Aplastic Anemia, it’s not just Anemia

Dianne McMahon

Professional Studies

Brigham Young University - Idaho

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'But I have to be at work tomorrow morning'. It was hard to comprehend what I was being told. The doctors had performed blood tests and found that some of my levels were dangerously low. I would not be going to work tomorrow, or as it turned out, for several months. I would be admitted to the hospital where I would stay for several weeks. 'But it was just a nose bleed and some bruising, oh, and the fatigue. How serious could it be?' I thought. This was my introduction to aplastic anemia (AA). They first thought it was leukemia. But they weren't sure. It took over a week and a transfer to another hospital to get the diagnosis. Dr. Kew broke it to me. She said she had some good news and some bad news. The good news was it was not leukemia. The bad news was my condition wasn't any better. I had never heard of it before. It was a weird name and I had no idea what it meant. 'But it was anemia, right? So, really, how serious could it be?' I was told a lot of things, it is rare, it is not cancer, I will have to make drastic lifestyle changes that seemed peculiar. I was told about treatments, medications, transfusions, bone marrow transplants, etc. It was a lot to take in. 'But it was just anemia, wasn't it?' No, as I was to find out, aplastic anemia is not just anemia. What it is, is a disease that prevents the bone marrow from producing blood cells. Clearly, it is a serious condition. By learning more about AA, dispelling misconceptions and increasing its recognition, perhaps more can be done to help those that have it.

When it comes to AA there are deficiencies in the bone marrow so the medical term 'aplastic' is appropriate. While anemia generally relates to issues with hemoglobin, AA relates to hemoglobin, platelets and neutrophils. As Brank Cuglievan, and his colleagues point out in

their paper entitled *Aplastic anemia: the correct nomenclature matters*; “The term ‘Aplastic Anemia’ does not describe the entity as it should. A bone marrow failure of this nature should be called ‘Aplastic Pancytopenia’” (Cuglievan B., et al, 2016). Thus, pancytopenia which means low counts of hemoglobin, platelets and neutrophils, would be a more accurate term. These are some of the ambiguities that may lead to misunderstandings about this condition. Now let’s look at what AA entails.

The bone marrow produces all the components of blood, including hemoglobin, platelets and neutrophils. The marrow has long been recognized as a core component to a healthy life. Hemoglobin carries oxygen to all parts of the body. Platelets coagulate the blood at the skin level so that a wound will not bleed out. Neutrophils help the body defend itself against infection. When all three of these components are not being produced by the marrow, the result affects oxygen levels, blood clotting and the body’s ability to fight off infections. Along with other bone marrow failure (BMF) diseases, such as leukemia, myelodysplastic syndrome (MDS) and paroxysmal nocturnal hemoglobinuria (PNH), AA is diagnosed by way of a bone marrow aspiration and biopsy. At a microscopic level the marrow will appear relatively empty of blood cells and fat cells take up the rest of the space. This is where the laboratory technicians and hematologists can tell the difference between BMF diseases. There are varying levels of AA. Moderate AA (AA), Severe (SAA) or Very Severe (VSAA). In many cases the cause of AA is not known. Sometimes it is found to be genetic and sometimes the cause is from medications used in chemotherapy or exposure to certain toxins and chemicals etc. AA also has an immune response element to it. “Normally, your immune system attacks only foreign substances. When your immune system attacks your own body, you are said to have an autoimmune disease”.

(Aplastic anemia causes, AAMDS International Foundation, retrieval April 2024.) The immune system, for a person with AA, attacks the newly formed blood cells. The lack of sufficient healthy blood affects the body and leads to symptoms that any person with AA will know very well.

Prior to receiving a diagnosis, the symptoms of AA can be chalked up to everyday stress and can easily be ignored. Due to the vagueness of the symptoms and overlap with other potential causes, it is not unusual for the disease to have progressed quite far before the patient presents themselves to a medical professional. Often meeting with a medical professional is because the fatigue symptom is just so unusual and deep that it affects their normal routine. Another common symptom is catching every cold, flu or infection that is going around and that the individual with undiagnosed AA just can't shake. Symptoms could also include significant bruising and/or bleeding of the gums, or a nose bleed that will not stop. These are all effects of pancytopenia. However, once the patient does go to a doctor and is diagnosed, the proper course of treatment can begin.

There has been a vast improvement in the treatment options for AA in the past few decades. As Dr. Bhavisha Patel and her colleagues outline in their scholarly article entitled *Long-Term outcomes in patients with severe aplastic anemia treated with immunosuppression and eltrombopag: a phase 2 study*, "SAA, a historically fatal disease, now has a much-improved outcome due to recent advances in therapeutics and supportive care"; progress has been made, but there is still room for improvement (Patel, B., 2020). One example from the 1970's is illustrated by a movie starring John Travolta called *The Boy in the Plastic Bubble* (Kleiser, 1976). The film is actually based on the lives of two boys. One of the boys was Ted DeVita.

He had aplastic anemia and, for eight and a half years, was confined to a hospital room that was specifically fitted for those with immune deficiencies (McNees, P., 2004). There are varying levels of severity of the disease. If the disease has reached the Severe or Very Severe level, the first treatment they may receive is a blood and/or platelet transfusion. They will most likely need quite a number of them at the beginning and hopefully the frequency will taper off as the other parts of the treatment are administered. As with the other treatments involved, blood transfusions have down-sides. One downside is well illustrated by Ted DeVita's story. He died of iron overload. Every blood transfusion he received added to the iron levels in his blood which did not come down (McNees, P., 2004). As discussed previously, AA is pancytopenia along with an autoimmune disease. All of these elements need to be addressed. Medications may include anti-fungal and anti-viral agents to prevent or curtail any infections, an immunosuppressant to counteract the autoimmune factor, and a prescription to help the platelet count to increase. These medications often have side effects, so additional medication is often prescribed to deal with those issues. There are two other treatment options that involve resetting the bone marrow. One is a bone marrow transplant, which is the only potential cure. This has elements of chemotherapy to it and a patient will be in the hospital for several months in an isolated room to minimize any contact with germs. The other is called anti-thymocyte globulin (ATG), which allows new and hopefully healthy bone marrow to develop. This treatment means a stay in the hospital of at least 4-5 days. However, if there are complications, the hospital stay can be longer. As such, these treatments can last a long time and consequently can seriously impact the patient's routines and day-to-day life.

The effect on one's lifestyle can be quite dramatic. The impact on the family and friends can be equally difficult. In an article written by M.C. Suhocki published on August 30th 2023 in the Today magazine, American soccer superstar Mia Hamm spoke about some of her experiences when her adopted brother was battling AA. An unusual bruising prompted his doctor to run some blood work. After some difficulty in finding a donor match, Hamm's brother had a bone marrow transplant. Unfortunately, due to an infection that could not be contained, Hamm's brother passed away. This, of course, affected the family deeply. Watching a loved one go through this health battle is heart wrenching. As for the patient, the routines and lifestyle changes that have to be made to accommodate recovery from the treatment and the disease are not easy. To prevent uncontrolled bleeding, patients are told not to brush their teeth with a toothbrush, but rather to do so with a softer foam-ended stick. Other precautions include: not blowing your nose, avoiding rubbing your eyes, avoiding anything that may cause a bruise, such as playing any type of contact sport, bumping into anything or, perhaps surprisingly, taking public transit. To prevent infection, wearing a mask anywhere there may be people, which sometimes even includes family. Avoiding potted plants or cut flowers, not eating raw fruit or vegetables, washing hands after touching anything, and having very limited contact with babies or young children are recommended. Public gatherings are to be avoided. While essential to avoid or reduce the likelihood of infection and bleeding, all of these precautions have the effect of being very isolating. Physical exertion has to be curtailed, though this is not by choice but due to extreme fatigue and difficulty breathing. Showering, preparing meals, standing for any length of time, and walking any distance are all things that inevitably become undoable for someone with AA. Sleeping, then eating whatever you can get down, and doing the bare minimum of

other essential activities often comprise the main activities of one's new reality. The numerous appointments to see doctors, have transfusions, have blood tests, and to have the dressings of intravenous ports cleaned take their toll physically. Texting a few of your close friends helps with the emotional toll. Time off work can be for weeks, months and sometimes years. As Dr. Kew said, although it is not leukemia, it is not any better. One of the reasons it is not any better is the rarity of the disease.

AA is a rare disease, and according to Christine A. Moore and Koyamangalath Krishnan in their scholarly article entitled '*Aplastic Anemia*', it affects only "0.6 to 6.1 cases per million population" in North America (Moore, A. et al, 2023). This presents challenges. First, it is hard to find another patient from whom to gain support. I only know one other person with AA in my city of 1 million people, and she is about fifty years younger than I am. The hematology team at the hospital here that specializes in BMF diseases see only a handful of patients that have AA. Fortunately, there are a couple of other resources available now to connect with other AA patients. For instance, through Facebook there is an international support group for those with AA. In the Eastern Canada chapter of Aplastic Anemia Myelodysplasia Association of Canada (AAMAC) there is only one active member with AA who attends the Zoom meetings. All the others have MDS. When doing research for this essay it was found that the doctors at the hospital in Ottawa, who specialize in BMF diseases, have not written any research articles on Aplastic Anemia, but a number of articles were found regarding leukemia and MDS. This scarcity of research brings us to some difficulties that can arise from lack of awareness.

As was in the case for Mia Hamm's brother, it can be very difficult to find a bone marrow donor who is also a match. Patients wait for months, years and sometimes will never

find one. There are very specific criteria that have to be met in order for someone to be a donor and a match. The bone marrow donor needs to have healthy blood, he or she (usually he) has to be of the same ethnicity, and preferably in their twenties. Some of these criteria may be adjusted if the donor is a family member. It can be a challenge socializing people to the importance of blood and blood product donation. Another challenge is the cost of AA medications, which can be outrageously expensive, and medical insurance is often required. Funding is difficult to get since few people are aware of the disease and the need. So, what can be done?

Five years on and I am still feeling the effects of AA. While most of the key AA indicators in my blood counts are within the normal range now, my platelets are still low. However, they are no longer in a range that is concerning to the doctors. I consider myself very fortunate. I have had the luxury of recovering without the need to worry about how I am going to support myself. With Medicare and disability insurance from work and the government I have been able to get by and I am not in debt. Living in a major city, I have had access to the necessary AA treatment options and have had an excellent level of care. Not everyone has had this same luxury. One of the ways to spur improvement in outcomes for AA patients could start with changing the name. Just as the change in terminology from amyotrophic lateral sclerosis (ALS) to Lou Gehrig's disease has helped increase awareness of that disease, rebranding AA as Ted DeVita's disease, for example, may improve recognition of this condition. Alternatively, using a more accurate nomenclature like aplastic pancytopenia, may help the medical research community and newly diagnosed patients gain a better understanding of the physiological and other impacts that occur in the lives of those affected by AA, which has been misunderstood and mislabeled for so long. Joining celebrities, like Mia Hamm, and advocating for more funding

and research may bring this disease out of obscurity. Additional awareness campaigns that leverage the uniqueness of the disease to tell a compelling story in the entertainment industry could go a long way. There are viable options to increase AA awareness and funding for research and to improve AA outcomes. All that is required is the will to carry them out.

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