

UK NEQAS Haematology

SURVEY 2302DN: DNA Diagnostics for Haemoglobinopathies

DISTRIBUTION DATE: 12th June 2023

CLOSING DATE: 23:59 (GMT); 24th July 2023

1.0 Distribution Package

This package only contains the specimens for the following survey:

- DNA Diagnostics for Haemoglobinopathies: Survey 2302DN.

A full package comprises a plastic postal bag, documentation and a moulded plastic specimen carrier holding vials of survey material and/or a slide carrier. The specimen carrier has an absorbent side that will absorb the entire contents of the package, in the event of a breakage.

The availability of spare specimens for this programme is limited. If you have problems with the specimens, please contact the Scheme by Email: haem@ukneqas.org.uk or Tel: +44 (0)1923 587111

2.0 Information required for Control of Substances Hazardous to Health (COSHH)

*This information is specific to the specimens for the DNA Diagnostics for Haemoglobinopathies survey **2302DN**. This information should be reviewed by your COSHH assessor for consideration of any changes necessary to your local work practices*

2.1 Each specimen contains DNA in TE buffer from cultured human cell lines or DNA isolated from human leucocytes.

3.0 Use of packaged material

This material is for laboratory proficiency testing as part of an External Quality Assessment programme only.

4.0 DNA Diagnostics for Haemoglobinopathies:

This survey contains 2 specimens of DNA in TE buffer: **2302DN1 & 2302DN2**.

The concentration of DNA in sample 2302DN1 is approximately 100ng/μl and in 2302DN2 is approximately 200ng/μl, as measured by the Nano Drop system.

Each sample prepared is between 20 - 25 μl in volume.

Analyse the specimens as follows, according to your laboratory's registration:

- Full alpha and beta genotype
- Alpha genotype only
- Beta genotyping only

Please note:

- For interpretation purposes, brief clinical details are supplied on the reverse of this instruction sheet.
- For performance assessment purposes, it is essential that you return results for both alpha and beta gene analysis if you are registered for them. Penalty points will be given for incomplete participation.

5.0 Specimen handling and disposal

- Store the specimens at 4 °C until tested.
- Handle and discard the material according to your local procedures for clinical specimens

6.0 Return of Results

PLEASE NOTE: The process for returning results for the DNA programme has changed.

To return your results please:

1. Go to <https://www.ukneqash.org/> and click the "Log in" button in the top right corner of the page.
2. Enter your alpha and/or beta globin genotyping results on the website and click submit when ready.
3. Refer to the "UK NEQAS (HAEM) DNA DIAGNOSTICS FOR HAEMOGLOBINOPATHIES PROGRAMME – INSTRUCTIONS FOR DATA ENTRY (PDF)" document for detailed instructions.

Background Details & Laboratory Results:

	2302DN1	2302DN2
Sex:	Female	Female
Age:(yrs)	28 years	1 year
Ethnic Origin:	Turkey	Pakistan
RBC (10¹²/L)	4.32	1.64
Hb (g/L)	77	37
PCV (L/L)	0.24	0.113
MCV (fl)	56	68.9
MCH (pg)	18	22.6
Haemoglobinopathy screen:	Hb variant detected in S window	No variant haemoglobin detected. No Hb A detected.
Hb A2 window (%)	3.8	1.1
Hb F (%)	2.2	84.1
Hb X (%)	24.5	~
Reason for referral:	Antenatal screening	Infant with severe anaemia. The family recently moved to the UK from Pakistan.

Reference ranges (adult) for laboratory results:

Parameter (Units)	Female	Male
RBC (10¹²/L)	3.80-5.10	4.50-5.80
Hb (g/L)	115-155	130-169
PCV (L/L)	0.370-0.470	0.400-0.540
MCV (fl)	79.0-96.0	79.0-96.0
MCH (pg)	27.0-32.0	27.0-32.0
MCHC (g/L)	315-360	315-360
RDW (%)	11.0-15.0	11.0-15.0
WBC (10⁹/L)	4.0-11.0	4.0-11.0
PLT (10⁹/L)	150-450	150-450

Coded comments for Recommendations:

Code	Comments
911	Testing of the baby's biological father is recommended
912	Reproductive (genetic) counselling is recommended
913	Parental testing indicated
914	Parental and/or family testing is recommended
915	Further molecular testing is recommended due to the method limitations of this laboratory
916	Information on prenatal diagnosis could be offered, according to local practice
917	Referral to a Clinical Haematologist is recommended
918	Iron studies are recommended
919	No further action is recommended on the basis of these results