To the CIRM Board of Governors, in support of Dr Bacchetta CLIN1 application 11591.

In February 2013 our lives changed forever when we were told that Giacomo, our son, had IPEX. Having a child diagnosed with IPEX was a traumatic experience. Moreover, I knew the only effective cure to this disease was a bone marrow transplant. This meant that he needed someone with the same tissue type as his to donate their stem cells to him and hopefully give him the opportunity to beat this disease, and live the rest of his life. Some people are lucky enough to find matches in their family, but that wasn't our case. Giacomo and our family were going to rely on an unknown person to save his life; I didn't know what to expect for myself, my family and, above all, for my little Giacomo.

The diagnosis for Giacomo came after 12 years (all his life) of being very sick. He could not eat anything without being sick, his body was skin and bones, and he could not have a normal life like his peers. IPEX was suspected based on a new test that was established at that time by the group of Dr. Bacchetta at San Raffaele Hospital in Milan, Italy. Giacomo did not have the typical disease presentation so, although we were devastated, we were also very thankful that he could be tested and correctly diagnosed.

At first Giacomo needed a long period of rehabilitation to a normal nutrition, and at the same time he started taking immunosuppressive drugs, which gave him immediate benefit, but were very difficult to keep at the right levels in the blood. All the time they went off range, Giacomo developed very painful ulcers in his mouth.

We saw improvement with medications, but he always had to go to the hospital to be checked and we all knew this could not be forever. The option of the transplant was difficult because of the many possible complications, especially when using an unrelated donor. After 3 years of immunosuppression we opted for the transplant and Giacomo luckily is now cured, but I know that this is not what many IPEX patients experience. The option of a personalized medication, that could specifically provide to Giacomo what he was missing, his own functionally normal regulatory T cells, would have been ideal. Therefore, we would like to strongly support the work of Dr. Bacchetta and her team at Stanford to have the possibility of advancing new treatments for IPEX patients.

How you choose to cope with IPEX depends on how you perceive it, your feelings, your personality and your own individual way of facing the problem. You may wish to be active and research all medical and alternative options, you may decide to hand over control to others, or combine the two attitudes. We opted for the last. Talking to Giacomo about his disease was difficult. It is hard to know how to talk to a child about being in hospital for several days a month.

IPEX can be treated with a bone marrow transplant but finding a compatible bone marrow donor can be difficult, and the transplant procedure is often risky because people with IPEX may be very sick. Due to the limited number of cases, it has been difficult up to now to compare different therapeutic strategies and their outcomes. Therefore, therapeutic approaches for the treatment of IPEX patients are still based on the experience of single patients. Research is their only hope.

Francesca, Domenico.

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