Finding a cure for the royal disease that is fit for a king
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Have you ever dreamed about being a member of the royal family? I know I have. Although it may seem like a great life where you get to jet around the world and be a part of the ruling family, there were serious downfalls to being a member of this exclusive family. During the 19th and 20th centuries, hemophilia was a major inherited disease associated with the royal family. In fact, hemophilia is called the royal disease since many royal families were affected. At least five members of royal families, including the Spanish and Prussian families, died from this disease with the youngest being only three years old [1]. Of these five, three died in car crashes, which doctors stated they would have survived with only minor injuries if not for this disease [2].

Hemophilia A and B are both bleeding disorders which are characterized by a severe lack of a protein—factor 8 and 9, respectively, both of which are produced in the liver. These two disorders have the same symptoms but are due to different protein deficiencies [1]. When factor 8, associated with hemophilia A, is absent, the body is not able to effectively stop bleeding following a cut or bruise (Figure 1). If this bleeding is not stopped, it can lead to muscle damage and death in severe cases. Hemophilia A is X-linked, which means that the males in the family are more likely to inherit this disease [3]. Hemophilia was a major problem in the 1800s for those that had the disease and were injured. Luckily, there are now treatments that can help patients stop this prolonged bleeding.

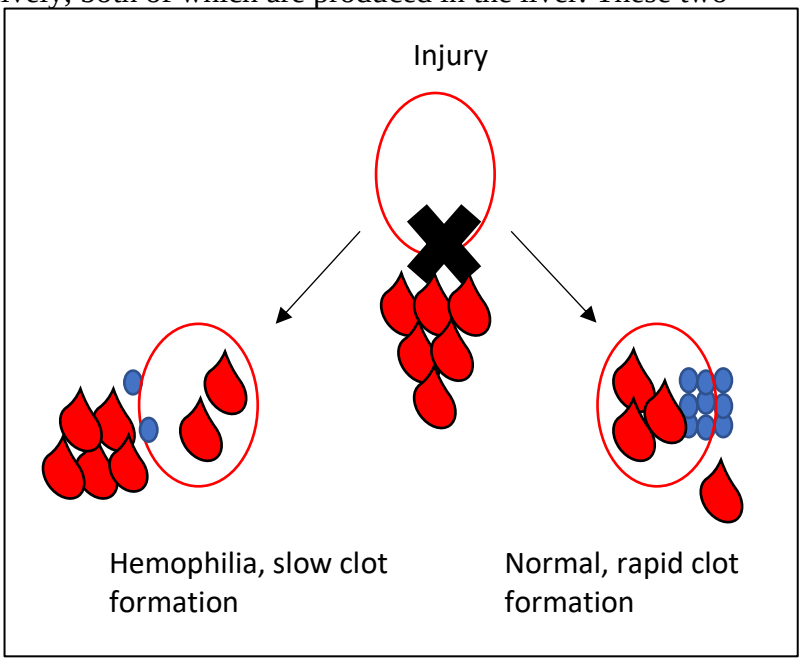

Figure 1. Illustration of hemophilia clot formation (left) following injury compared to a normal condition (right). Blue circles represent clot formation

Currently, treatment exists which allows patients to live their lives without fear of bleeding to death, but this treatment is not a cure. The treatment consists of regular injections of factor 8 protein to promote clotting during an injury and to prevent muscle and joint damage from occurring before it happens [3]. Although this treatment is effective, it requires lifelong injections and is quite costly, approximately $100,000 per year [4]. Due to these downfalls in current treatment, a cure needs to be developed.

Research into cures for hemophilia is ongoing. Scientists are analyzing the effects on mice following transplantation of cells that were derived from human stem cells. Human stem cells have the potential to become any cell in the body, which scientists can utilize for treatment. Researchers take cells from the urine of hemophilia patients and these cells can then be transformed back into stem cells for editing and future use [5]. These stem cells are selectively

transformed back into endothelial cells, which are cells that line blood vessels. Endothelial cells are also the cells that normally produce factor 8. Once derived, the endothelial cells from the hemophilia patient will be edited so that they produce healthy factor 8. Scientists then transplant the factor 8-producing cells into a mouse to determine if they can engraft into vessels and efficiently produce factor 8. This research is ongoing but holds promise for treatment options for hemophilia A patients.

Even though the so-called royal disease is no longer found in the British royal family, hemophilia A continues to afflict people. Currently,1 in 5,000 males in the United States are afflicted with this disorder, and an estimated 400,000 individuals are afflicted worldwide. The scientific community is doing everything it can to create a stem cell-based cure for all of those suffering that is fit for a king.

References

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