UK NEQAS Red Cell Genotyping

UK NEQAS
BBTS Technology SIG
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Findings of the UK NEQAS Red Cell Genotyping pilot

- Nomenclature
- FY GATA
- Bad luck!
  - RH variants
  - Scoring
Voting Warm Up

• The best sweet is:
  – Werther’s Originals
  – Haribo
  – Wine Gums
  – Jelly Babies
Voting Warm Up

• The best chocolate is:
  – Cadbury’s Dairy Milk
  – Galaxy Milk
  – Wispa
  – Lindt
Voting

• KEL*02.06 / 01.07
  – I know, with 100% confidence, exactly what this means
  – I know the principle of what this means but might have to check literature for specifics
  – I recognise this as something to do with genotyping
  – This is a random collection of letters, numbers and symbols
Nomenclature

– Kell is the system name that K and k belong to
– KEL is the ISBT prefix for Kell system
– K can also be written as KEL01
– k = KEL02
– Antithetical
– The Kell blood group system antigens are encoded by the Kell gene (!)
- *KEL* is the ISBT prefix code for the Kell gene
  - Note the italics for genetic information
- *KEL*\(^{*}01\) Denotes a Kell gene, with an allele responsible for K expression
- *KEL*\(^{*}01/01\)
The predicted phenotype for KEL*01 / 01 is:

- K+ k+
- K+ k-
- K- k+
Nomenclature

K SNP
  c.578C>T
  p.Met193Thr

Bs SNP
  c.1790T
  p.Leu597

KEL*01

KEL*01.07

KEL*02

KEL*02.06 / 01.07

KEL*02.06

k SNP
  c.578C
  p.Met193

Ja SNP
  c.1790T>C
  p.Leu597Pro
The predicted phenotype of \textit{KEL}*02.06 / 01.07 is:

- K- k+ Js(a-b+)
- K+ k+ Js(a+b+)
- K+ k+ Js(a-b+)
Voting

- **KEL*02.06 / 01.07**
  - I know, with 100% confidence, exactly what this means
  - I know the principle of what this means but might have to check literature for specifics
  - I recognise this as something to do with genotyping
  - This is a random collection of letters, numbers and symbols
FY GATA

- FY*01 = FY01 = Fya
- FY*02 = FY02 = Fyb
- GATA
  - A mutation in the GATA promoter region of FY gene
  - c.-67T>C
  - When present the FY gene is not expressed
  - Found only on FY*02
  - FY*02N.01
FY GATA

- Common allele in Blacks
- Essential to include in genotyping assays
- It is possible to carry an FY*02 allele but have a predicted phenotype of Fy(b-)
The predicted phenotype of \( FY^*02 / 02 \) is:
- \( Fy(a-b+) \)
- \( Fy(a+b-) \)
- \( Fy(a+b+) \)
- \( Fy(a-b-) \)
Voting

- FY*02N.01 / 02N.01. What **alleles** are present?
- FY*A/A
- FY*B/B
- FY*A/B
- FY*02N.01 / 02N.01. What is the predicted red cell phenotype?
  - Fy(a-b+)
  - Fy(a+b-)
  - Fy(a+b+)
  - Fy(a-b-)
WELL DONE!
 RH variants and RCG

– We really didn’t mean to…
– D+ phenotype
– What depth of genotyping is required?
  • 2 to 4 targets to ensure detection of psi, D-CE-Ds, DVI
  • D- phenotype
– HUNDREDS of RH variants
  • Impossible to detect all by serology
  • Unnecessary to detect all by genotyping
RH variants and RCG

– But when some participants identify a variant
  • more accurate result
  • unfair to penalise the minority
  • unfair to penalise the majority
– NEQAS try (very) hard to ensure scoring is fair and proportionate
  • Safety is paramount
Thank you very much.

Questions?