NHS
Blood and Transplant

NHSBT Board Meeting
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A Patient Story: Our Unique Role in International Rare Cases

In hospital transfusion laboratories around the world, routine pre-transfusion testing is carried out to determine if a patient has any unexpected antibodies, to ensure the safest selection of blood for transfusion. In most cases this routine testing is all that is required and compatible blood can be found easily and transfused to the patient without any problems. When unexpected antibodies are detected in a patient's sample it may be necessary for the hospital to refer samples to a national reference laboratory for further investigation. In England our Red Cell Immunohaematology (RCI) laboratories fulfil this role. However, there are rare cases that national reference laboratories may not be able to solve. These cases are often patients who have a very rare blood group and have antibodies that react with the red blood cells of almost all other humans, therefore solving these cases and subsequently finding blood for these individuals is extremely challenging.

The International Blood Group Reference Laboratory (IBGRL), Red Cell Reference (RCR) department, is an internationally renowned tertiary reference laboratory which has been in existence since 1946. The laboratory has always been at the forefront of blood group discoveries and transfusion science and have amassed a comprehensive collection of rare cells and antibodies which make up a reference collection that is unparalleled worldwide. The collection, coupled with the unique expertise of the staff in the department, enables the RCR team to solve the most complex immunohaematology cases in the world. In addition, the department has been responsible for the compilation and administration of the International Rare Donor Panel since it was established in 1964. The panel is a database of rare donors from around the world and enables effective exchange of rare blood between countries for patients who need it. The laboratory receives samples from all over the world and here we describe the case of a baby boy from Croatia who needed our help.

Baby M was born with a rare X-linked metabolic disorder which was detected within his first month of life. The condition means that he is unable to produce an enzyme required to remove ammonia from his blood and the accumulation of ammonia in his blood, prior to diagnosis, lead to it crossing the blood-brain barrier which resulted in brain damage. We first received samples from Baby M when he was 6 months old. Unexpected red cell antibodies had been detected during routine pre-transfusion testing and despite extensive investigation by the referring laboratory, the antibodies could not be identified. Baby M needed a red cell transfusion but no compatible blood could be found. Our colleagues in Croatia were asking for our help to identify the unknown antibodies so that the safest blood for transfusion could be selected. We investigated Baby M's samples and found that he had an extremely rare blood group known as the McLeod

phenotype and the antibody present in his plasma was identified as anti-KL. We thawed two examples of McLeod phenotype cells from our rare cells collection and these cells were found to be compatible with Baby M's plasma. The McLeod phenotype is caused by mutations in a gene on the X-chromosome and when we sequenced this gene from Baby M's DNA we showed that a portion of the gene was missing. Clues which lead to the resolution of this case were recognised early in the investigation due to the experience of RCR scientists. Our specialist serology techniques and the rare antibodies and red cells required to solve this case were part of our reference collection. Our expertise in blood group genetics enabled us to provide molecular evidence and an explanation for the rare blood group found in Baby M. Now that the case was resolved the next challenge was to find compatible blood for Baby M. We informed our colleagues in Croatia that there were only three McLeod phenotype donors, who were the appropriate ABO group, known worldwide. One in France, one in New Zealand and one in England. The donor in France was considered the most suitable (due to matching for other blood group antigens to prevent formation of additional antibodies). Our colleagues in France responded quickly to the request from Croatia and rare frozen units were thawed and shipped directly. We received notification that Baby M responded well to transfusion of the rare McLeod phenotype red cells and the desired outcome was achieved to improve the life of Baby M. This case illustrates the unique and important role that NHSBT plays in saving and improving lives, not only in the UK but all over the world.

Rare red cells are frozen in droplets so that the very minimum amount can be thawed for testing. The collection is unparalleled worldwide.

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