

UK NEQAS Red Cell Genotyping

UK NEQAS
BBTS Technology SIG
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Intro

- Findings of the UK NEQAS Red Cell Genotyping pilot
 - Nomenclature
 - *FY GATA*
 - Bad luck!
 - *RH* variants
 - Scoring





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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 (!)



Nomenclature

- *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



Voting

- The predicted phenotype for *KEL**01 / 01 is:
- K+ k+
- K+ k-
- K- k+



Nomenclature

K SNP
c.578C>T
p.Met193Thr

Js^b SNP
c.1790T
p.Leu597

*KEL*01*



*KEL*01.07*

*KEL*02.06 / 01.07*

*KEL*02*



*KEL*02.06*

k SNP
c.578C
p.Met193

Js^a SNP
c.1790T>C
p.Leu597Pro



Voting

- The predicted phenotype of *KEL**02.06 / 01.07 is:
- K- k+ Js(a-b+)
- K+ k+ Js(a+b+)
- K+ k+ Js(a-b+)



Voting

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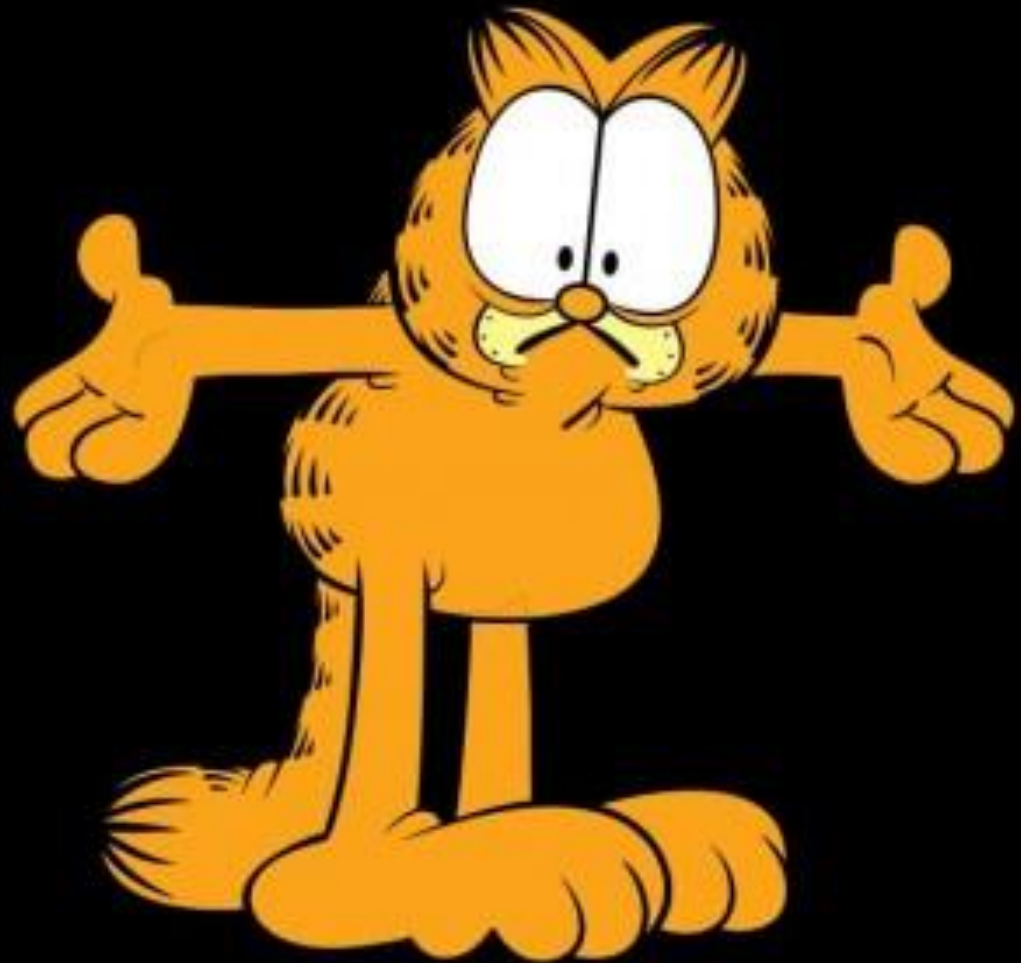


FY GATA

- $FY^{*01} = FY01 = Fy^a$
- $FY^{*02} = FY02 = Fy^b$
- 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}
 - $FY^{*02N.01}$



SO WHAT?



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-)



Voting

- 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*



Voting

- *FY*02N.01 / 02N.01*. What is the predicted red cell phenotype?
- Fy(a-b+)
- Fy(a+b-)
- Fy(a+b+)
- Fy(a-b-)



An illustration on a blue background showing five hands in business suits clapping in a circle. A white speech bubble in the center contains the text 'WELL DONE!'. Small blue triangles radiate from each hand, suggesting motion or sound. The suits are in shades of brown, black, and dark green.

**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-D^S, 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





Questions?