



OP-EDS

Congress members must strive to keep the spotlight on lesser known illnesses

By Rep. Carolyn McCarthy (D-N.Y.)

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Several years ago, I was introduced to a brave woman named Marie Arturi. Marie came to me because her daughter had recently fallen victim to an exceptionally rare bone marrow failure disorder called Diamond Blackfan Anemia (DBA). She wanted people to know about the existence of the disease and the devastation it has caused to her family, amongst only a scattered handful of others. There currently is no cure for DBA other than stem cell (bone marrow) transplantation, and the treatments have limitations.

I began to explore what I could do from my position as a member of Congress to help encourage research into rare diseases like DBA. I was fortunate enough to secure appropriations funding for upkeep of the Diamond Blackfan Anemia Registry of North America, maintained at Schneider's Children's Hospital. Registries like the DBA registry at Schneider's have the ability to collect clinical and demographic information on diseases that show significant gaps in the natural history of the disorder.

Recent years have been a very good time for research on orphaned and rare diseases like DBA. The National Institutes of Health and Centers for Disease Control and Prevention have recognized for years the great value of supporting DBA initiatives that have yielded valuable insights into the biology of blood disorders, blood cell formation, leukemia, the genetics of birth defects and more.

Thanks to these research efforts and the efforts of the DBA registry, there are exciting new areas of science being explored following the identification of DBA's link to this intercellular structure, which is an important example of the profound potential of these initiatives and justification for their continued support. This discovery may well yield the keys to finding cures for a host of rare blood diseases.

Last fall, by passing H.Res. 524, legislation I introduced to express the sense of Congress regarding Diamond Blackfan Anemia, the House formally acknowledged these scientific and clinical achievements and praised the NIH and CDC for the foresight demonstrated in their active involvement. As well, the work of the Daniella Maria Arturi Foundation, Schneider's Children's Hospital, and the Diamond Blackfan Anemia Foundation were commended for their dedication and cooperation in identifying the markers of the disease.

While I am very pleased to have been successful in helping to bring both research dollars and recognition to DBA, it is unfortunate that so many rare diseases continue to go without such recognition.

Recently, I have been working to raise awareness for another rare illness known as Inflammatory

Breast Cancer. IBC is a very rare yet very aggressive type of breast cancer that is little known and often misdiagnosed until it is too late. My interest was piqued when I was approached by one of my staff to ask if I had ever heard of the disease — even with over 30 years of work as a nurse, I had not.

As it turns out, most medical professionals and women are unaware of the disease and its deadly potential (survival rates are significantly lower than with other types of breast cancer). Unfortunately for many women, this lack of awareness has cost them their lives.

I set out to learn more about IBC, and in the process determined that I needed to work to make sure that other women and medical professionals knew about the disease and its deadly potential. For this reason, I introduced H.Res. 1300, supporting efforts to raise awareness, improve education and encourage research of inflammatory breast cancer. If only one woman or doctor becomes aware of IBC and is able to self-diagnose or diagnose the disease before it is too late, then the goal will have been achieved.

I will continue to work to raise awareness about orphan diseases. We cannot risk people losing their lives or watching their loved ones suffer from illnesses simply because of a lack of awareness. Americans need to know the risks of illnesses that might not currently be on the tips of most people's tongues but have devastating consequences. Congress needs to work with the next president to make sure that all Americans have affordable access to healthcare to cover the costs of treating these lesser-known but devastating diseases.

McCarthy is a member of the House Education and Labor Committee.