INDIVIDUAL LABORATORY REPORT (ILR)*

2023 Primary Immunodeficiency disorders (PID)

Version: Final

Participant G11487



Member of UK NEQAS consortium

Case 1

Category	Score ¹	Comments	
Genotyping	2.00	Correct result.	1
Interpretation	1.50	Not included: Hemizygous for BTK c.1516T>C p.(Cys506Arg) likely pathogenic variant. (-0.5 marks)	\
		Variant evidence is insufficient for class 5 (-0.5 marks).	
Clerical Accuracy	2.00	No clerical errors.	

Case 2

Category	Score ¹	Comments
Genotyping	2.00	Correct result.
Interpretation	2.00	All essential interpretative elements provided.
Clerical Accuracy	2.00	No clerical errors.

Case 3

Category	Score ¹	Comments
Genotyping	2.00	Correct result.
Interpretation	2.00	Not included: Testing of Thomas's mother is recommended to determine her carrier status and to assess the risk of recurrence.
Clerical Accuracy	2.00	No clerical errors.

Mean Overall Score				
Genotyping	2.00			
Interpretation	1.83			
Clerical Accuracy	2.00			

Conclusions		
Recommendations	None	
Performance Statement ²	Satisfactory	

Thank you for your participation

- * To be read in conjunction with the EQA Summary Report
- 1 The maximum score for each category is 2.00
- 2 There are only two categories of performance for any EQA; "Satisfactory" and "Poor"

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