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INSIGHTS

Family reflections: thalassemia and Ammar

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My name is Ali Oonwala. My wife Khadija Oonwala and I both carry the thalassemia trait. This means that for each pregnancy we have there is a 25% chance of a child having Thalassemia Major. Our second child was diagnosed with Thalassemia Major at 2 months of age. At the time our family was devastated by the news and did not know what to expect.

We did not want to put Ammar through all the testing and many needle pokes at such a young age. But from such a young age we knew our son was a trooper and so strong that he could deal with it. It's so funny how things work, but our older son, Yusuf, who is only a carrier and healthy like my wife, and I literally will faint at the sight of a needle. But can you blame him? He could not take watching his younger brother getting poked all the time. He would say "Papa, I hate all the doctors and nurses. Why do they keep hurting Ammar?". Yusuf was only 3 years old at the time Ammar was born and diagnosed.

Now fast forward 10 years and Ammar although living with Thalassemia is an otherwise healthy and completely normal 10-year-old boy. His best friend and older brother Yusuf is 13 years old. They are rough and tough with each other like any other siblings.

The only impact to Ammar's life currently is having to go get a blood transfusion once every 4 weeks. This means he misses half a day of school once a month. Ammar has learned to live with it and like I said from day 1 has been a trooper. He's the kid that falls, doesn't cry, gets right up and keeps moving. Yes, does he ask why I have to go every month…. of course! But we explain why, and he gets it. He is in great hands at the Alberta Children's Hospital. The nurses and doctors have literally seen him grow up and we owe a debt of gratitude to the team there for taking such good care of him.

From the age of 2 months, Ammar has received a blood transfusion every 4 weeks. This is essential to his life. Without these transfusions, his hemoglobin would drop to dangerously low levels eventually where he would have potentially major issues. However, since he was diagnosed so early in life, he has been immune to any issues. But he does need to be monitored annually for iron build up in vital organs (mainly the heart and liver). This is done through magnetic resonance imaging (MRI). Like anything, Ammar struggled with it at first, but now has become accustomed to it. With his blood transfusions iron can build up in the body, so he needs to take an oral medication known as a chelator to remove excess iron, so it does not build up in his heart or liver. He has to take that medicine daily. The annual MRI is a routine test just to make sure the medicine is working and there is no excess build up in the heart or liver.

There is no cure for Thalassemia Major other than a transplant. However, finding a match is very tough as there is only a 25% chance that a sibling could be a match for a bone marrow transplant. Without a full match, most transplant teams do not recommend going through a transplant because of the risks and also the fact that treatment is so good these days and patients live nearly normal lives even into their 70s in some cases.

That being said there is much attention being paid to Gene Therapy currently. I would love to see research focused on this area. The major benefit is that potential risks are much less with Gene Therapy versus transplants. I would also like to see more research being done on unmatched donors and transplants. With science evolving so rapidly this may open more doors for patients looking for a cure, but unable to find a match, which is the majority of patients, unfortunately.

Overall, the outlook is great for Thalassemia especially in first-world countries where access to treatment is not a barrier.

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However, through my volunteering overseas I have seen firsthand how different the situation is for Thalassemia patients living in third-world countries. I would love to see research in these areas where access to therapy and more knowledge about the condition could be more readily available. Of course, as more immigrants move to Canada, the same will need to be done here. New novel drugs are becoming available where hemoglobin levels can be maintained in the body without the need for transfusions. These are currently only available to adult patients. It would be great to see more research done in this area for younger patients to have access to treatment sooner. This is probably currently the biggest issue for Ammar as there are new novel treatments available (i.e., gene therapy, oral meds), but he is too young to start them. It would be great to see more research efforts focused on pediatric patients where the impact could be vast. The sooner the access to therapy the sooner younger patients can stop getting routine i.v.'s, transfusions, and other regular monitoring tests like MRI's being done. This in turn could also save the health care system major dollars as the costs to care for patients annually is extremely high.

Lastly, I think another effective approach would be for young parents and patients to have more resources to discuss at diagnosis. Many families feel overwhelmed at such a fragile time and having someone to reach out to that has been through it already could be profound. The Thalassemia Foundation of Canada does a great job of outreach and maybe this could be a joint venture working together with treatment centers. But as always living in Canada allows us so much already in terms of access and treatment, but if we can get other countries similar resources we would be able to help so many more families.

COMPETING INTERESTS

The author declares no competing interests.

ADDITIONAL INFORMATION

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