

Dear Members of the CIRM Board,

Cade Jobsis, my 3-year-old grandson, is one of the children diagnosed with SPG50, a rare neurodegenerative disorder caused by mutations in the AP4M1 gene. These mutations have limited Cade's development in a global sense. He is delayed in speech, movement, and cognition, generally. For a long time, doctors, nurses and therapists who worked with Cade told his parents he was just "slow" and that he would "catch up" to other children eventually. But he wasn't catching up, or in the ways that he was, it was much too slowly not to be a red flag. That is when he was genetically tested and was found to have the above-mentioned gene mutation, a mutation for which there currently exists no approved treatment, a condition that has devastating effects on affected children's lives as it causes the breakdown of neurons that support both motor function and cognition.

It sounds cliché to say that Cade is the light of my life and that he brings light and laughter everywhere he goes, but it's true. Despite his disabilities, his language-deficits, cognitive difficulties, and physical struggles, he is always smiling and always trying so hard to do the things he cannot do because of his genetic disorder. And because he tries so hard, and his mom (especially his mom), dad and therapists work with him so long, sometimes he makes progress. He can actually walk independently for a short distance now, something we didn't know if he'd ever do. Then we imagine how much more he could gain with the gene therapy that could be made available with support from CIRM, not to mention now much he could lose without it, which is virtually everything: his ability to walk, even with assistance, his ability to move his arms and legs and other extremities; ultimately, he likely would become nonverbal. A vegetable is what they used to call such people.

Right now gene therapy is the only possible treatment, and it is one that has shown some efficacy, both in rodent testing and in compassionate use in humans. Right now this therapy is my grandson's only chance. Please let him have it. Eight doses of the gene therapy medication are ready, waiting to be administered. If you won't fund a trial to test efficacy so that this treatment can be available to all of the children who need it or will need it, then please publicly suggest that Elpida release these doses for compassionate use, if such use is legally possible.

Sincerely,

Rosalyn Rossignol