Module 10 Discussion: Case Study in Savior Siblings

Cell & Tissue Engineering

Introduction

Some diseases, such as certain leukemias and anemias, require a bone marrow transplant as part of treatment. However, bone marrow transplantation requires a donor that is an immunological match to the recipient. Typically, the best option is a family member, specifically a sibling due to shared parents. Siblings have a 1 in 4 chance of matching each other based on inheritance of the same HLA genes from their parents (there are 4 possible combinations; see Figure 1). Given these odds, it is not surprising that patients often lack a sibling match. However, today a technology called preimplantation genetic diagnosis (PGD) is available for parents that are willing to have another baby to save their sick child. The technique involves in vitro fertilization (Mom's eggs and Dad's sperm combined in a petri dish) followed by genetic testing of the embryos' HLA genes approximately three days after fertilization. Embryos that are a match are implanted into the mother. At birth, cord blood is collected from the umbilical cord and transplanted into the sick sibling to replace the failing bone marrow. The baby is not harmed by the procedure. Children born in this way are known as "savior siblings"; no statistics are kept on how many such children exist today.

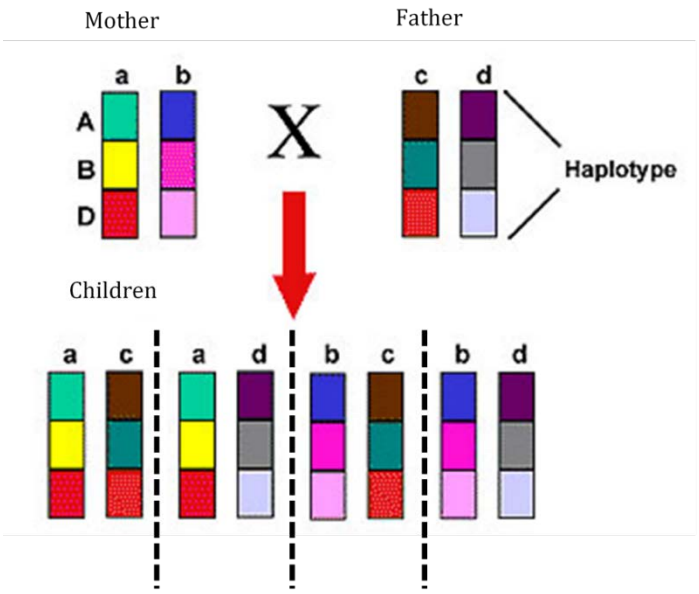

Figure 1: Inheritance of HLA haplotypes. Reference: modified from medsacpe.com

Case Study (a true story)

Jack and Lisa Nash's daughter Molly was born with a rare, incurable genetic condition called Fanconi anemia, which rendered her body unable to produce enough blood cells. Molly's best, and likely only, chance of survival was to find a bone marrow donor. The best bone marrow match is typically with a sibling, but Molly was an only child. The Nashes had always wanted to

have more children. But, because Fanconi anemia is an inherited condition, they knew that any future children also had a chance of getting the disease, for which Jack and Lisa were carriers. Eventually, Jack and Lisa Nash underwent in vitro fertilization followed by pre-implantation genetic diagnosis to choose an embryo that would have HLA genes that matched Molly and that would also be free of Fanconi anemia. After four in vitro fertilization attempts, Lisa Nash gave birth to a baby boy, Adam, on August 29, 2000. Adam's placenta was gathered immediately and all the cord blood saved. Molly started chemotherapy to destroy her bone marrow and received a transfusion of the cord blood cells a month later. The transplant cured Molly's bone marrow failure, but she still suffers from Fanconi anemia and visits the doctors 35-40 times a year to screen for solid-tumor cancers. A common cold could have dire consequences for her, but her bone marrow is functioning normally.

Reference: Rivard, L. (June 11, 2013). Case study in savior siblings. Scitable by Nature Education. https://www.nature.com/scitable/forums/genetics-generation/case-study-in-savior-siblings-104229158/