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1<sup>st</sup> November 2022

Dear Porphyrin Laboratory users,

We are writing to inform you that we will **be withdrawing the porphyrin hydroxymethylbilane synthase (HMBS) enzyme activity assay** from **1<sup>st</sup> December 2022**.

We were prompted to review the clinical utility of the service following the recent discontinuation of the commercial uroporphyrin fluorescence standard used to calibrate the assay. Although HMBS activity is markedly low in Acute Intermittent Porphyria (AIP) patients, there is an overlap with values in normal patients and this limits clinical utility of the assay. As an alternative, genetic testing of the HMBS gene is available and has superseded the need for an HMBS assay for family screening. For suspected AIP patients experiencing current acute symptoms, a urine PBG remains the most appropriate test. Withdrawing the HMBS test will align our test repertoire to the other SAS porphyria laboratory.

We would also like to take this opportunity to remind you that all porphyrin samples should be **protected from light** and ideally be collected during a suspected porphyria attack. Please do not use surgical or masking tape to protect samples from light and ensure the sample containers themselves are labelled, rather than the protective wrapping.

If you would like to discuss this further or have any other porphyrin-related laboratory enquiries, please contact me using the details below.

Yours sincerely

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