# *Haemophilia Molecular Genetics — UK NEQAS (Blood Coagulation)*

**Sample ID Number:** HG22:04 **Exercise: 40** **Circulated: 5 October 2022**

| LAB ID NUMBER 28<br><br>EXERCISE NUMBER<br><br>40 | CLERICAL | | GENOTYPING | | INTERPRETATION | |
|---|---|---|---|---|---|---|
| | Full Name<br>DOB or Hospital Number | 1.5/1.5 | Technique/Extent of analysis | 0.25/0.25 | Consistent with the observed phenotype. | 1.5/1.5 |
| | Clinical Question Disorder<br>What is the clinical question & the reason for the Investigation? | 0.25/0.25 | Clear and unambiguous identification of variant | 1.5/1.5 | Mentions risk to offspring | 0.25/0.25 |
| | Referring Hospital & consultant | 0.25/0.25 | Hemizygous | 0.25/0.25 | Answers the clinical question. Can be used in family studies | 0.25/0.25 |
| **Comments** | | | | | | |
| **Score** | **2 (Max = 2)** | | **2 (Max = 2)** | | **2 (Max = 2)** | |

Scoring based upon ACGS guidelines Maximum overall score = 6 A score of <1 in any column equates to a fail.

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| SCORES IN SURVEY 39 | Sample HG22:01 | Clerical | 2 | Genotype | 2 | Interpretation | 2 |
| | | Clerical | | Genotype | | Interpretation | |

# *Haemophilia Molecular Genetics — UK NEQAS (Blood Coagulation)*

**Sample ID Number:** HG22:05 **Exercise: 40** **Circulated: 5 October 2022**

| LAB ID NUMBER 28<br><br>EXERCISE NUMBER<br><br>40 | CLERICAL | | GENOTYPING | | INTERPRETATION | |
|---|---|---|---|---|---|---|
| | Full Name<br>DOB or Hospital Number | 1.5/1.5 | Technique/Extent of analysis | 0.25/0.25 | Consistent with the observed phenotype. | 1.5/1.5 |
| | Clinical Question<br>Disorder<br>What is the clinical question & the reason for the Investigation? | 0.25/0.25 | Clear and unambiguous identification of variant | 1.5/1.5 | | |
| | Referring Hospital & consultant | 0.25/0.25 | Heterozygous | 0.25/0.25 | Answers the clinical question. Can be used in family studies | 0.5/0.5 |
| **Comments** | | | | | | |
| **Score** | **2 (Max = 2)** | | **2 (Max = 2)** | | **2 (Max = 2)** | |