

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

Good morning,

We are the parents of Riccardo, a boy, who is six years old. Four years ago, Riccardo was very sick. He had been seen by multiple doctors, hospitalized in multiple hospitals where he was discharged with multiple diagnosis and no effective treatment. We watched him lose weight, becoming weaker and weaker every day, skinny but with a puffy tummy. He did not have any desire to play and always asked to rest on our arm to seek comfort from what seemed to be a dark evil. Then, finally we received the correct diagnosis, that no parent would ever want to hear; Riccardo, we were told, was suffering from IPEX, a rare disease of the immune system. We feel no need to give the scientific description of his disease nor recount the desperation we felt when we were told that the only solution for a possible cure was a bone marrow transplantation, provided we could find a compatible donor.

Riccardo started receiving immunosuppression that fortunately made him feel better, but for how long and at what price was a constant fear, and immunosuppression was not a cure and has to be given for the rest of his life. The doctors were our hero's, providing hope and light because they dedicated their lives and their work to search for a cure that can save our child, and other children just like him. Our second son Marco tragically also has the same mutated gene as his little brother, but he has not so far developed the disease. Of course our hope is that he will not get sick, but we must be prepared. Our hope and dream are to have a cure, for us and all the other families living with this chronic and debilitating illness.

For this reason, with a heart swollen with gratitude, we support Dr. Bacchetta and her collaborators. Indeed, our children's future depends on the dedication and passion with which these studies are carried out. Because maybe one day, this terrible experience will just be a very bad memory, and because one day IPEX will no longer be the incurable monster we know and fear. At that time, we will know that the monster has been defeated.

Signed letter

Paolo Machnig e Natasha Michielan