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Beyond compliance: How external quality assessment drives laboratory improvement over time

Evidence from a longitudinal study of EQA data from genomic testing laboratories

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Background

External Quality Assessment (EQA) plays a critical role in safeguarding the accuracy, reliability, and clinical utility of genomic testing. While its immediate value in identifying errors and benchmarking performance is well recognized, the long-term impact of sustained EQA participation on laboratory quality and improvement is less well studied.

EQA contributes to quality assurance by:

- Monitoring laboratory performance and test accuracy
- Detecting and reducing critical analytical and interpretive errors
- Supporting continuous quality improvement through feedback and benchmarking

This study investigates the utilization of longitudinal EQA performance data, highlighting its role in not only evaluating compliance but also fostering significant and lasting improvement in genomic testing laboratories over time.

Methods

We selected ten EQA schemes—six from our germline genetic testing and four from our molecular pathology portfolios—based on the following inclusion criteria:

- A minimum of 10 consecutive years of EQA data
- More than 50 participating laboratories per EQA run
- Consistent sample number and sample type across all years
- Unchanged scope and structure of the EQA scheme over time

We evaluated three core performance metrics:

1. Genotyping accuracy
2. Interpretation quality
3. Rate of poor performance (PP)

Genotyping and interpretation were assessed using a deductive scoring system. Each participant began with full marks, from which points are deducted (0.20–2.0) for each error depending on the severity and potential clinical impact of the mistake.

Poor performance (PP) was defined as any critical genotyping or interpretive error with the potential to cause patient harm.

Incomplete submissions were excluded from the analysis. All data were anonymised and results aggregated to identify broader trends. We used Microsoft Excel for data handling and linear regression analysis to explore the relationship between frequency of participation (1–10 years) and:

- Average genotyping and interpretation scores
- Frequency of poor performance

Results

- Genotyping scores show a modest upward trend over time as EQA schemes run consecutively (Figure 1).
- Interpretation scores demonstrate a more pronounced and consistent improvement across EQA cycles (Figure 2).
- When analysed by laboratory participation frequency, both genotyping and interpretation scores improve with regular involvement in EQA, for both germline and molecular pathology schemes (Figures 3 and 4).
- A clear decline in the rate of poor performance (PP) is observed with increased participation: laboratories with more EQA cycles exhibit fewer critical errors (Figure 5).

Figure 1. Average genotyping scores for all EQAs in 2015-2024

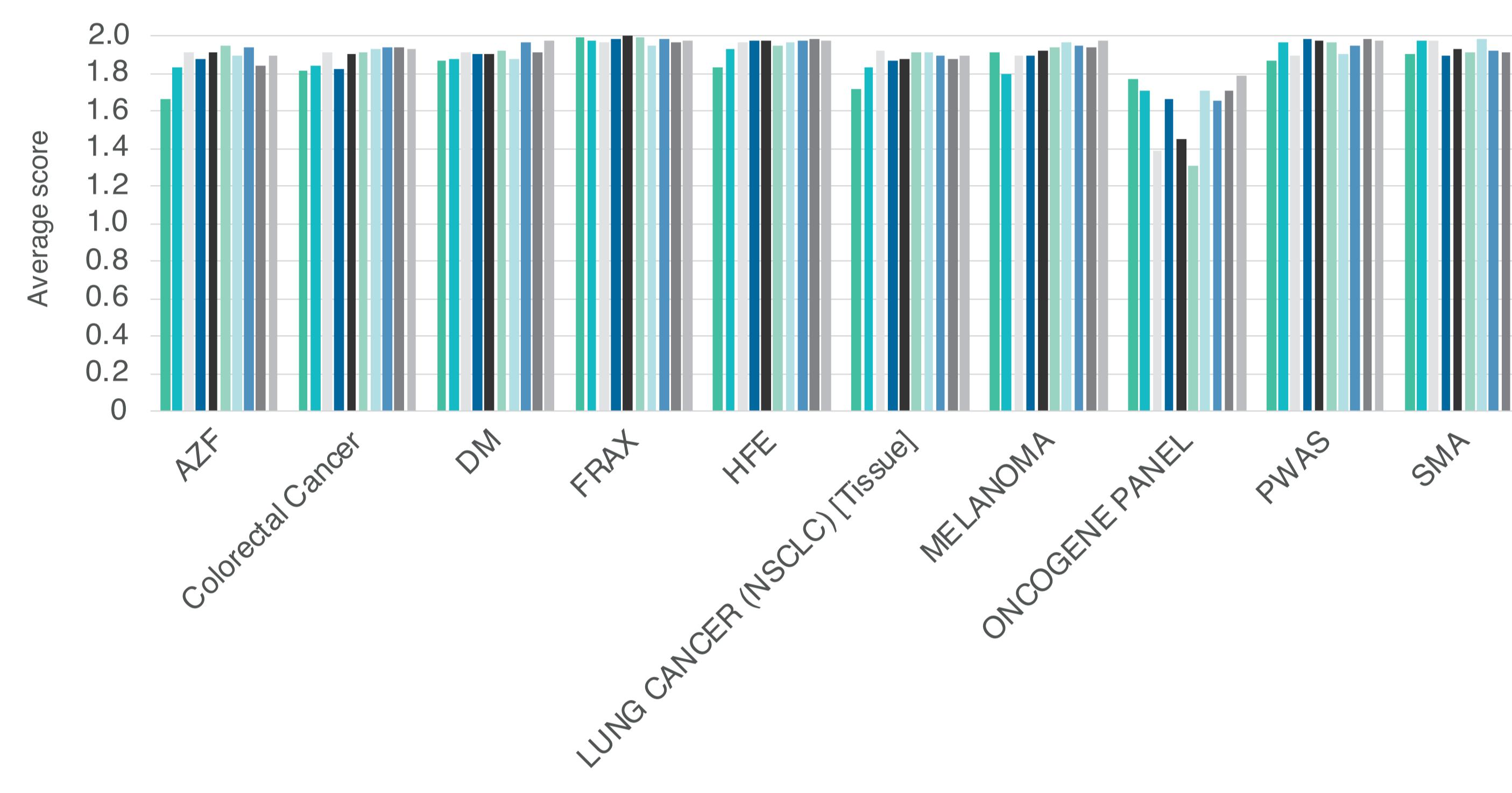


Figure 2. Average interpretation scores for all EQAs in 2015-2024 (oncogene panel does not assess interpretation)

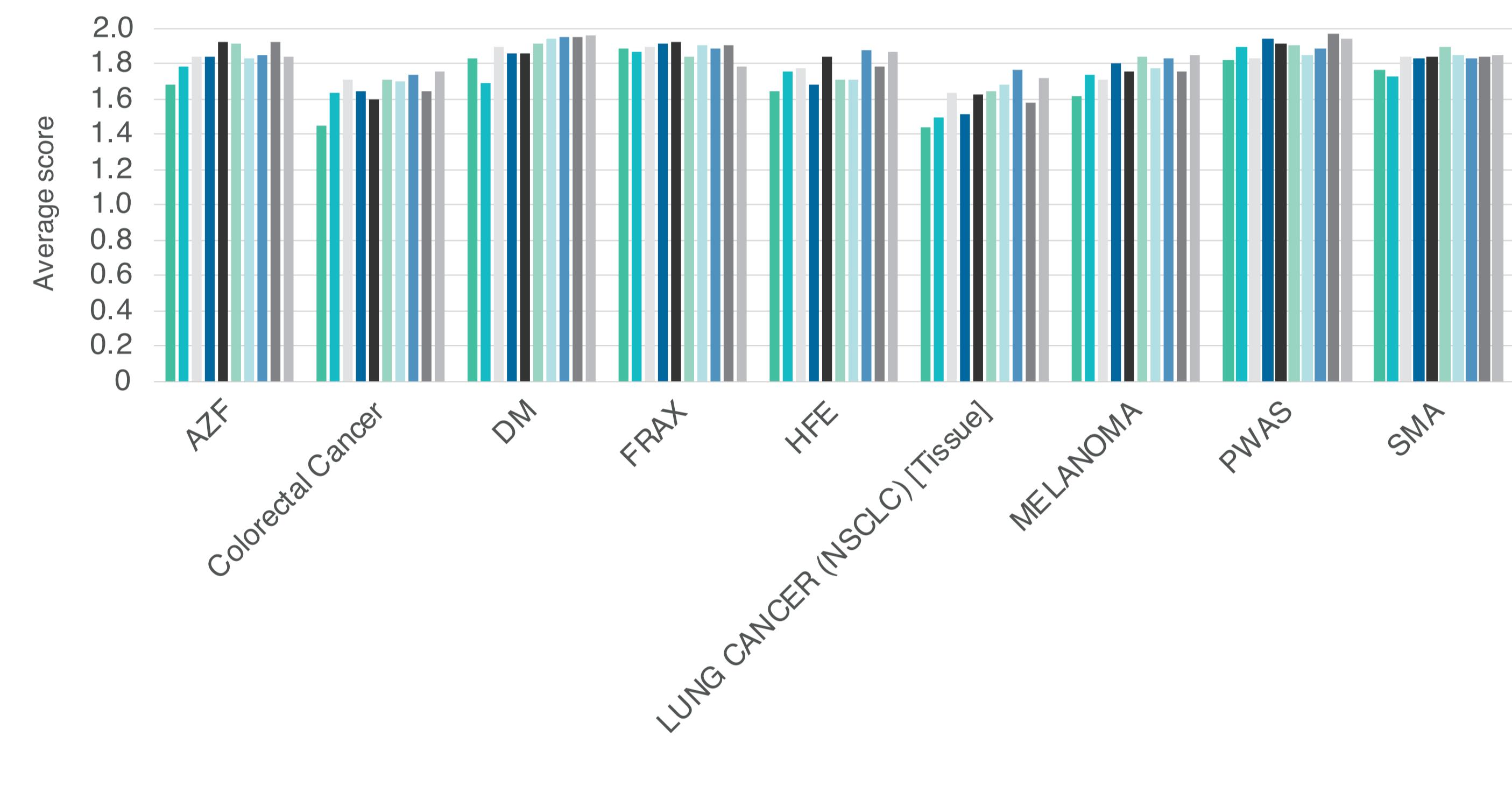


Figure 3. Average score per number of participations (1-10) for the combined germline EQAs (AZF, DM, FRAX, HFE, PWAS, SMA)

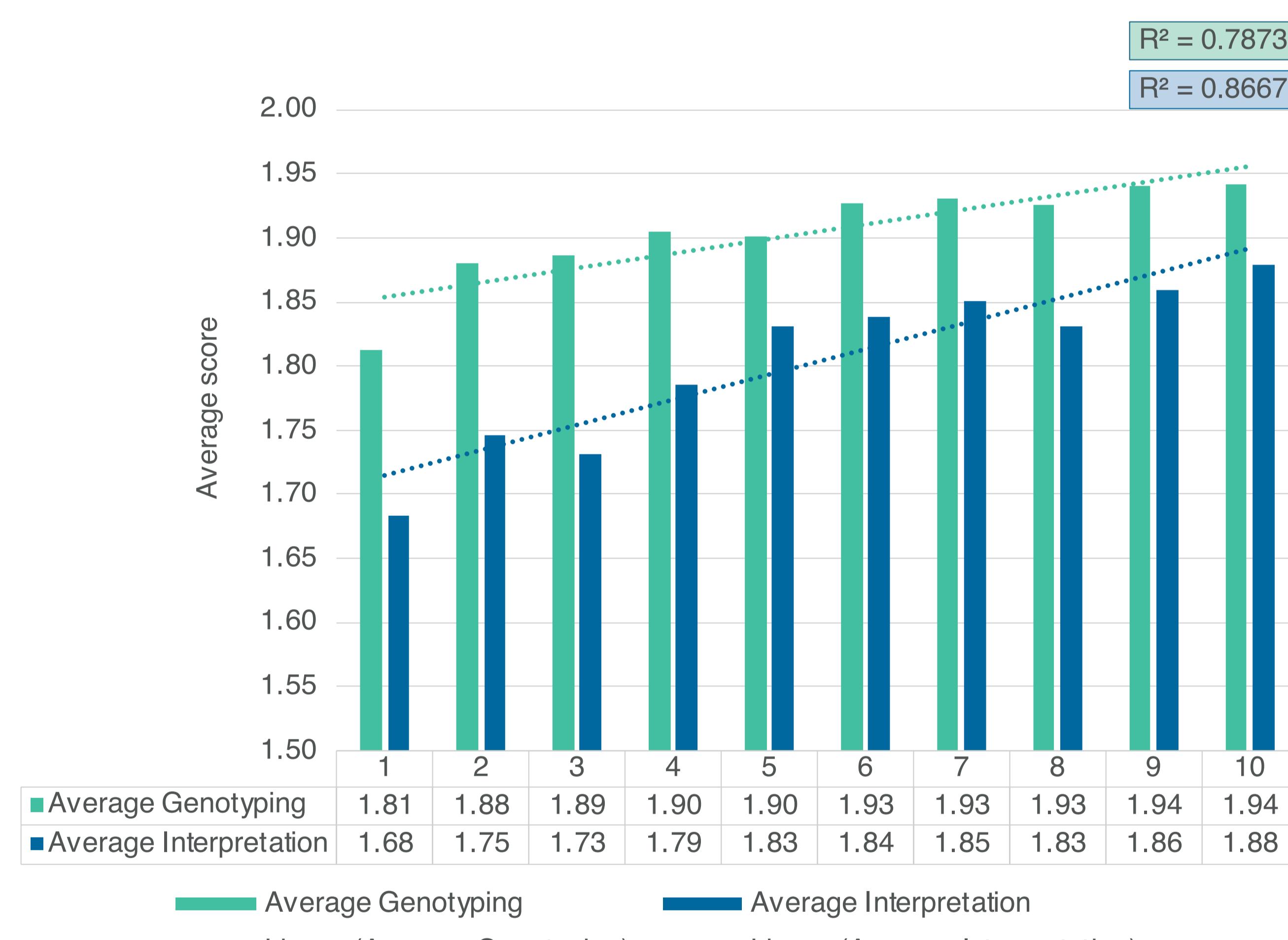


Figure 4. Average scores per number of participations (1-10) for the combined molecular pathology EQAs (Colorectal, Melanoma, Lung cancer)

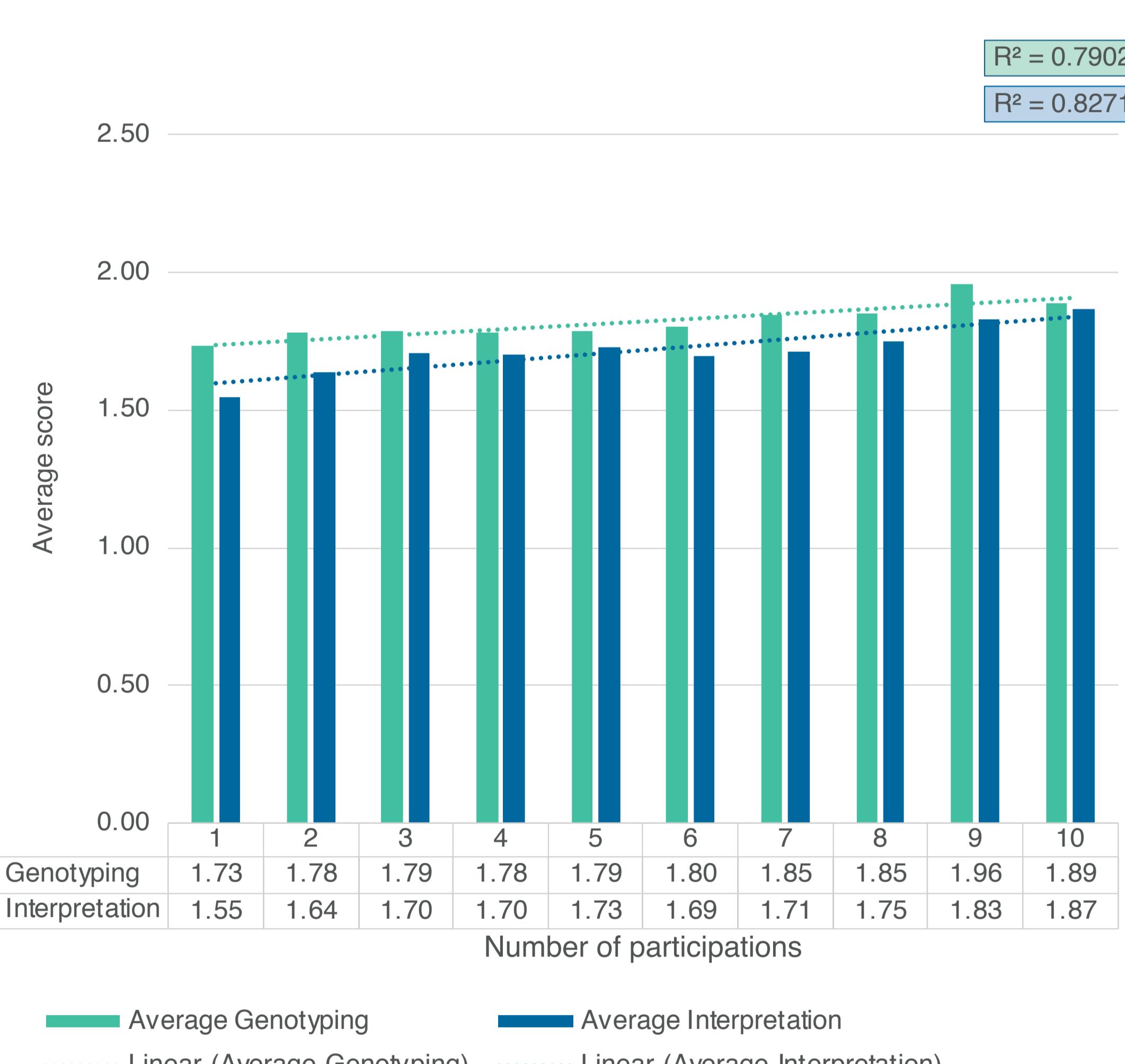
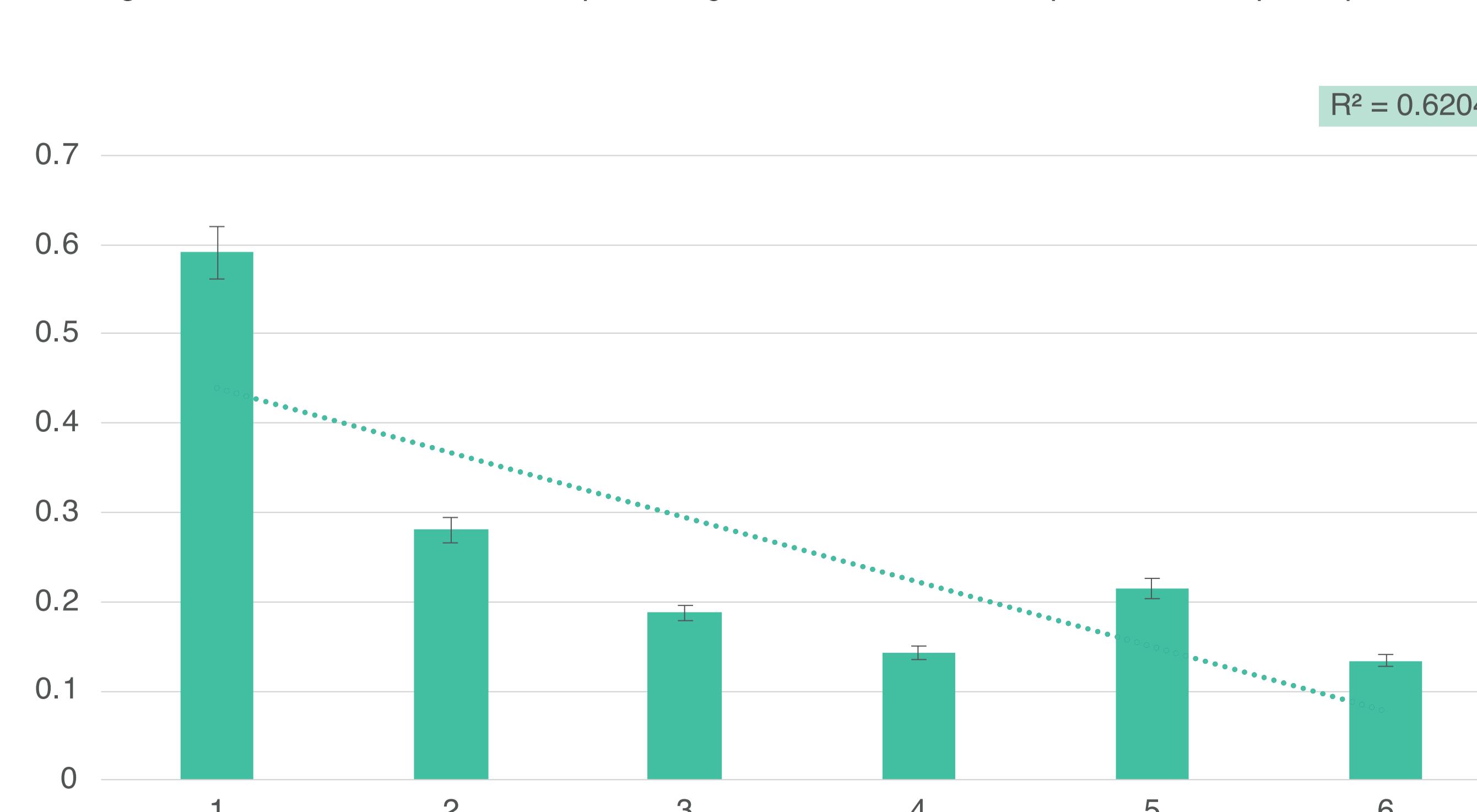


Figure 5. Combined results for the percentage of Poor Performance per number of participations.



Discussion

Our analyses of longitudinal EQA data provide clear evidence that sustained participation in EQA schemes contributes to measurable quality improvement in genomic testing laboratories. While both genotyping and interpretation performance improved over time, the gains were most evident in interpretation accuracy, a domain often influenced by evolving clinical knowledge and reporting standards.

The decreasing rate of poor performance with increased participation further underscores EQA's important role beyond compliance—it functions as a feedback and learning mechanism that enables laboratories to detect errors, maintain quality as processes change, benchmark progress, and implement corrective actions.

Interestingly, a small subset of laboratories exhibited persistent variability in performance across multiple cycles. This highlights the need for tailored interventions, such as targeted feedback or training, to address persistent gaps and ensure all participants benefit from the quality improvement potential of EQA.

These findings reinforce the value of EQA not only as a regulatory or accreditation tool but as a driver of long-term, system-wide improvement in genomic testing services.

Conclusions

Long-term participation in EQA programs contributes to more than regulatory compliance—it drives measurable, lasting improvements in laboratory quality. Sustained involvement is linked to fewer critical errors and greater accuracy in both genotyping and interpretation. However, while these errors decrease over time, they are not entirely eliminated. They can arise from process or personnel changes, equipment updates, or the inclusion of unfamiliar or complex variants. As such, ongoing engagement with EQA remains essential. Ultimately, EQA is a cornerstone for promoting excellence, consistency, and confidence in genetic testing.