

Haemoglobinopathy insights from **Variant** the ~~Abnormal~~ Haemoglobins

Programme

Egg Donation and Bone Marrow Transplant

Bianca Olivier

Haemoglobinopathy EQA Lead Scientist

Outline

Registered Participants

Why these cases?

Current NHS/SCT guidelines

Reporting UKNEQAS results

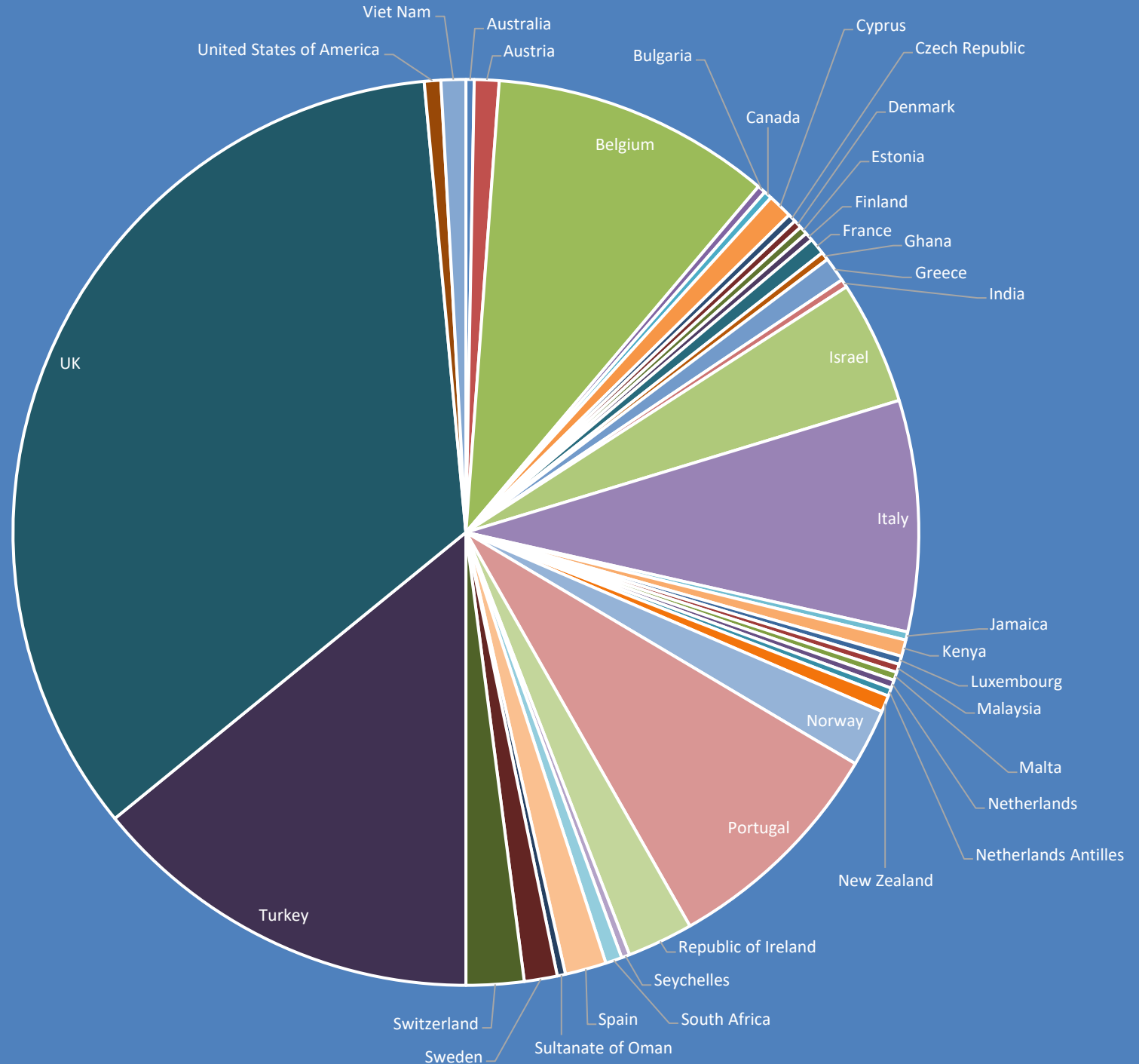
Egg Donation cases & Bone Marrow Transplant (BMT) cases

Next steps for the AH programme

UK NEQAS Participants in AH programme

2504AH (August 2025)	Total
Registered participants	340
Registered instruments	498
UK participants	117
NHS laboratories	109

Number of
participants for
the
2504AH
survey
by country



Why these cases?

- Increasing number of patients
- Maternal sample may not represent fetal genetic material in donor-egg pregnancies.
- Bone marrow transplant recipients have haematopoietic DNA that may mask fetal/parental genotypes.
- Clinical implications: missed carrier couples, incorrect referrals and missed high risk pregnancies.

NHS Sickle Cell and Thalassaemia guideline



4.1 Fertility treatment – donor gametes

If the pregnancy was achieved using a donor egg then the screening results on the woman will not be informative. The baby's biological father must be offered testing and, if screen positive, the report must recommend that the fertility clinic is contacted to obtain the biological mother's haemoglobinopathy results.

4.3 Bone marrow transplant (BMT)

Where either biological parent has had a BMT it is likely the results obtained will reflect the BMT donor rather than the biological parent and so will not accurately represent the genetic status of the fetus. If the biological mother has had a BMT, the baby's biological father must be offered testing to make sure this is not a high risk pregnancy. If DNA confirmation of the biological mother's status is required, or if the baby's biological father is post BMT and requires testing, then options that can be considered include:

NHS Sickle Cell and Thalassaemia Screening Programme

Family Origin Questionnaire

If using a pre-printed label please attach one to each copy

H. DON'T KNOW

Adoption/unknown ancestry

Donor egg/sperm (if pregnancy results from donor egg, order test for mother and offer biological father test immediately)

Bone marrow transplant (if mother has had a bone marrow transplant, order test for mother and offer biological father test immediately)

Coded Comments

Expected:

400: No evidence of a haemoglobin variant or thalassaemia

452: Testing the baby's biological father should be offered **or**

453: Testing the baby's biological father should be offered if he is from a high-risk area

Free text comment mentioning the reason for testing the baby's biological father (i.e. Egg Donation or BMT)

Incorrect Comment:

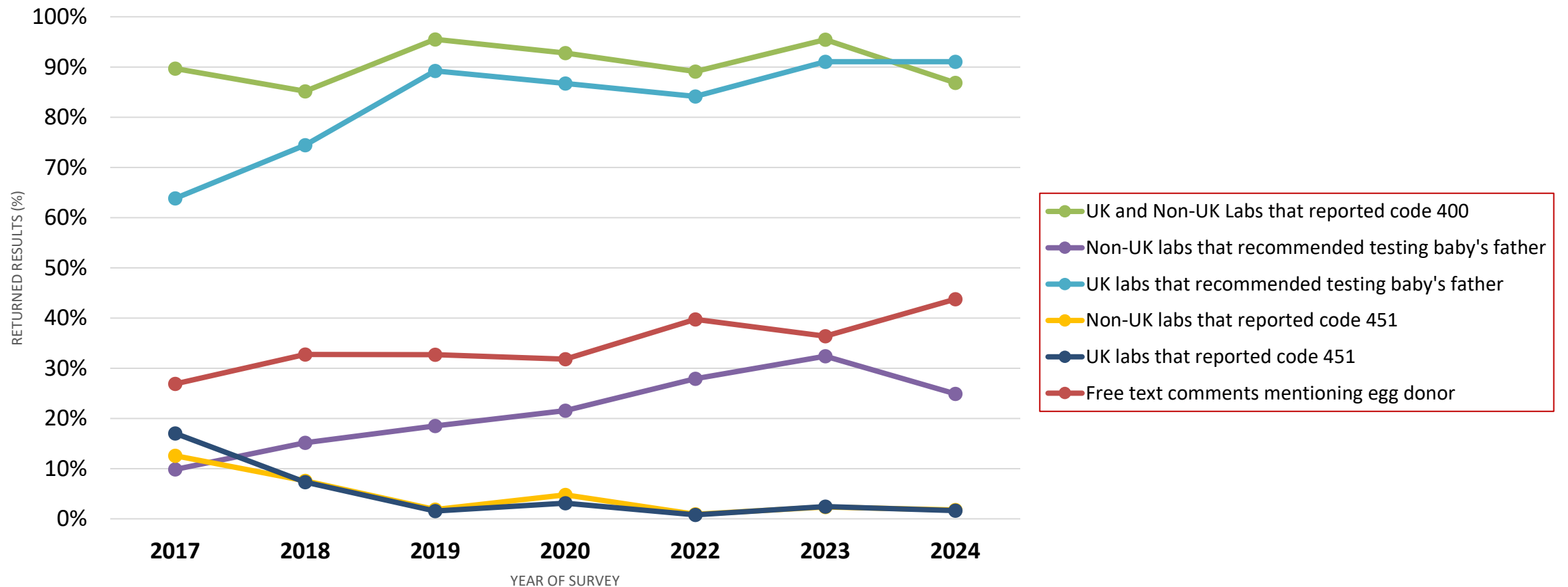
451: Testing the baby's biological father is **not** required

UK NEQAS Egg Donation Cases

	Year	Survey	Specimen	Case Details
1	2017	1706	AH1	Female, South American, 39 years old
				Pregnant via egg donation
2	2018	1804	AH2	Female, South Asian, 41 years old
				Pregnant via egg donation. Antenatal screening
3	2019	1906	AH3	Female, Russian, 33 years old
				Pregnant via egg donation
4	2020	2005	AH1	Female, Greek Cypriot, 35 years old
				Antenatal screening; pregnant by egg donation
5	2022	2201	AH3	Female, Greek, 38 years old
				Antenatal screening - pregnant using an egg donor
6	2023	2302	AH1	Female, White British, 41 years old
				Antenatal Screening (donor egg pregnancy)
7	2024	2403	AH2	Female. Indian, 40 years old
				Antenatal screening – pregnant by egg donation

Pregnancies via Egg Donation

The UK vs The Rest of the World

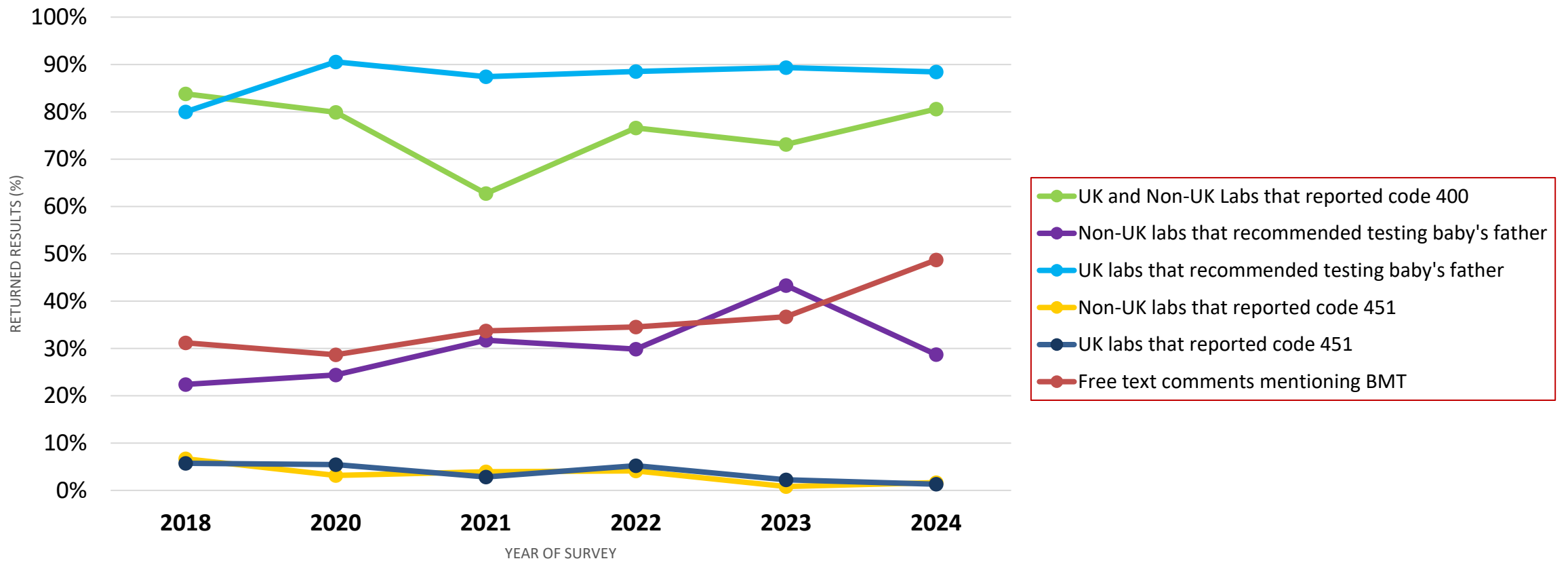


UK NEQAS BMT Cases

	Year	Survey	Specimen	Case Details
1	2018	1805	AH2	Female, African, 27 years old
				Antenatal screening. Childhood history of bone marrow transplant
2	2020	2002	AH2	Female, Caucasian, 31 years old
				Antenatal screening. Bone marrow transplant in childhood.
3	2021	2105	AH1	Female, Greek Cypriot, 25 years old
				Antenatal screening. Patient received a bone marrow transplant in childhood.
4	2022	2205	AH2	Female, Cyprus, 36 years old
				Antenatal Screening. Patient had a BMT in childhood.
5	2023	2305	AH2	Female, Greek Cypriot, 29 years old
				Antenatal screening (Had a BMT in childhood for beta thalassaemia major)
6	2024	2406	AH2	Female, Greek Cypriot, 23 years old
				Antenatal screening - patient had a bone marrow transplant in childhood

Antenatal BMT's

The UK vs The Rest of the World



Overall Trends

Code 400 is used in Egg-Donor cases more than BMT cases.

There is more consistency for BMT cases when requesting testing of the baby's biological father.

There is a general decrease in participants using code 451 in both Egg donation and BMT cases for both UK and Non-UK participants.

Appropriate free text comments mentioning that the sample does not represent the genetics of the foetus are on the rise.

The sudden decrease in Non-UK laboratories recommending testing of baby's biological father in 2024 is likely due to new registrations.

Thoughts?

- Are these cases still worth while?
- Should we be sending more or less of these cases?
- Are there other cases you would like us to mimic?

UK NEQAS Stand

Come and find me!

Email us: haem@ukneqas.org.uk

Next Steps

- Reviewing our available comments

“Due to patients' clinical history, these results do not necessarily reflect the genetic status of the baby.”

- Scoring AH interpretive comments

Interpretation Trial 2 will be sent out in November

- Exploring the possibility of providing more UK specific analysis

Change takes time



Thank you for listening