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## INDIVIDUAL LABORATORY REPORT (ILR) - Lab 0254

**SCHEME:** DNA SEQUENCING - SANGER

**SEASON:** 2022

Case 1		
Assessment Category	Score <sup>1</sup>	Comments (& deductions <sup>2</sup> )
Genotyping	2.00	No comments (0)
cDNA Interpretation	2.00	No comments (0)
Protein Interpretation	2.00	No comments (0)

Case 2		
Assessment Category	Score <sup>1</sup>	Comments (& deductions <sup>2</sup> )
Genotyping	2.00	No comments (0)
cDNA Interpretation	2.00	Comment only (0)
Protein Interpretation	2.00	Comment only (0)  Correct allelic nomenclature is: CFTR: LRG_663t1;p.[(Phe342HisfTer28)]; [Thr291_Gln372=]. Phe342= falsely indicates that only the amino acid at position 342 was analysed

Case 3		
Assessment Category	Score <sup>1</sup>	Comments (& deductions <sup>2</sup> )
Genotyping	2.00	No comments (0)
cDNA Interpretation	2.00	No comments (0)
Protein Interpretation	2.00	No comments (0)

<sup>1</sup> Maximum score is 2.00

<sup>2</sup> Deductions from the maximum score

<sup>3</sup> Green >= Scheme mean, Orange < Scheme mean, Red Poor performance.

NRS no results submitted. WDS withdrew from scheme


<b>General Comments</b>	Thank you for participating in EMQN Sanger Sequencing scheme. We look forward to welcoming you back in 2023.
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<b>Appeal response</b>	
<b>Decision</b>	Upheld
<b>Comments</b>	Appeal decision: upheld. You are correct it is also acceptable to reference the bases affected in the heterozygous change. Comment removed.

### SUMMARY OF YOUR PERFORMANCE IN THIS SCHEME

Assessment Category	Performance <sup>3</sup> (mean score)
Genotyping	2.00
cDNA Interpretation	2.00
Protein Interpretation	2.00
<b>Scheme result (SATISFACTORY or POOR)</b>	<b>SATISFACTORY</b>

Report approved and authorised by Simon Patton (23 February 2023) on behalf of EMQN.  
Signed:



<sup>1</sup> Maximum score is 2.00

<sup>2</sup> Deductions from the maximum score

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