ERNDiM

Quality Assurance in Laboratory Testing for IEM

Common DPT sample

Petr Chrastina

ERNDIM Workshop, 29th August 2023

Jerusalem, Israel

Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM

Clinical picture provided with the sample:

This boy was referred at the age of 7 years with attention deficit disorder.

The sample was collected at the age of 25 years on the specific treatment.

Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM

Diagnosis

mild form of argininosuccinic aciduria

due to argininosuccinate lyase deficiency

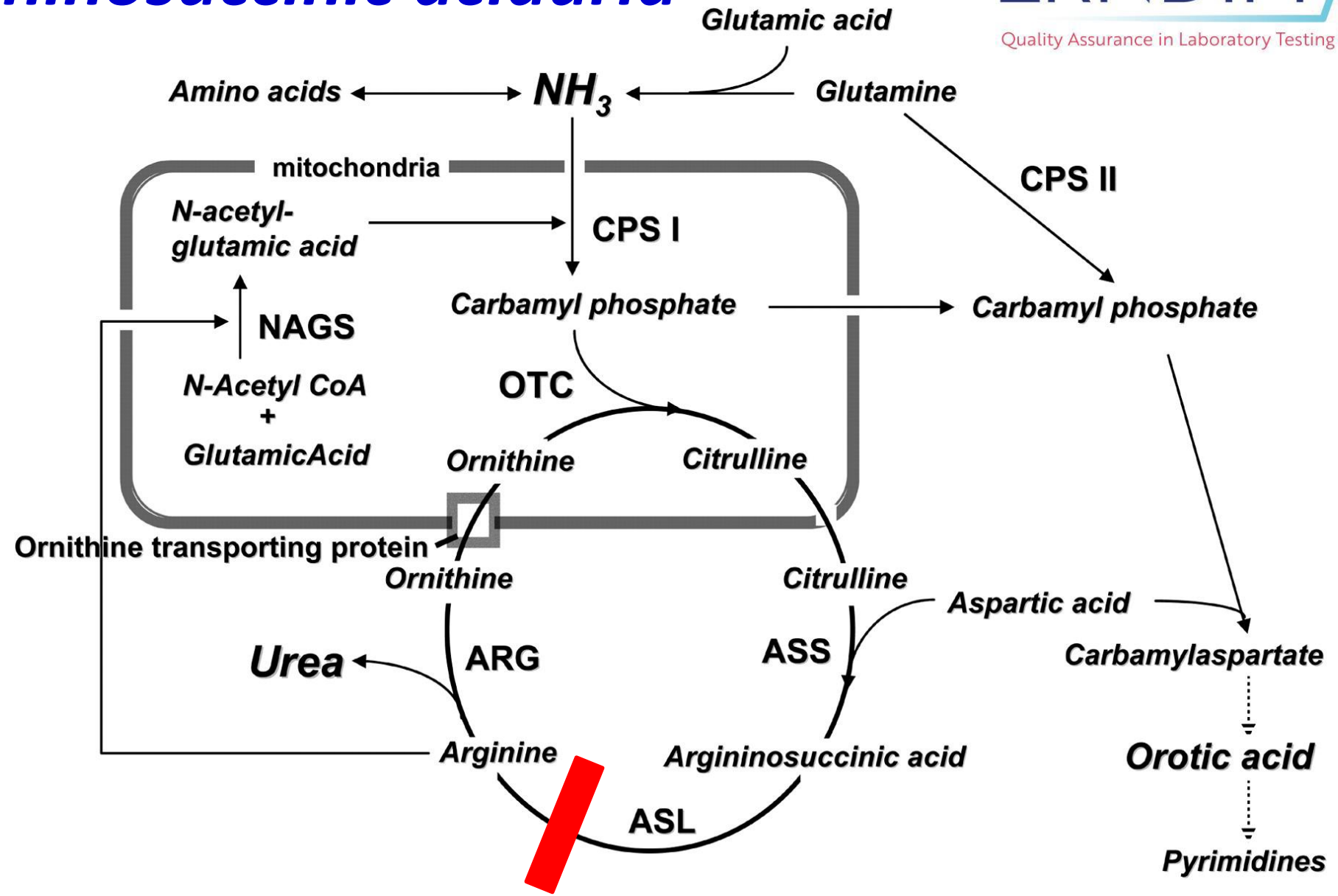
- The diagnosis was confirmed by molecular genetic analysis.

25 years old man

- treated with low protein diet (0,6 g/kg body weight)
- no metabolic decompensation
- obese suffers from hypertension and depression

argininosuccinic aciduria

ERNDiM
Quality Assurance in Laboratory Testing for IEM



DPT Centers

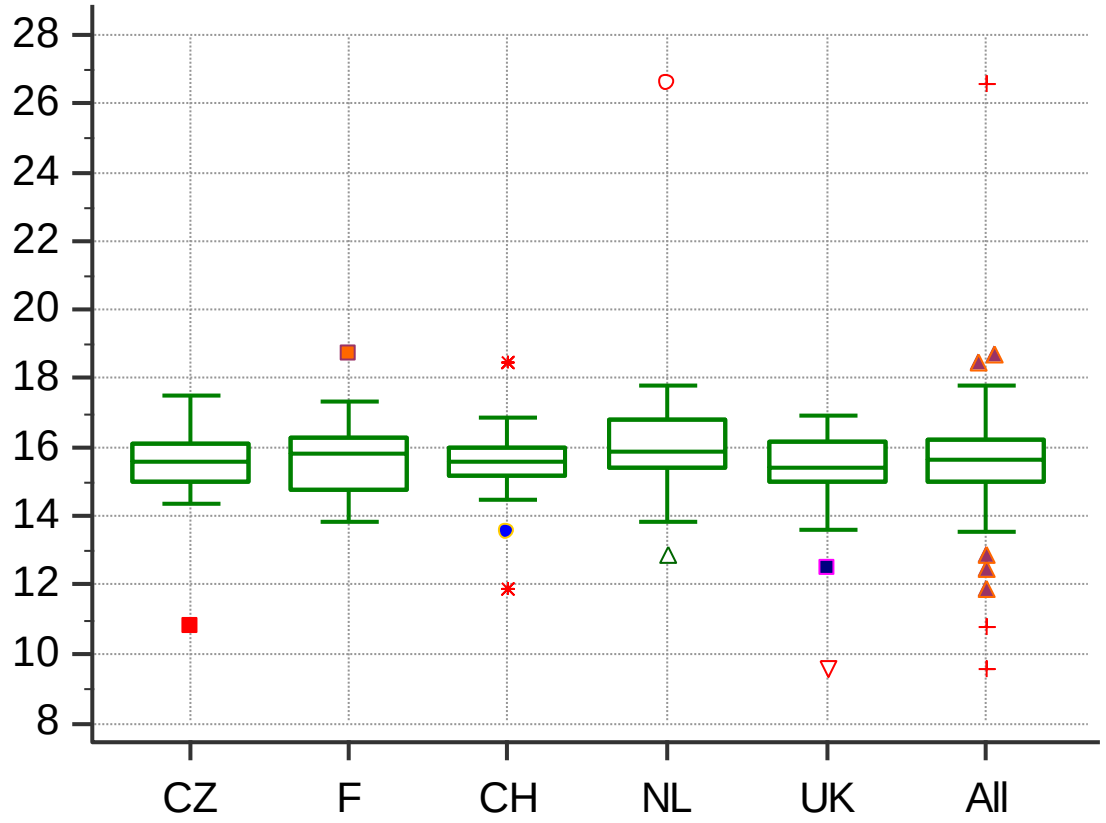
ERNDiM
Quality Assurance in Laboratory Testing for IEM

• Czech Republic	19 labs
• France	22 labs (2 no answer)
• Netherlands	18 labs
• Switzerland	21 labs
• United Kingdom	20 labs
• **Reports**	**98 labs**

Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM

Creatinine in urine

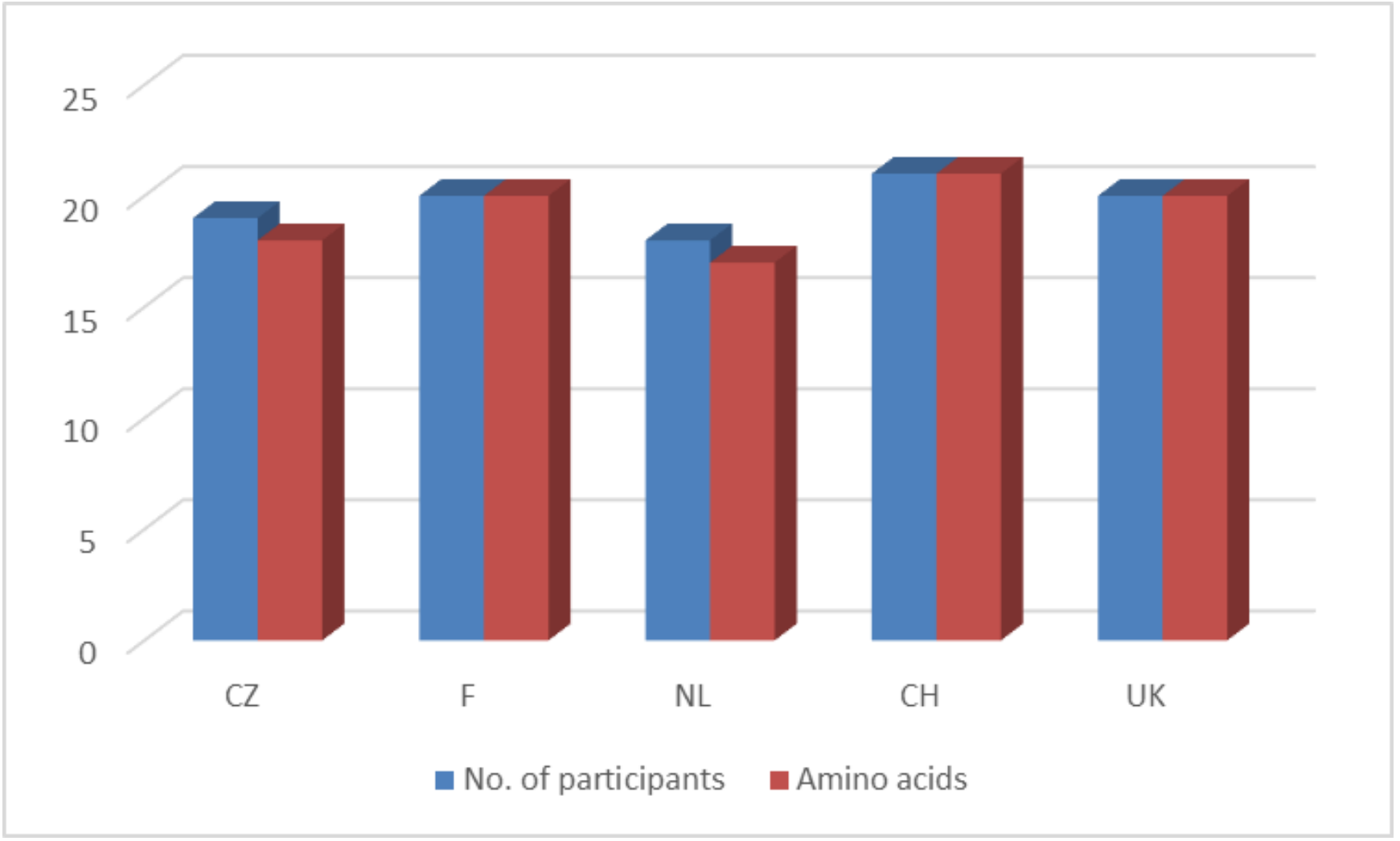


Average

15,6 mmol/l

Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM



Common DPT sample

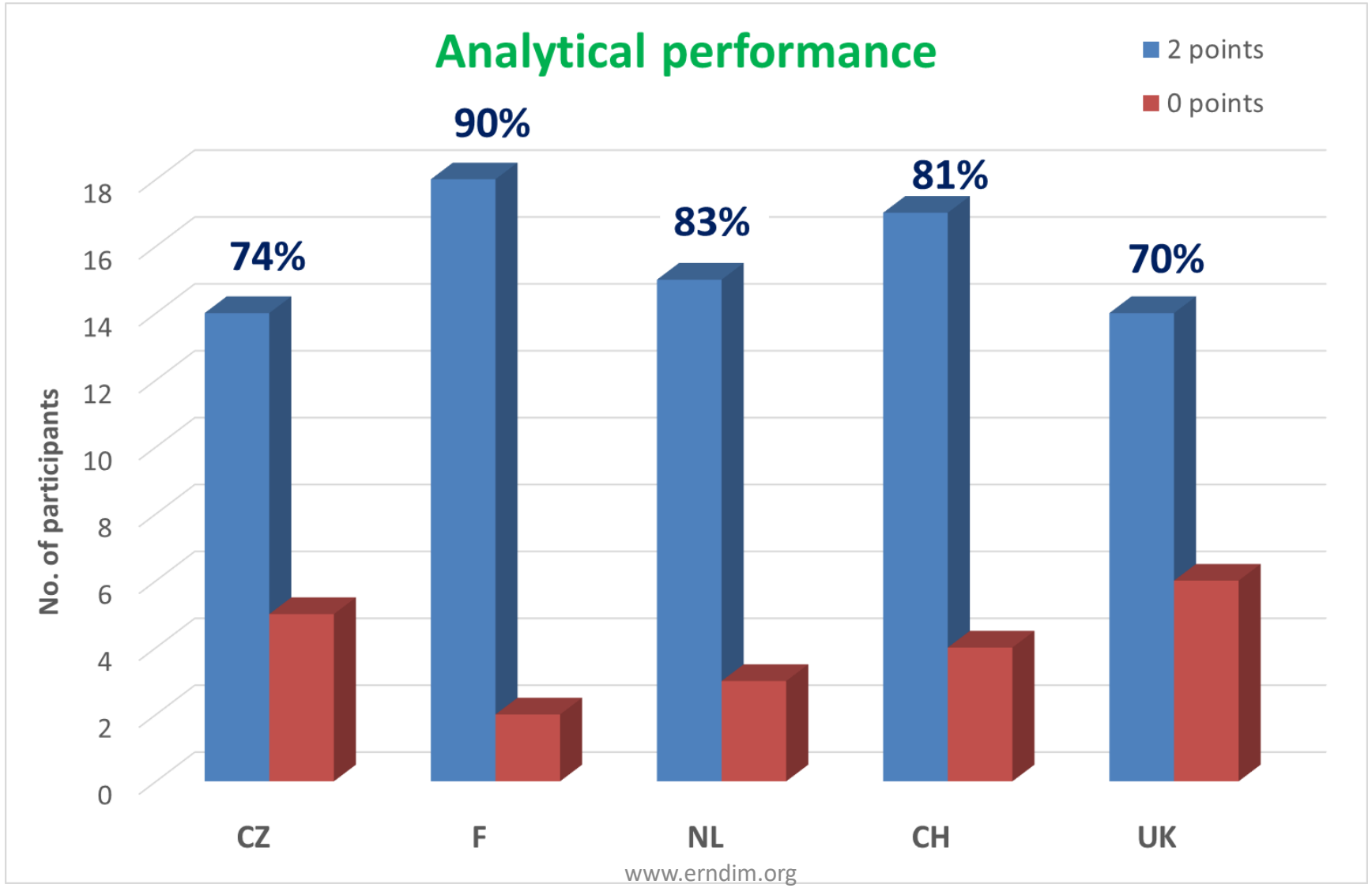
ERNDiM
Quality Assurance in Laboratory Testing for IEM

Scoring of analytical performance

- increased excretion of **argininosuccinic acid**
  and/or **its anhydrides** **2 points**

Common DPT sample

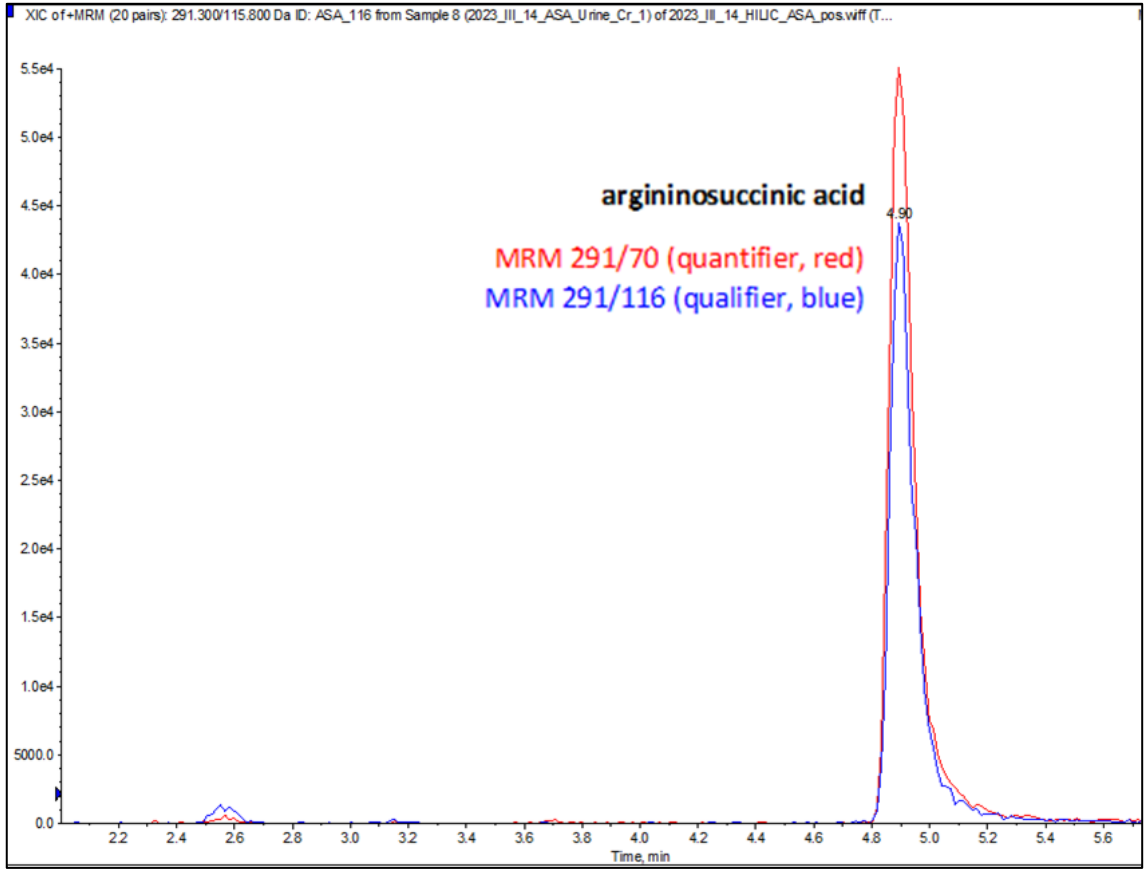
ERNDiM
Quality Assurance in Laboratory Testing for IEM



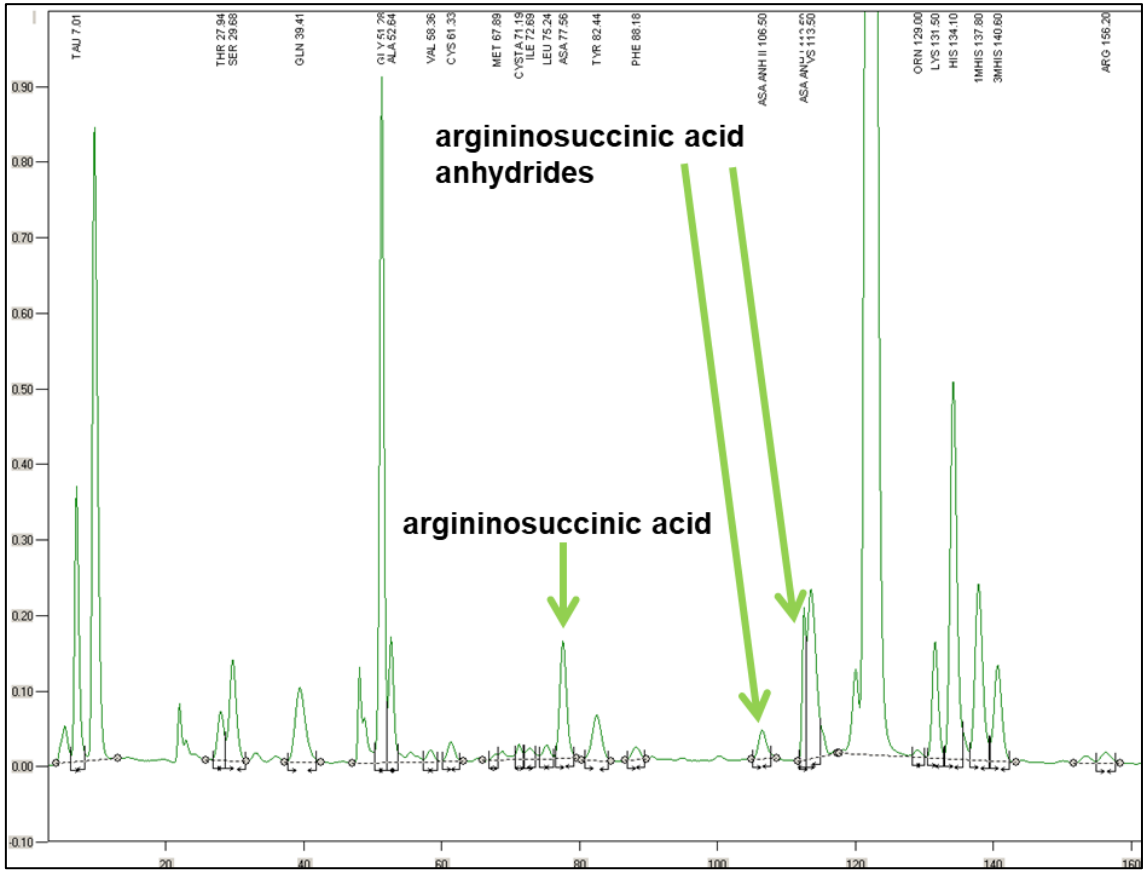
Common DPT sample

amino acids analysis

LC-MS/MS

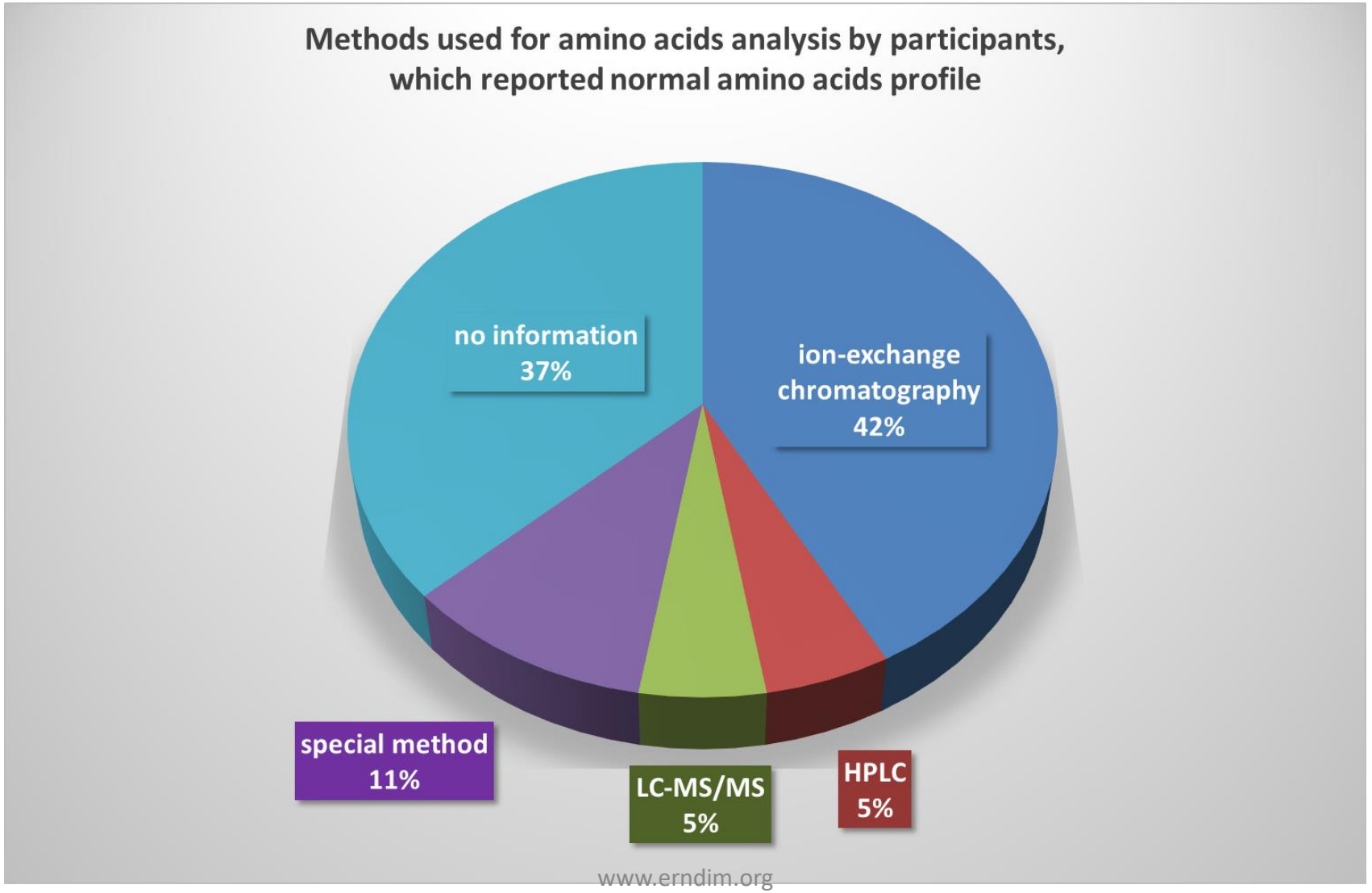


IEC-NHD



Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM



Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM

Interpretation and recommendation

argininosuccinic aciduria 2 points

n = 78 80%

+ mutation analysis of the *ASL* gene

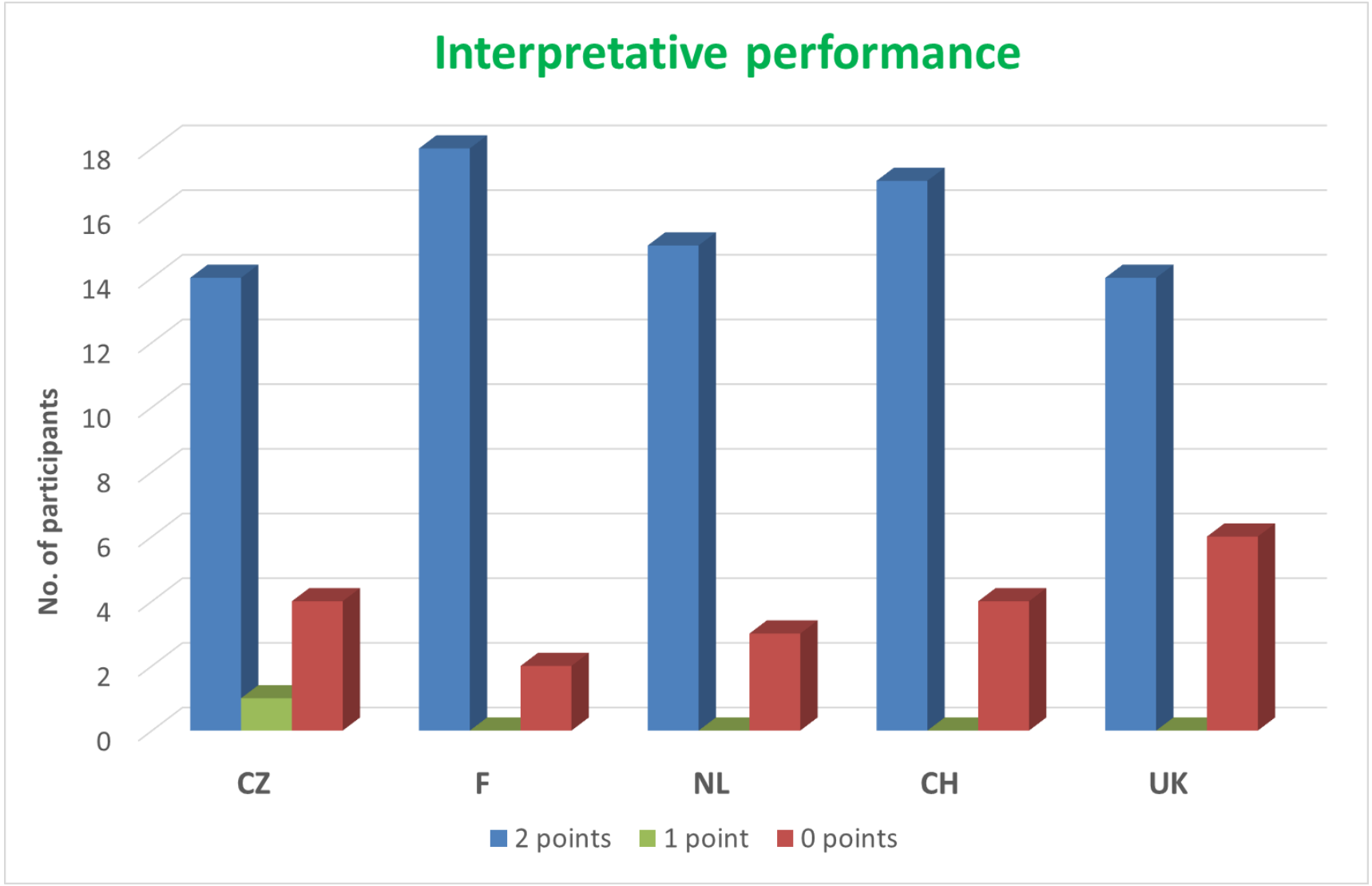
Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM

Interpretation and recommendation	0 points
thiamine metabolism dysfunction syndrome	2
mucopolysaccharidosis type III	2
tryptophan dioxygenase deficiency	1
MSUD	1
tyrosinemia type III	1
MTHFR deficiency	1
no metabolic disorder	12

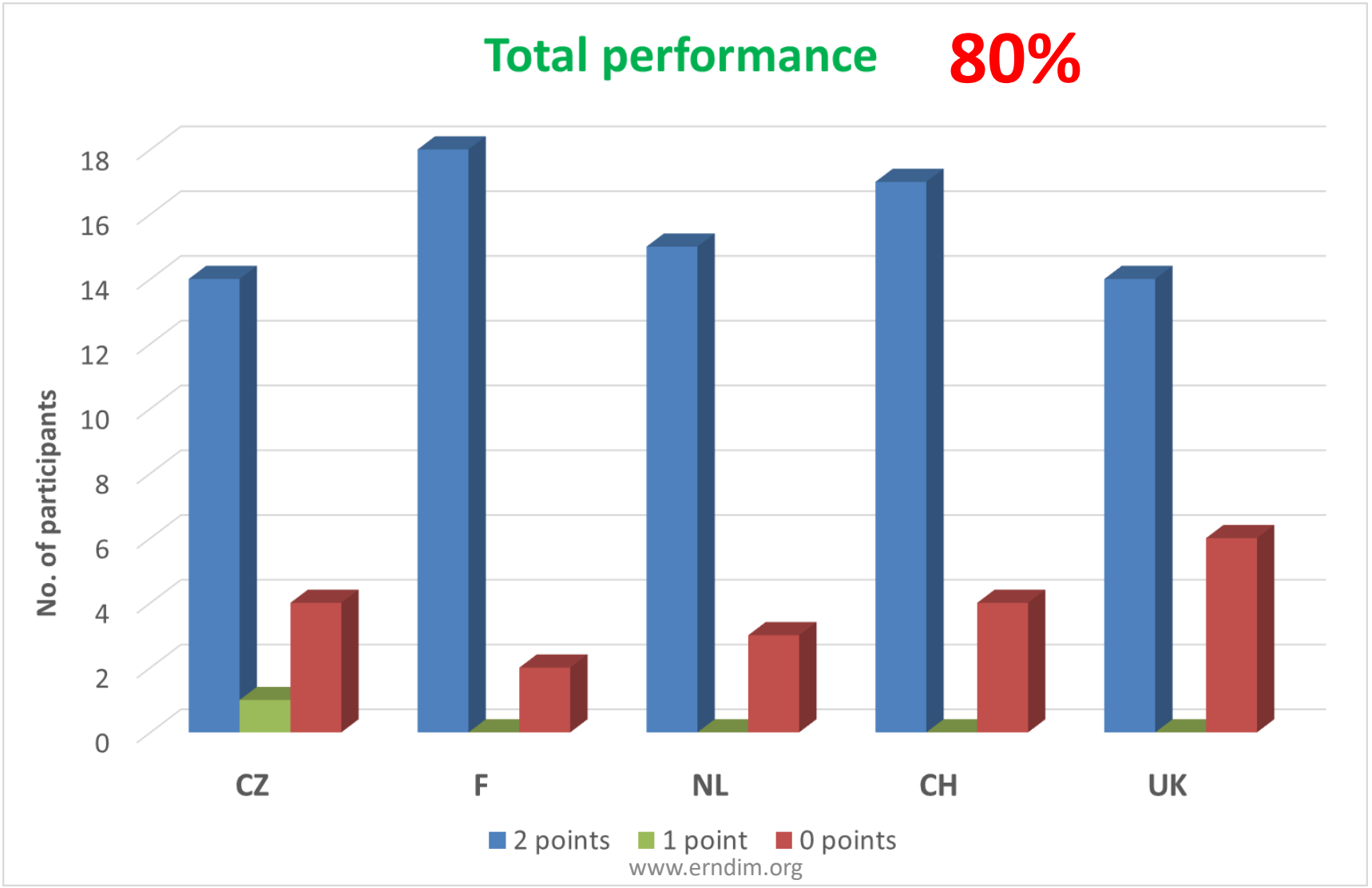
Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM



Common DPT sample

ERNDiM
Quality Assurance in Laboratory Testing for IEM



Summary

mild argininosuccinic aciduria

- **typical DPT sample**

  with suboptimal proficiency score (80%)

- **20/98 participants did not detect elevated excretion of argininosuccinic acid**


Thank you
for your attention!