INDIVIDUAL LABORATORY REPORT (ILR)*

2024 Preimplantation genetic testing for monogenic disorders (PGT-M)

Version: Final

Participant G00446

PGT-M Beta Thal 2024 - Stage 1: Feasibility Study: PGT-M Beta Thal 2024 - Stage 1: Feasibility Study

Category	Score[1]	Comments
Genotyping	1.80	Comment(s) with deduction (-0.2 marks) The online data collection form requested Distance in Mb in respect to HBB gene. The data provided does not have any units and are not Mb. (-0.1) The online data collection form requested the position of the STRs should be stated as centromeric/telomeric to the gene (-0.1)
Interpretation	1.20	Multiple markers with incorrect informativity (-0.5 marks) Limitations of test with regards to risk of recombination not discussed. (-0.1 marks) Limitations of the test were discussed but no overall figure was given. (-0.2 marks) Markers D11S1760, D11S2352, D11S4181 and D11S4891 are partially informative, not fully informative. Fully informative is when there are four unique parental alleles for autosomal disorders. (-0.5)

Embryo 1

Category	Score[1]	Comments
Genotyping	2.00	Correct result.
Interpretation	1.50	Test methodology not provided. (-0.5 marks)
Clerical Accuracy	1.70	Pagination should be included e.g. page 1 of 2. No indication that the results have been verified and the release of the report authorised. (-0.1 marks) Comment(s) with deduction. (-0.2 marks) The first column of the genotyping results table has no header and the unit of cM is incorrect. This should be Mb. (-0.2)

Embryo 2

Category	Score[1]	Comments
Genotyping	2.00	Correct result.
Interpretation	1.50	Comment(s) with deduction (-0.5 marks) High risk maternal allele should be stated instead of HGVS description because direct maternal variant testing was not performed. (-0.5)
Clerical Accuracy	--	Marked for embryo 1 only.

Member of UK NEQAS consortium

Embryo 3

Category	Score[1]	Comments
Genotyping	2.00	Correct result.
Interpretation	2.00	Correct interpretation.
Clerical Accuracy	--	Marked for embryo 1 only.

Mean Overall Score	
Genotyping	1.95
Interpretation	1.55
Clerical Accuracy	1.70

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"