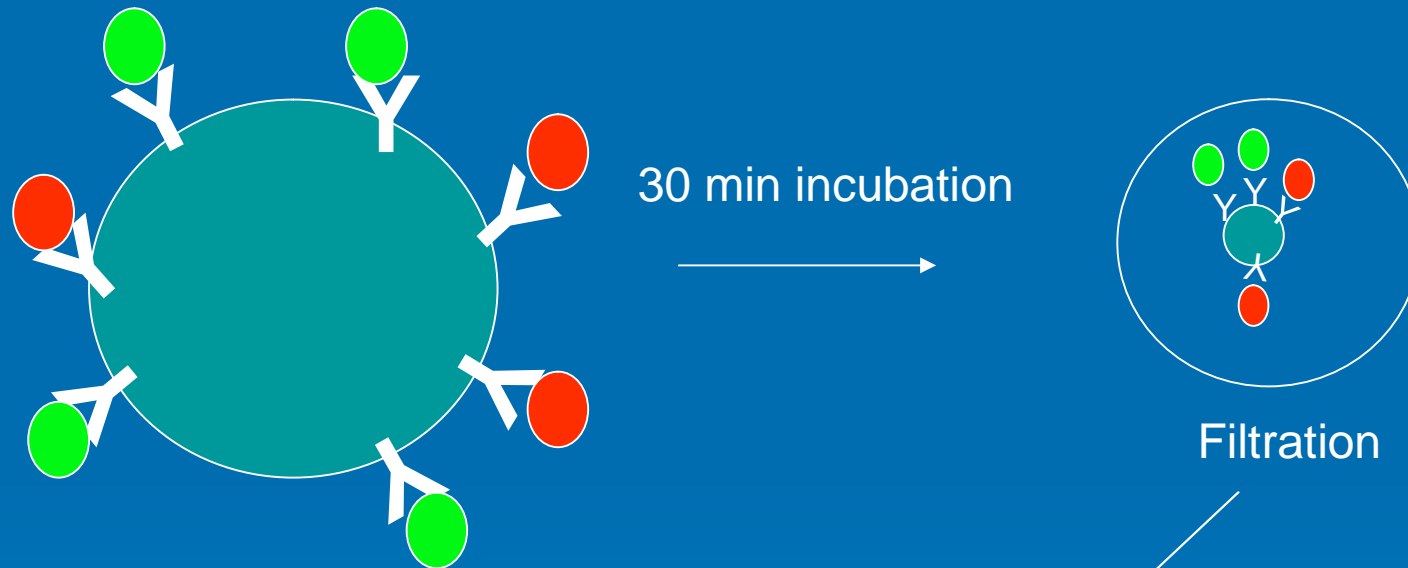
# Cystinosis

- Rare autosomal recessive lysosomal storage disease
- Defect in the lysosomal membrane protein cystinosin
- Infants may present with failure to thrive and begins to show signs of Fanconi syndrome
- Treatment by cysteamine

# Current laboratory method- Cystine binding protein (CBP) method

● Sample

●  $^{14}\text{C}$  - cystine



CPM

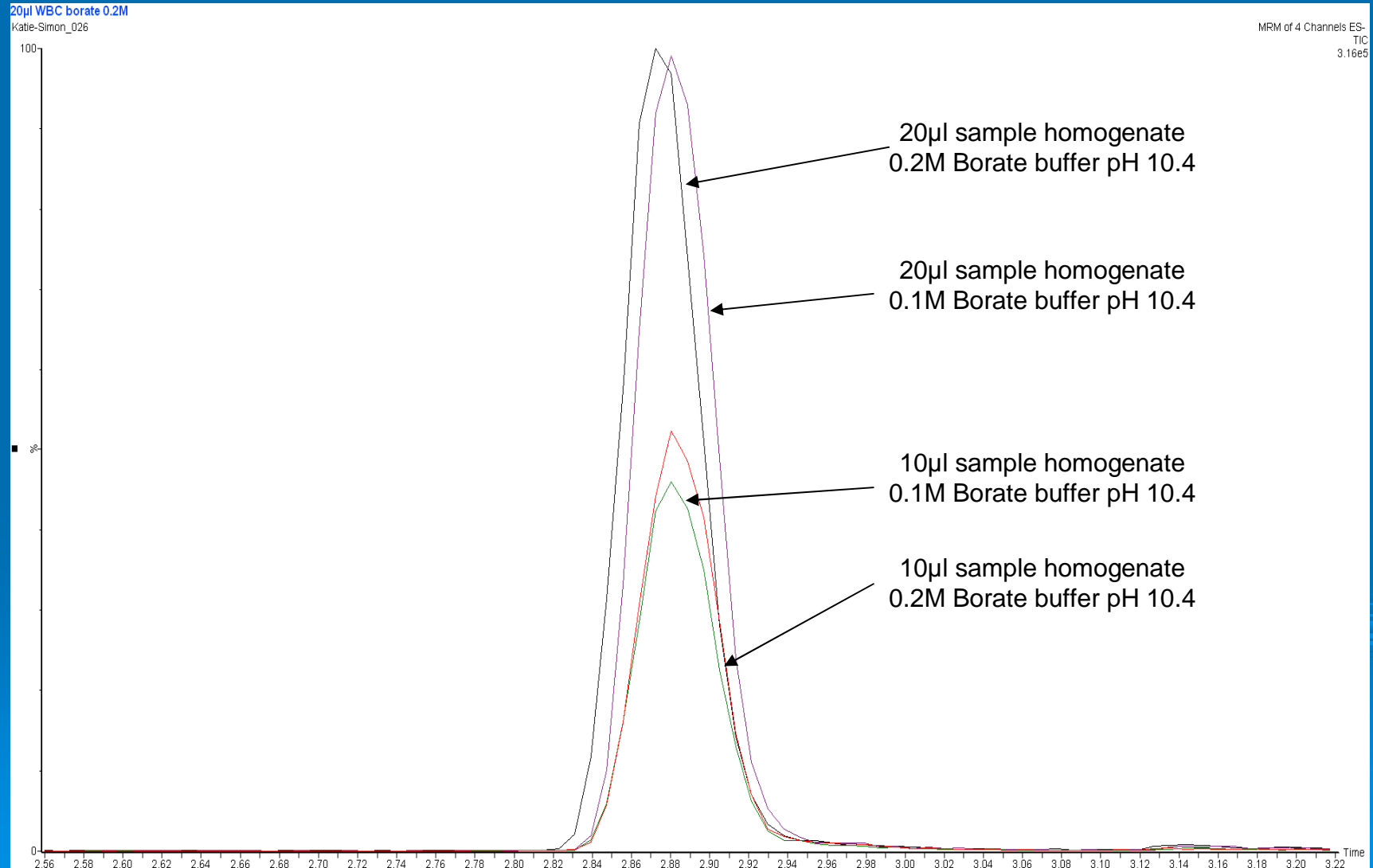
Scintillation counting

[cystine]

# LC-MS method (in brief)

- Sample preparation
- 10µl of sample + 10µl d3-cystine
- 125 µl 0.2M borate buffer, pH 10.4,
- 125 µl 6M derivative
- HPLC-MS/MS analyses using a C18 guard column, 5 min per sample.

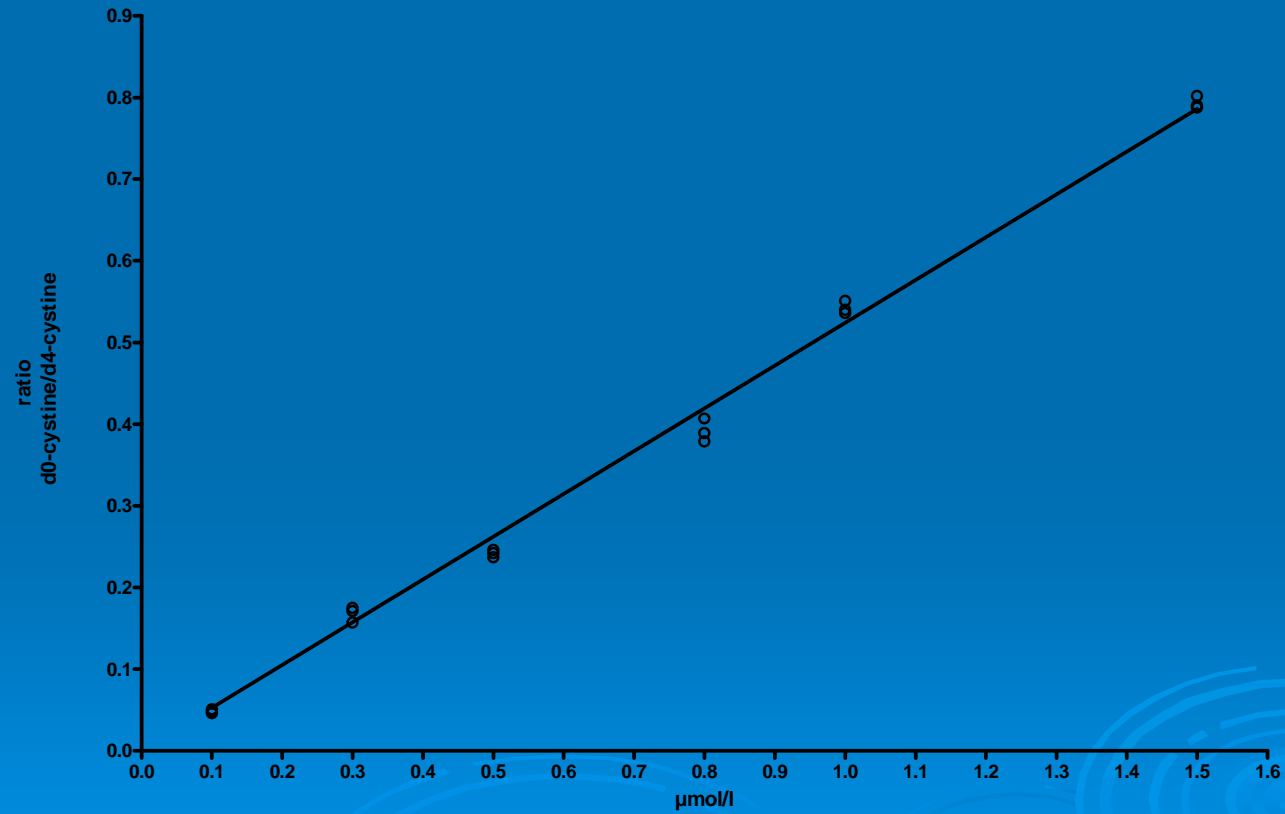
# Optimisation of method



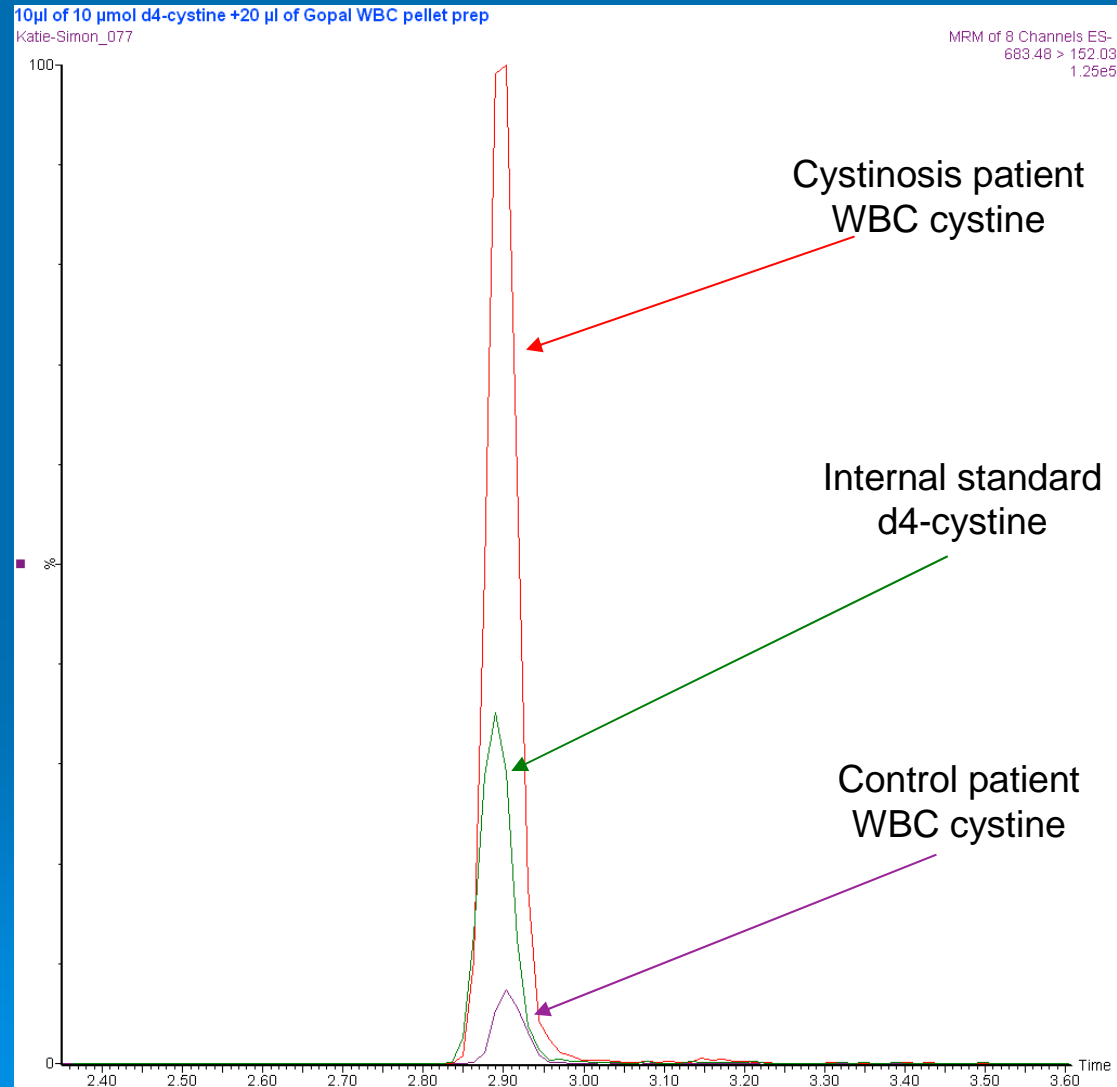
# Linearity

## Cystine calibration curve

683.83>152.03 transition  
(3 replicates for each point)



# Evaluation with patient samples

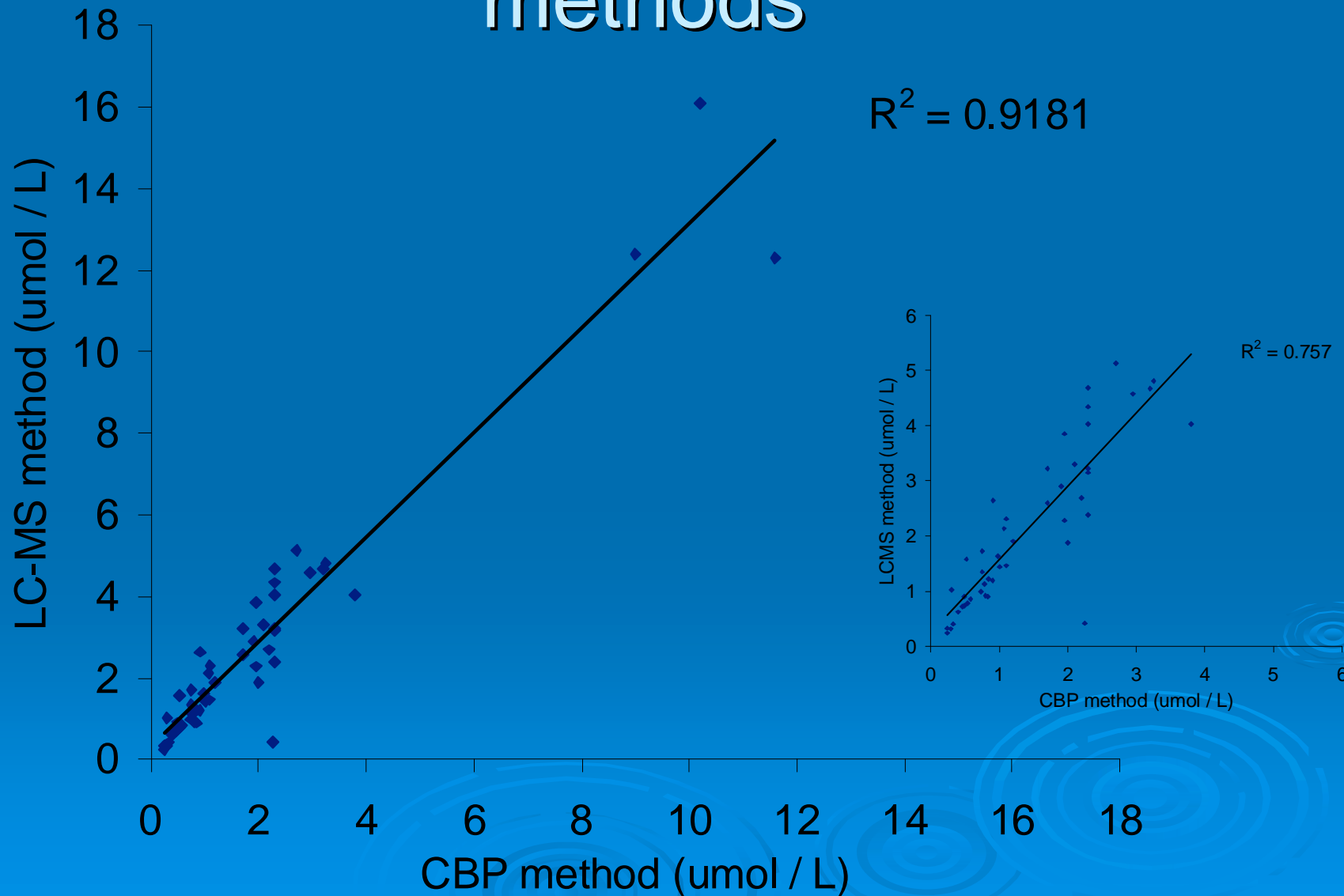


# Imprecision

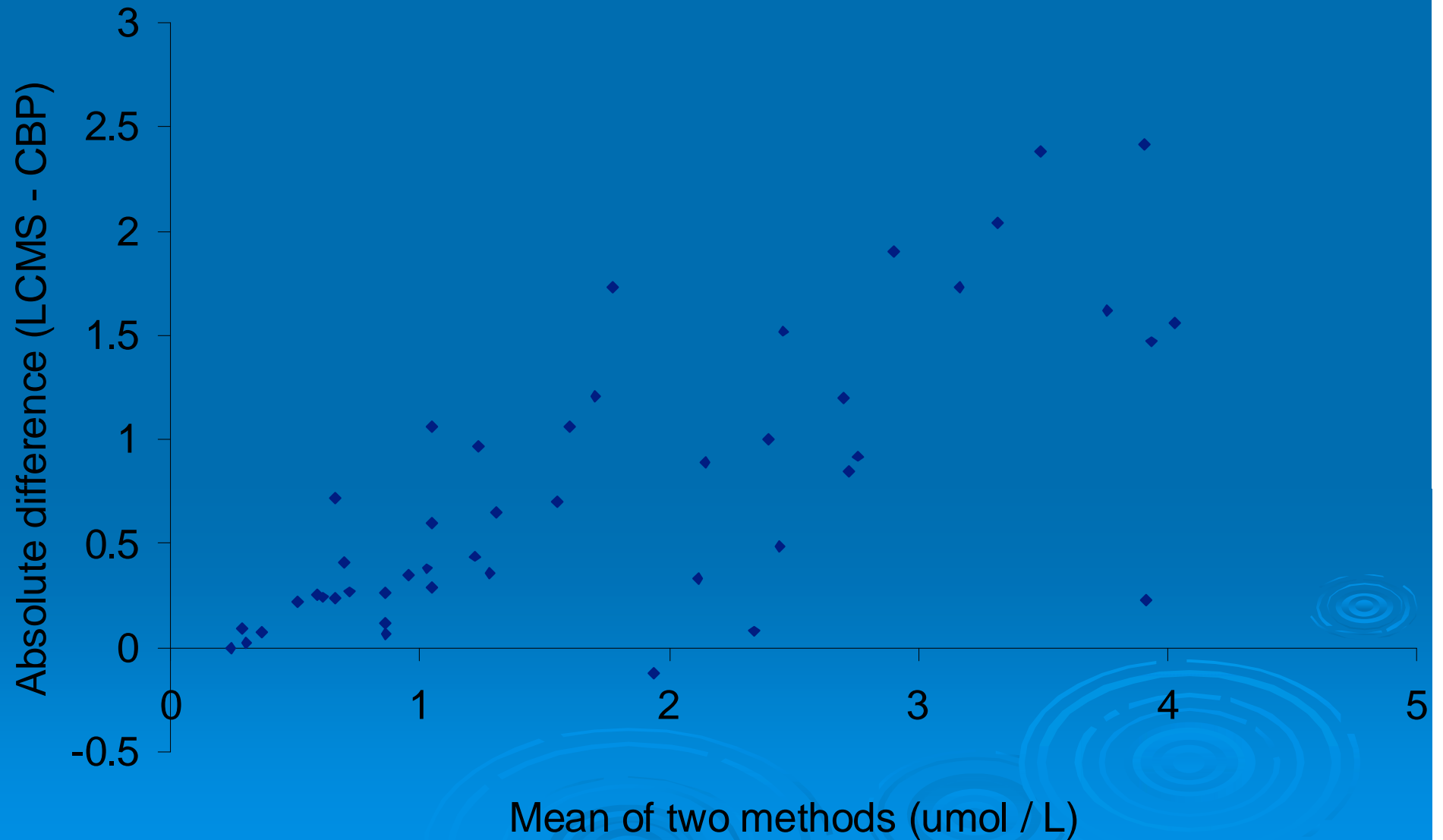
	Within batch %CV	Between batch %CV
Low QC (1.8 $\mu\text{mol} / \text{L}$ )	4.0 (n = 16)	5.8 (6 batches)
High QC (3.1 $\mu\text{mol} / \text{L}$ )	6.2 (n = 19)	5.7 (6 batches)



# Comparison of CBP and LCMS methods



# Bland Altman plot



# Acknowledgments

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