

## **EMQN Office**

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## INDIVIDUAL LABORATORY REPORT (ILR) - Lab 1678

SCHEME: HBOC (Panel Testing) SEASON: 2023

Case 1				
Assessment Category	Score <sup>1</sup>	Comments (& deductions²)		
Genotyping	2.00	Version number missing / incorrect / inconsistent for reference sequence (0.00)		
Interpretation	2.00	All essential interpretative elements provided (0.00)		
Patient Identifiers and Clerical Accuracy	1.30	Failure to provide patient identifiers on each page of the report (0.20)		
		Errors in sample batch number or no sample batch number (0.50)		
		Reason for referral not restated (0.00)		
		Incorrect or no pagination (eg X of Y) (0.00)		

Case 2				
Assessment Category	Score <sup>1</sup>	Comments (& deductions <sup>2</sup> )		
Genotyping	2.00	VUS in PALB2 not mentioned (NM_024675.4(PALB2):c.3320T>C p.(Leu1107Pro)) (0.00)		
		Synonymous protein change reported (0.00)		
		Version number missing / incorrect / inconsistent for reference sequence (0.00) HGVS advice is to use p.? instead of the synonymous protein nomenclature for a splicing variant as this is not the effect seen in the protein. https://varnomen.hgvs.org/recommendations/RNA/variant/splicing/		
Interpretation	1.75	No/insufficient evidence for classification of variant (0.25) Further information about the expected effect of the ATM variant would ideally be required.		
Patient Identifiers and Clerical Accuracy	1.30	Failure to provide patient identifiers on each page of the report (0.20)		
		Errors in sample batch number or no sample batch number (0.50)		
		Reason for referral not restated (0.00)		
		Incorrect or no pagination (eg X of Y) (0.00)		

Case 3				
Assessment Category	Score <sup>1</sup>	Comments (& deductions²)		
Genotyping	2.00	Correct result reported (0.00)		
		Version number missing / incorrect / inconsistent for reference sequence (0.00)		
Interpretation	2.00	All essential interpretative elements provided (0.00)		
Patient Identifiers and Clerical Accuracy	1.30	Failure to provide patient identifiers on each page of the report (0.20)		
		Errors in sample batch number or no sample batch number (0.50)		
		Reason for referral not restated (0.00)		
		Incorrect or no pagination (eg X of Y) (0.00)		

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<sup>&</sup>lt;sup>1</sup> Maximum score is 2.00

<sup>&</sup>lt;sup>2</sup> Deductions from the maximum score

<sup>&</sup>lt;sup>3</sup> Green >= Scheme mean, Orange < Scheme mean, Red Poor performance. NRS no results submitted. WDS withdrew from scheme

## SUMMARY OF YOUR PERFORMANCE IN THIS SCHEME

Assessment Category	Performance <sup>3</sup> (mean score)
Genotyping	2.00
Interpretation	1.92
Patient Identifiers and Clerical Accuracy	1.30
Scheme result (SATISFACTORY or POOR)	SATISFACTORY

Report approved and authorised by Simon Patton (13 September 2023) on behalf of EMQN. Signed:

Mai bettan

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<sup>&</sup>lt;sup>3</sup> Green >= Scheme mean, Orange < Scheme mean, Red Poor performance. NRS no results submitted. WDS withdrew from scheme