Latest Tooting Rare Findings.

Joint TAG on 02/04/14 at St. George’s.

1 x apparent Weak/Partial D in pregnancy, but with a normal RHD gene (as yet, unexplained). Colindale and Sheffield RCIs have seen several examples of RhD antigen expression getting weaker during pregnancy, even from Dweak to D-negative. Question of anti-D prophylaxis. Advice has been to give Anti-D.

1 x anti-Js\textsuperscript{b} + a CR1-related antibody in a sickle cell patient. Colindale SCD patient sent for full phenotype in July’13 no transfusion Hx. Referred on call in last week of March’14 – panreactive antibody, identified as anti-Js\textsuperscript{b}. NHSBT policy is to advise Rh matched K- until the antibody is made.

1 x S-, s-, U- in a sickle cell patient.

1 x \textit{RHD*DAU-0} in a sickle cell patient.

The genetic background to this type is a mutation at position 1136C>T (exon 8), resulting in an amino acid change of Threonine to Methionine at position 379 of the mature RhD protein (T379M).

1 x DOL with alloanti-D+C+E.

The genetic background to this type is mutations at positions 509T>C (exon 4) and 667T>G (exon 5), resulting in amino acid changes at positions 170 (Methionine to Threonine) and 223 (Phenylalanine to Valine) of the mature RhD protein (M170T, F223V).

1 x \textit{d(C)ce\textsuperscript{S}/Dce\textsuperscript{S}} with anti-C+E, and an auto-antibody.

The \textit{RHD} gene contains a heterozygous mutation in exon 2 of 186G>T, resulting in a Leucine to Phenylalanine change at position 62 of the mature RhD protein (L62F), but the other \textit{RHD} is normal.

The \textit{RHCE} gene contains a heterozygous mutation in exon 7 of 1006G>T, resulting in a Glycine to Cysteine change at position 336 of the mature RhCcEe protein (G336C) and a homozygous mutation in exon 5 of 733C>G, resulting in a Leucine to Valine change at position 245 of the mature RhCcEe protein (L245V).

1 x anti-k in a patient going for a TKR.

A lot of k- (KK) donor screening has been undertaken for many years. Not unusual to find ‘wet’ units on the shelf, rather than have to use the NFBB.

1 x alloanti-H in a pregnant O\textsubscript{1} lady. Patient recently referred to RCI Colindale for weak antibody investigation. Patient grouped as O Pos and reported with a weak papain+IAT antibody
showing no obvious specificity. HTL now says they have a record that the patient is a Bombay (Oh). Requested repeat samples.

1 x U- sickle cell patient.

1 x novel D variant (RHD*G209A).

The RHD gene contains a mutation in exon 2 of 209G>A, resulting in an Arginine to Glutamine change at position 70 of the mature RhD protein (R70Q).

No mutations were detected in the RHCE gene, except for the expected polymorphisms in exons 2 and 5 that are associated with the RHCE*Ce/Ce genotype. It is probable that, given the rarity of this mutation, the patient is D*Ce/dCe.

The D--/D-- patient, first reported in September 2008, with the novel mutation of del907C in exon 6 of the RHCE gene, who originally had only anti-E, and later developed the expected anti-Rh17, has now also developed anti-C and anti-c.

Colindale has had three patients with anti-Rh17 in recent years. At the time there were no D--/D-- donors on record. Had to source units from USA and Japan. NHSBT donor search showed 1 D+ C-c-E-e- donor on the system. Not picked up by anyone.

2 x Partial D DAR.

DAR has a mutation at position 602 (C>G), resulting in an amino acid change at position 201 of the mature D protein (Threonine to Argenine), a mutation at position 667 (T>G), resulting in an amino acid change at position 223 of the mature D protein (Phenylalanine to Valine) and a mutation at position 1025 (T>C), resulting in an amino acid change at position 342 of the mature D protein (Isoleucine to Threonine).

RCI Colindale referred an antenatal patient with an allo anti-e like antibody to IBGRL. Report showed that the patient was DAR/DAR with anti-Dr8. The patient had been grouped as RhD positive by the HTL and RCI but was a D variant and would require anti-D prophylaxis. Written in to RCI grouping procedures to investigate apparent RhD positives with allo anti-e like antibodies.

Apparent r’ r’ patients with anti-c are also investigated for D weak. Case seen where an apparent r’ r’ patient with anti-c was in fact RhD positive. The D expression suppressed by the presence of RhC in cis (Ceppellini Effect). r’ r’ frequency <1 in 10 000 (R1R1 frequency 1 in 5).

Fetomaternal haemorrhage estimation

HTL reported a FMH bleed of >4mL by Kleihauer. Mum O Neg, baby O Pos. RCI grouping showed mixed field for RhC and K1. Evidence of FMH bleed? FMH by flow cytometry using BRAD-3 FITC labelled anti-D was negative. FMH by FC using anti-HbF showed a minor maternal population only.
Contacted the HTL and found out:

1. The mother has βThal-trait associated with persistent adult HbF
2. The mother had been recently transfused 4 units, one of which was RhC positive and all were K1 negative.

Which explains:

1. The positive Kleihauer of >4mL, the mother's HbF is ~5%
2. The negative FMH estimation by FC using the anti-D reagent.
3. The mother's RhC and K1 mixed field results. The mother is probably K1 positive.

And emphasises:

1. The need to question results and not jump to conclusions
2. The need to have all of the facts.