



Diagnostic Proficiency Testing Discussion common sample 2016

**ERNDIM workshop, SSIEM Rome
September 6, 2016
George Ruijter**



Patient information

Clinical info provided:

At the age of 5 years this boy was referred for the first time to a pediatric nephrologist, because of urolithiasis. At ages 7 and 10, again renal stones were found. At the time of the urine collection, he was 10 y old and in good health. He used no medication, had a normal diet and adequate renal function.

Diagnosis: Hyperoxaluria type 2 (GRHPR deficiency)

Sample provided by dr W Onkenhout & dr P Vos
(Leiden University Medical Centre, The Netherlands)

Patient information

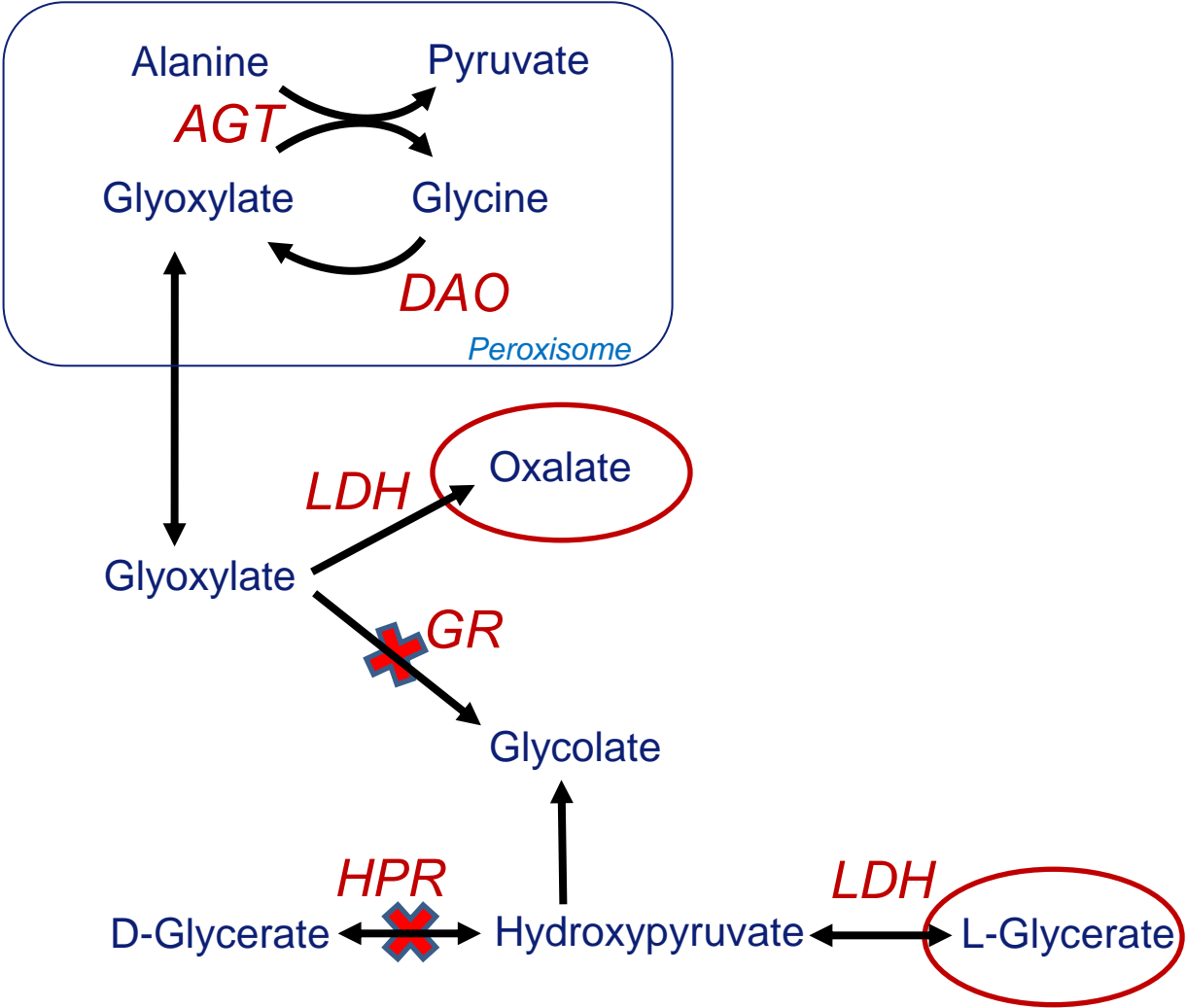
First metabolic workup:

	Patient	(Ref. values)
Amino acids	normal	
Purines	normal	
Oxalate	234	0-110 mmol/mol creat
Glycerate	2448	0-30
Glycolate	61	0-90

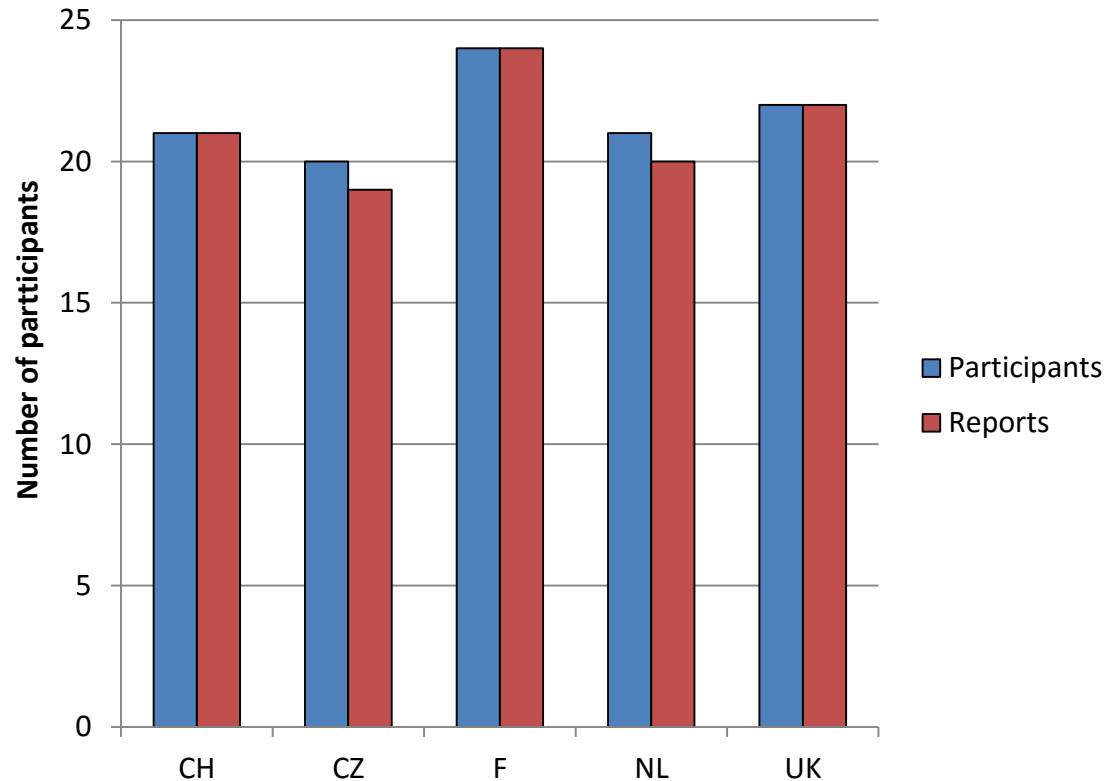
Mutation analysis *GRHPR*: homozygous c.103delG (p.Asp35ThrfsTer11)
Common mutation

Enzyme activity: not performed

Metabolism oxalate/glycerate/glycolate

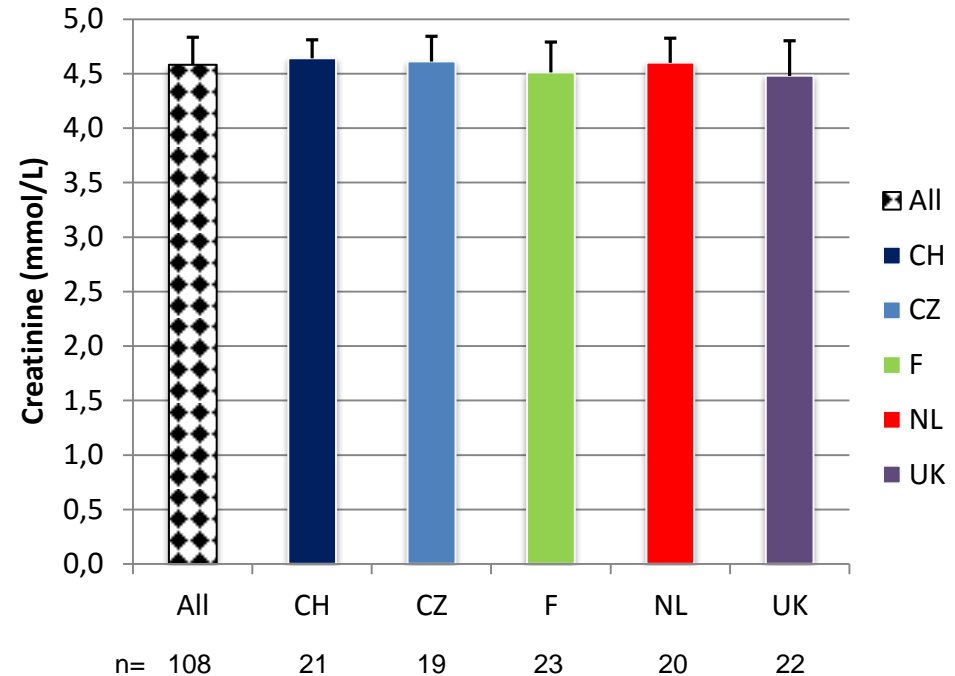
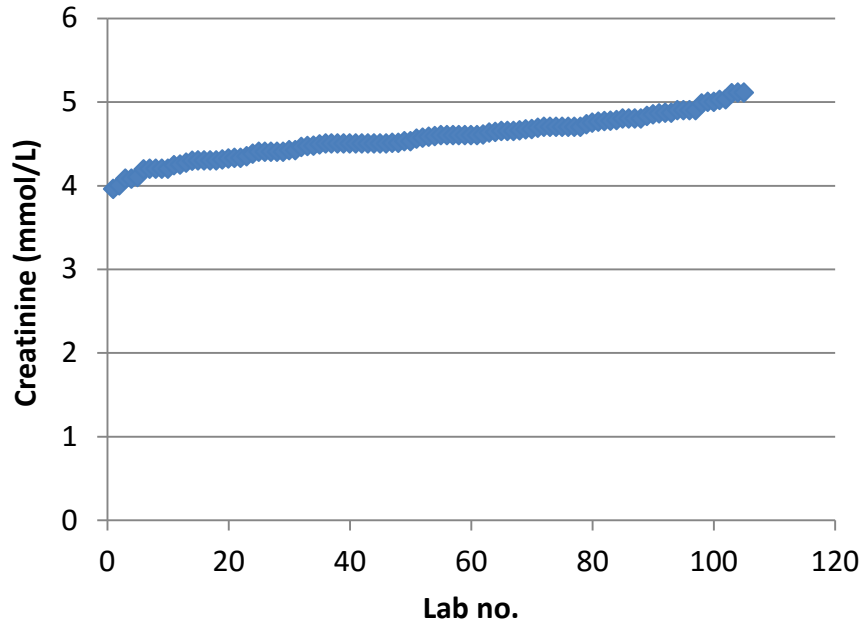


Diagnostic Proficiency Testing participants



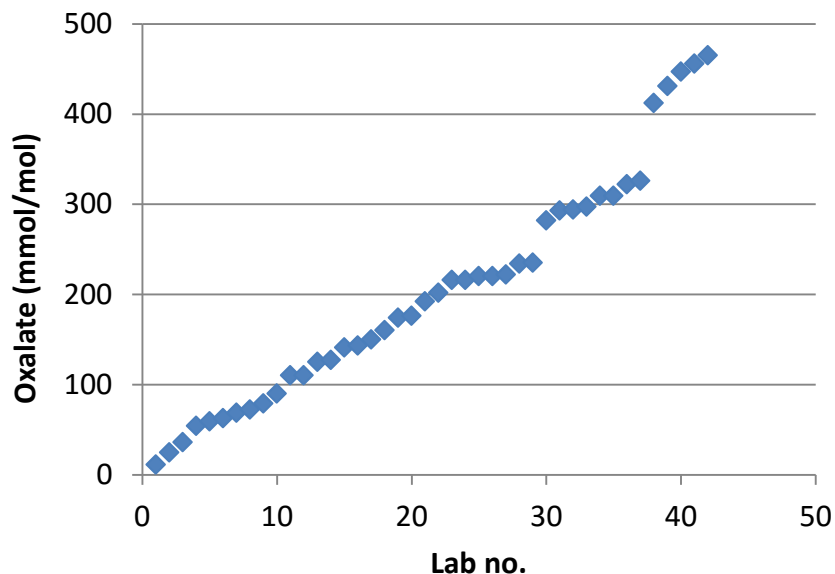
108 participants
106 reports

Creatinine results



Creatinine: 4.58 ± 0.25 mmol/L
(CV 5.5 %; similar to SA Urine)
(1 outlier removed; 8.9 mmol/L)

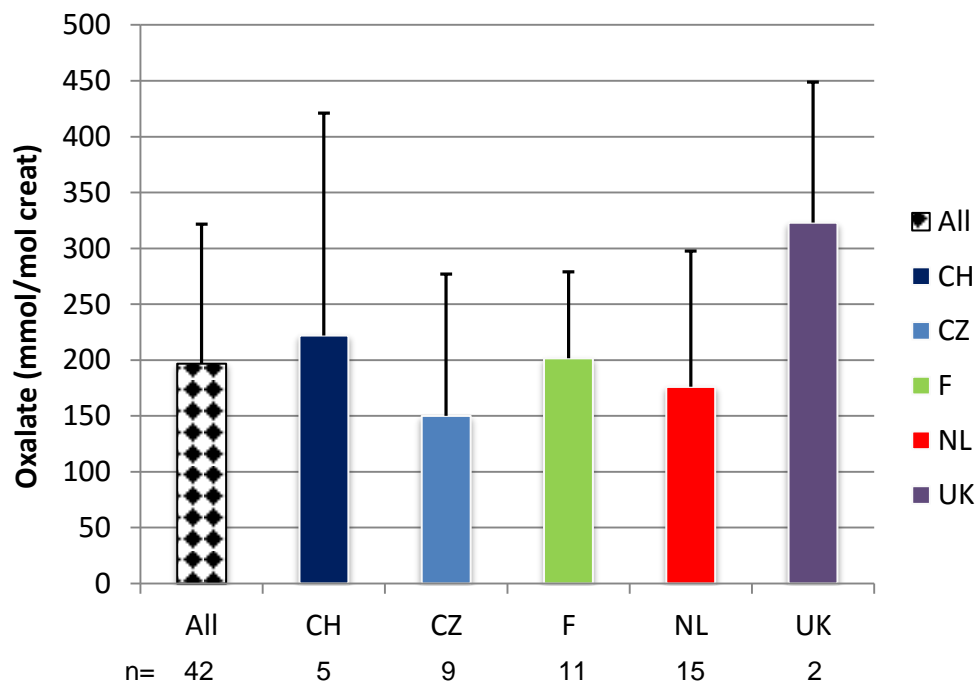
Oxalate results



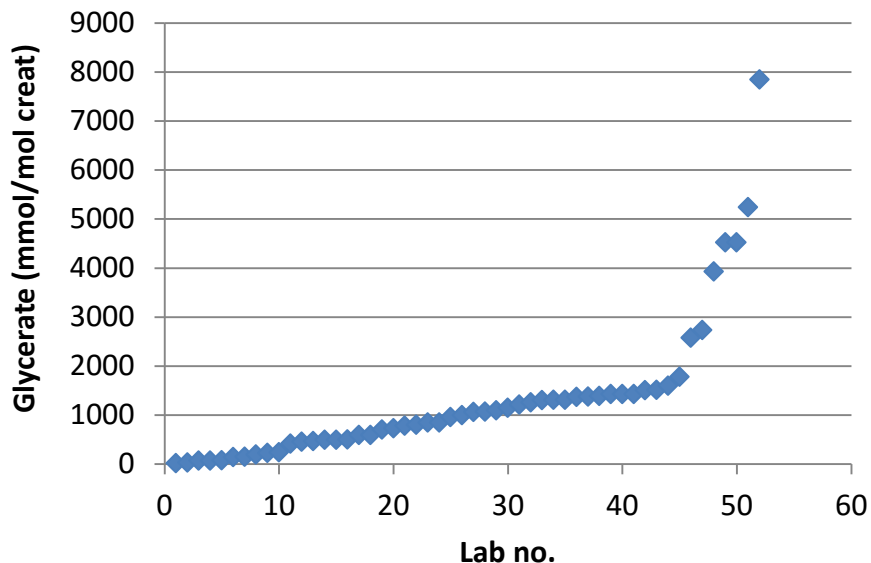
42 quantitative results reported for oxalate

Oxalate: 197 ± 125 mmol/mol
(CV 63 %)

➤ Extraction efficiency?



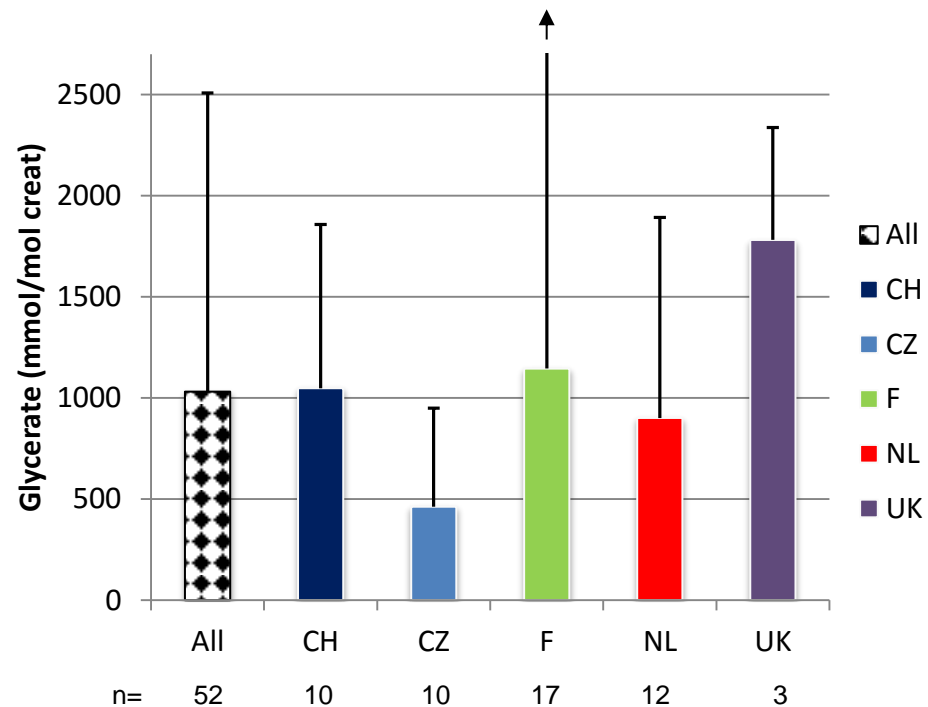
Glycerate results



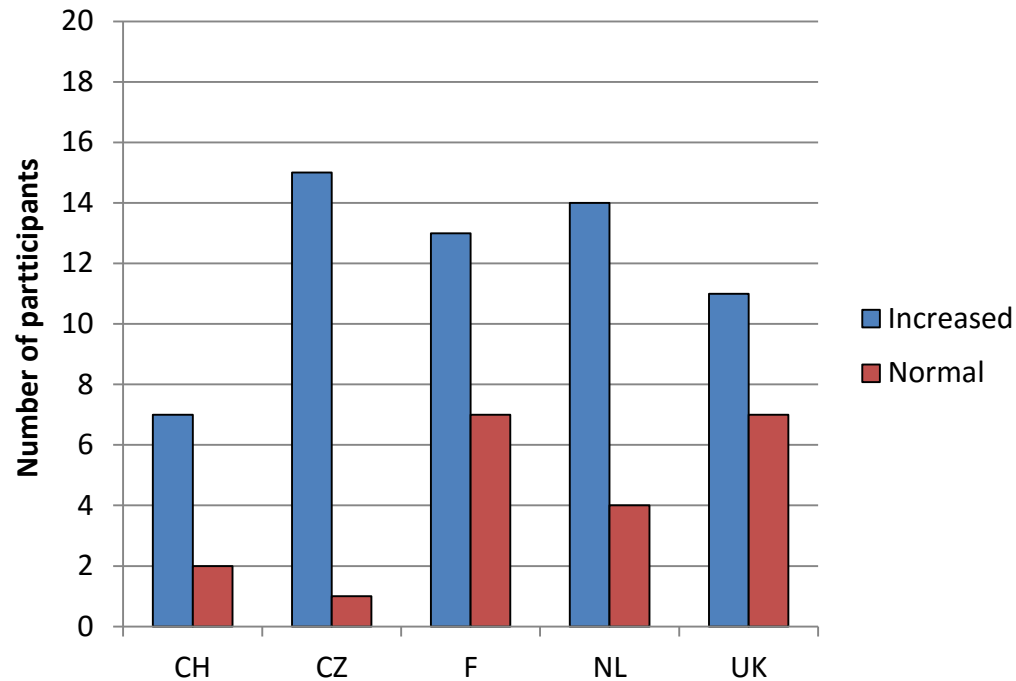
Glycerate: 1031 ± 1477 mmol/mol
(CV 143 %)

- Extraction efficiency, calibration?
- 102/106 participants reported glycerate elevated

52 quantitative results reported for glycerate



Oxalate: interpretation of results (1)

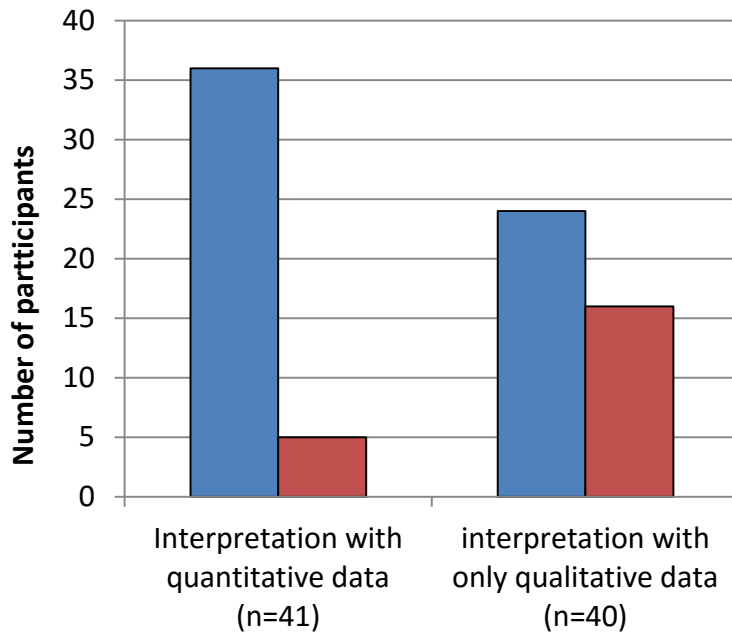


All: oxalate increased: 60
 oxalate normal: 21

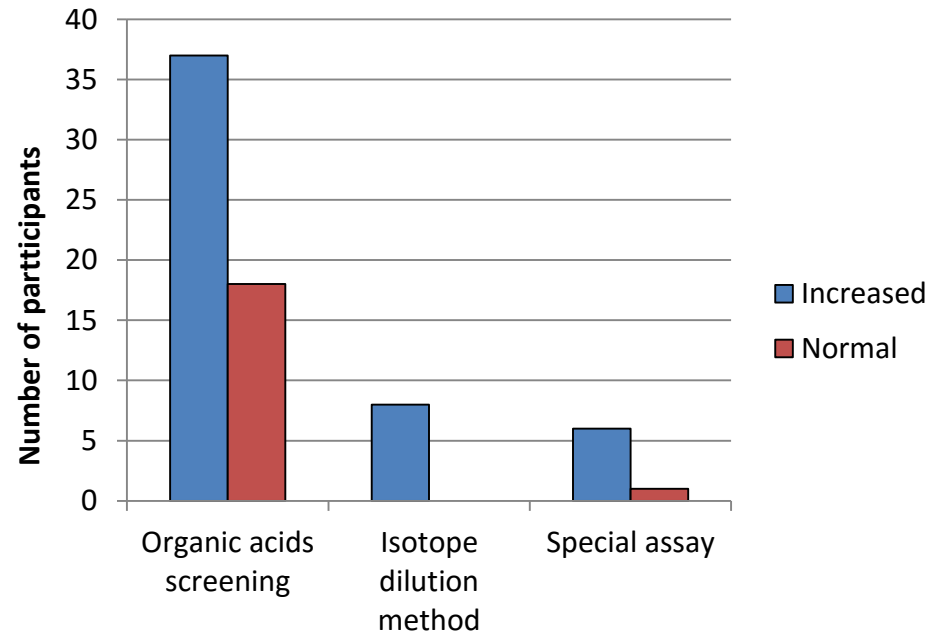
Oxalate interpretation of results (2)

Effect of quantitation & analytical method

Quantitation yes/no



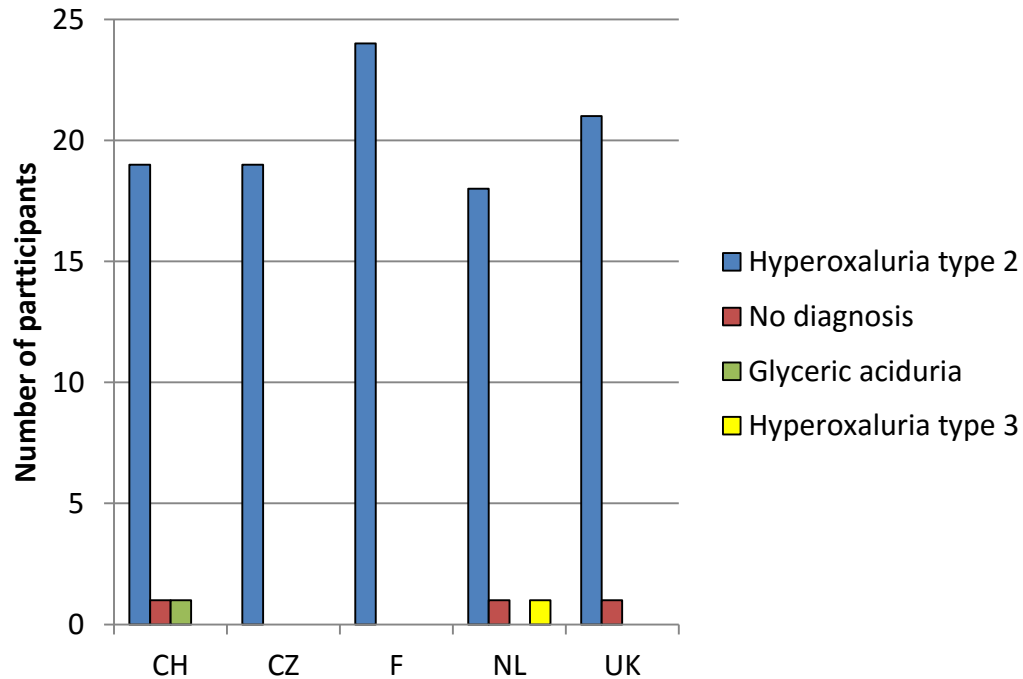
Analytical method



- Quantitative analysis of oxalate improves sensitivity (detection of hyperoxaluria)

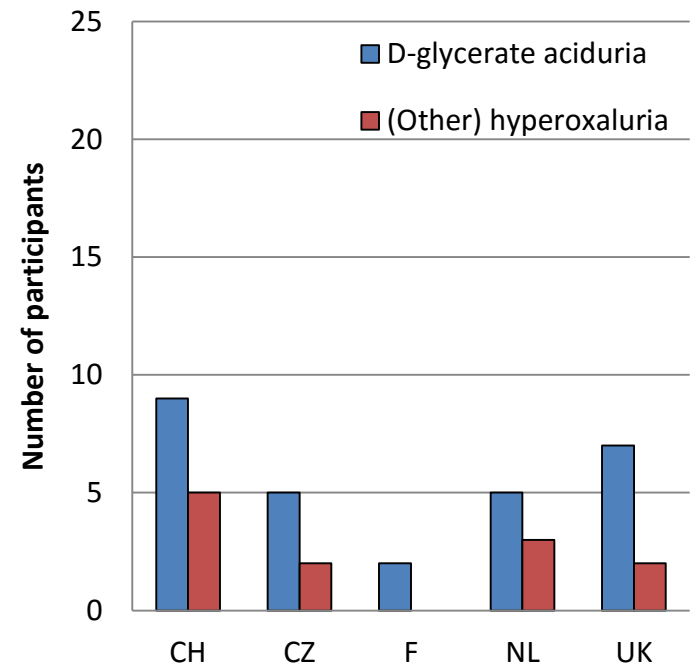
Interpretation of results

Most likely diagnosis

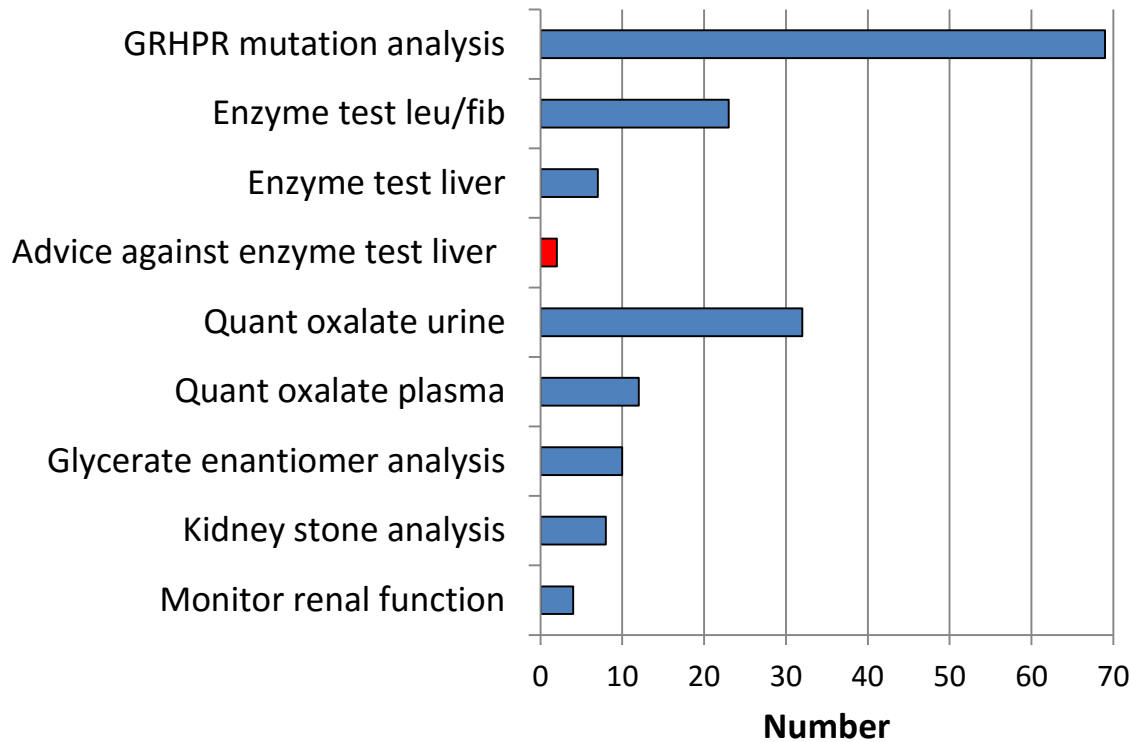


DPT participants: 108
 Reports: 106
 Correct diagnosis: 102 (96%)

Other possible diagnoses



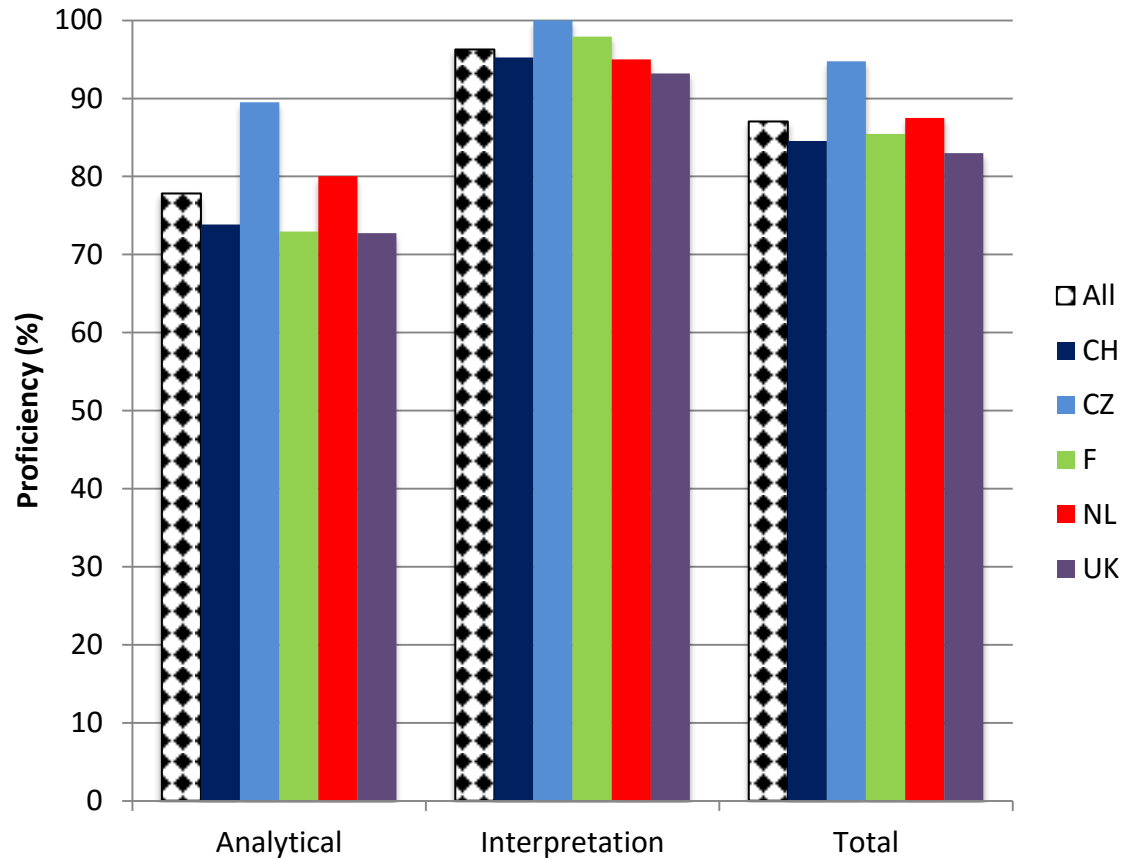
Advice for further investigations



Scoring criteria

Analytical	points
Oxalate elevated	1
Glycerate elevated	1
Interpretation of results	
Hyperoxaluria type 2	2
Hyperoxaluria unspecified or wrong type	1
Advice to measure oxalate (when oxalate analysis is not performed and no diagnosis reported)	1
Maximum score	4

Diagnostic proficiency



Overall proficiency: 87%

Conclusions

- Increased oxalate and glycerate → hyperoxaluria type 2
- 106 out of 108 participants reported results
- Analytical proficiency: satisfactory
 - Quantitative results oxalate, glycerate reported by <50% of participants
 - Lack of precision and accuracy in quantitation oxalate, glycerate (extraction efficiency, calibration, reference values?)
 - Quantitative oxalate test → better detection of hyperoxaluria
- Interpretation of results: excellent
 - Correct diagnosis by all participants that detected elevated glycerate

Recommendations:

1. In patients with kidney stones: perform quantitative oxalate test
2. Harmonisation oxalate, glycerate (OA) testing required