ERNDIM Urine MPS: an External Quality Assurance scheme for diagnostic testing of mucopolysaccharidoses in urine

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Introduction

The Urine MPS scheme is a regular ERNDIM scheme as of 2012 (pilot study in 2010 – 2011)
Cost (2014): € 230
Aims:
1. to evaluate diagnostic proficiency in urine MPS screening
2. to assist IEM diagnostic laboratories in improving or maintaining their skills in MPS screening.

Scheme format

- Each year 6 authentic (lyophilized) urine samples are circulated (usually 5 MPS, 1 normal control)
- 107 participants in 2014
- Participants are asked to analyze 3 samples in April and 3 samples in June:
  ➢ Perform quantitative GAG analysis with interpretation, i.e. normal or increased
  ➢ Perform qualitative GAG analysis and give the most likely diagnosis
  ➢ Report results on CSCQ website
- Feedback is provided by:
  ➢ 2 interim reports and 1 annual report
  ➢ Letters of support to poor-performers

Methods used to analyse GAG
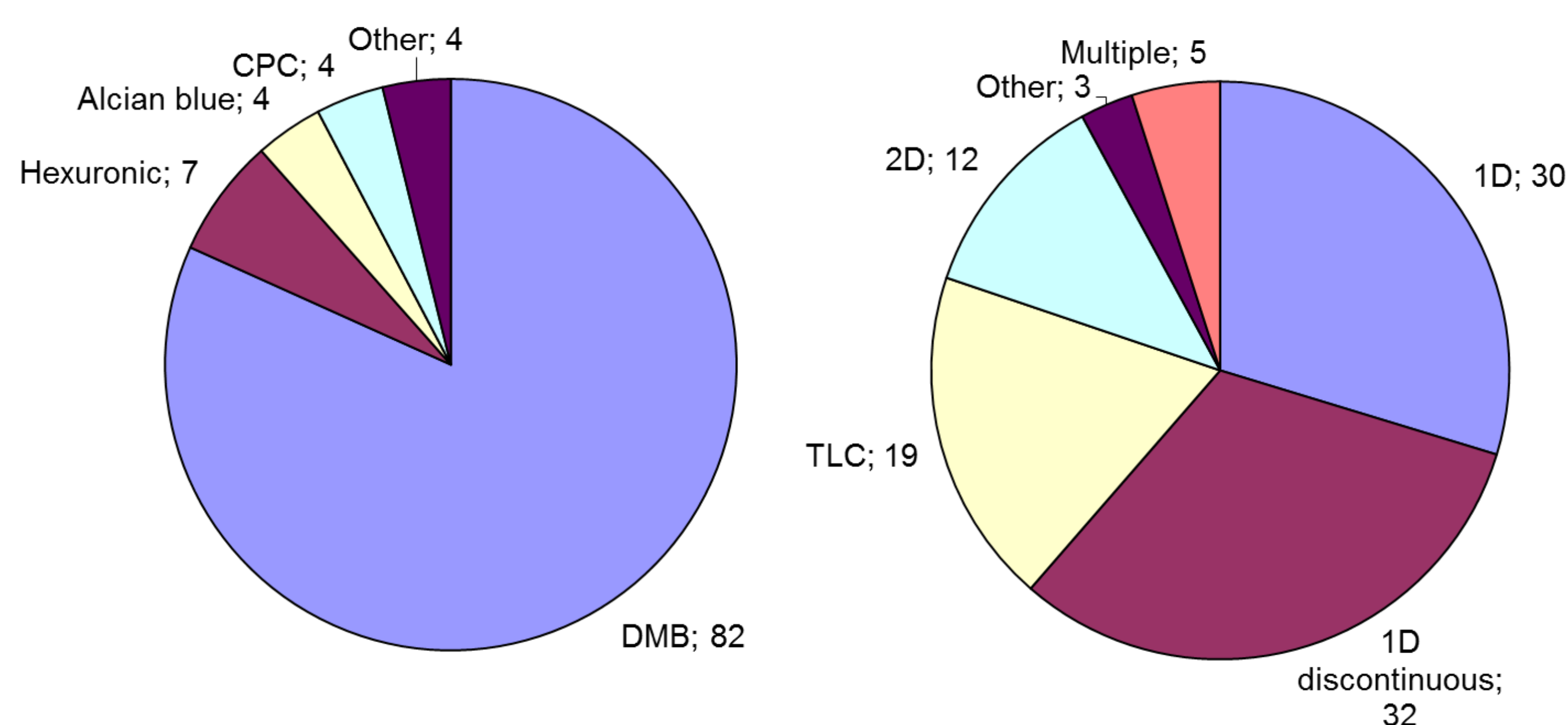
Methods used by the participants of the ERNDIM Urine MPS scheme (2010-2013). Left panel: quantitative analysis (methods codes are the reagents used; n=104), Right panel: qualitative analysis of GAG subfractions (methods codes are various electrophoresis/ chromatography systems n=101). Data: % of total.

Quality of quantitative MPS screening
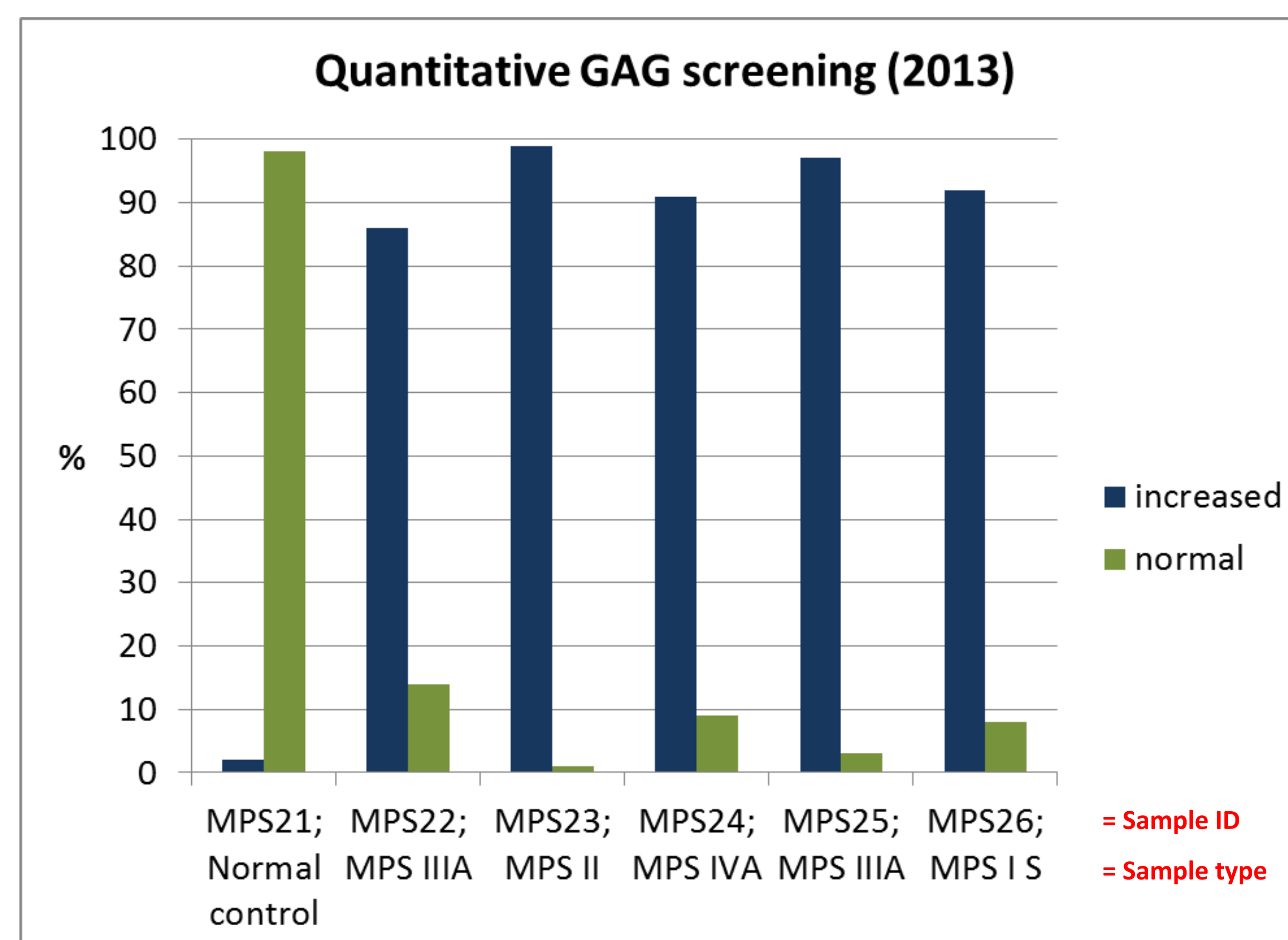
Interpretation (normal/increased) of quantitative GAG screening test results is usually >90% correct (figure shows 2013 scheme; n=95). Mild cases of MPS I and, particularly, MPS III and IV are frequently missed.

Proficiency of urine MPS screening
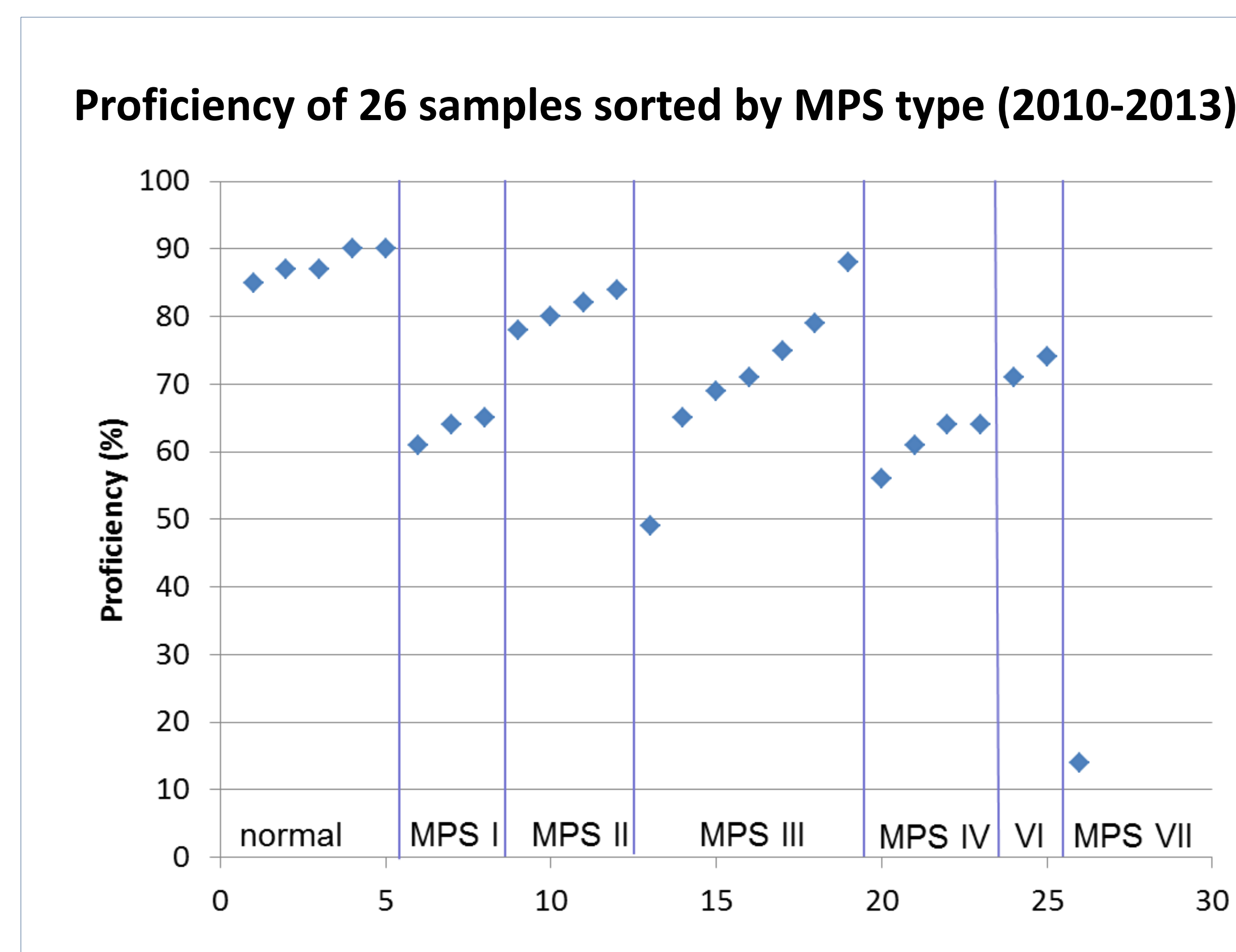
Diagnostic proficiency is 50-90% (one MPS VII sample excluded) with relatively low proficiency for MPS IV and mild MPS III. MPS I and VI apparently are difficult to distinguish using electrophoretic/chromatographic analysis.

Conclusion

The Urine MPS scheme shows that current GAG analysis methods are not sufficiently robust to diagnose all MPS patients. Novel methods, such as recently reported LC-MS/MS based GAG assays, will hopefully improve this.

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

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