

Live Healthy and Be Well!

Christmas disease

--Stephen Jarrard, MD, FACS

For this month's article, I wanted to do something related to the holidays, but this is often hard when writing about different medical topics. But, there is such a thing as Christmas disease, and it has nothing to do with shopping or eggnog addiction, or anything related to this most happy time of the year. But, the name seems appropriate, and it is an interesting topic in a medical and historical sense, especially for those who might be closet Europhiles – as it has had an impact on the royal families of Europe over the centuries. The disease is named after a British man named Stephen Christmas, the first person described with the disease. Coincidentally, this “description” appeared in the Christmas edition of the British Medical Journal in 1952.

Christmas disease is more commonly known as Hemophilia B. There are two major types of this condition, A and B. Both are bleeding disorders caused by genetic mutations which bring about a deficiency of clotting factors. In the case of type A – it is Factor VIII (8). Christmas disease results from a deficiency in clotting Factor IX (9). The gene that controls the production of this factor is located on the X chromosome. As males (XY) only have one X chromosome, they are almost always the ones affected by the actual disease. Females (XX) with two X chromosomes are almost always only “carriers” of the condition, as the gene on the other X chromosome usually can produce enough to be sufficient. However, these females who are carriers can pass along the trait to their sons, who have a very good chance of having the disease.

The disease is passed through the female line of a family. Fathers who have the mutation cannot pass to their sons, but all their daughters will be carriers. Mothers who are carriers will have sons with a 50% chance of having the disease, and their daughters will each have a 50% chance of also being carriers. Women can only get the disease if both parents carry the mutation, which is very rare. In the situation of an affected family who inter-marries and thus magnifies the genetic defect, it will present itself much more often than the general population. Thus, the Royal connection to this and other genetic diseases, which leads to a “weakening” of the family line over time.

So, if you have Christmas disease, you are almost always a male with a mother who is a carrier, and you will have a serious blood clotting disorder which will lead to bleeding episodes. You are born with it, but it may not show up until later in life. At that point, blood testing will determine that you are missing Factor IX which is an important part of the clotting cascade, and thus your blood will not clot properly. Severe cases may show up in early childhood, such as excessive bleeding after circumcision or from cuts and scratches. Other cases will present later in childhood – heavy bruising or bleeding with tooth extractions or any surgical procedures.

As Christmas disease results from a genetic malformation – there is no cure. Treatment involves regular injections of Factor IX, or transfusions before a surgical procedure. These individuals must be very careful to avoid wounds and the normal lacerations and such that we all experience with mild inconvenience, but could be serious or even fatal to one afflicted. However, using

caution and regular treatment from healthcare providers, one can be expected to lead a fairly normal life in most cases. With knowledge and treatments known today, the prognosis is much better than Stephen Christmas was given during this month in 1952.

During this special time of the year, we are reminded where all credit is due, to the Great Physician who is ultimately in charge of all things. Your healthcare providers do all they can to help you with health and wellness, but your health is ultimately up to Him, who will help you if you help yourself, so we are told in the Scriptures. Merry CHRISTmas to you and your families, and best wishes for a happy and prosperous new year!

We really do enjoy hearing from you with any questions, concerns, or ideas for future columns and/or health and wellness related issues for the *Georgia Mountain Laurel*. Please send an email to rabundoctor@gmail.com, or call Felichia at 706-782-3572, and we will be sure to consider your input. This and previous articles can be now be found on the web at www.rabundoctor.com in an archived format. If you use Twitter, then follow us for health tips and wellness advice @rabundoctor. Until next month, live healthy and be well!