



May 4, 2023

Tomás J. Aragón, M.D., Dr. P.H.
Director and State Public Health Officer
California Department of Health | Newborn Screening Program
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Dear Dr. Aragón

We, the parents, grandparents, and clinicians of children with Pyruvate Dehydrogenase Complex Deficiency (PDCD), are submitting this letter of support for Dr. Jirair K. Bedoyan's application to expand his pilot study of adding PDCD to the newborn screen in California. **Hope for PDCD** is a patient advocacy foundation based in Folsom, California. It was founded by UC Davis alumni, Jon and Frances Pimentel, after their daughter Violet was diagnosed with the condition. Hope for PDCD is dedicated to improving the lives of patients with PDCD through research of new precision/genetic medicine, increasing equitable access to diagnosis, creating standards of care, and more. PDCD is a disorder of carbohydrate metabolism and patients need to be on a ketogenic diet as soon as possible. The early implementation of a ketogenic diet is critical for a good outcome in PDCD. From Dr. Jirair K. Bedoyan's pilot newborn screen study in Ohio, we now know the prevalence of PDCD to be 1 in 40,000 births. That means in the next year, 10 babies will be born in California with the condition. 10 lives that can be saved with newborn screening.

Spend five minutes getting to know our community and you will know our lived experiences with the positive effects of a medical ketogenic diet (or, for some milder cases, a modified Atkins diet). The diet stabilizes our children, brings down their lactate levels, allows them to meet developmental milestones that were previously missed, and improves sleep and mood regulation. For as many uplifting and life changing stories about the ketogenic diet, there are just as many heart wrenching stories about the diagnostic odysseys our families face. A majority of patients have a de novo (meaning, not inherited) mutation, making it all the more critical that PDCD be added to the newborn screen.

While we could write a book of testimonials, we will share just a few:



My daughter Aela is an angel on earth. She is six years old, but for the first two years of her life she did not have a diagnosis. Two years of potentially avoidable seizures, harsh medications, and expensive steroid injections. Two years of severe swelling, irritability, dangerously high blood pressure, further brain damage, and daily vomiting from feeding her carbohydrates. Unbeknownst to us she was in lactic acidosis. We rarely saw her smile or make eye contact. Fast forward to today. Finding her PDCD diagnosis and a plan of care has brought Aela to life. She has eyes that look into your soul, she smiles when she looks at you. She is stronger, she doesn't vomit daily and, most importantly, she is here with us today.

- Katy Cairo, Mom to Aela, Diagnosed at 2 years | San Diego, CA



Violet Pimentel was born at Mercy Folsom after a healthy pregnancy and uncomplicated delivery. After passing all her newborn screens in the hospital, she came home 24 hours later. In the months following, we raised our concerns with doctor after doctor about her feeding difficulties, developmental delays, and lethargy. After months of being dismissed and being told that she would probably be fine with a little physical therapy, Violet was finally admitted to UCSF and diagnosed with PDCD. After starting the keto diet, Violet went from not tracking objects to tracking objects with no visual delay. She went from constant discomfort to smiling and babbling. She went from a floppy baby with a severe hypotonia diagnosis to a content baby that can prop sit, roll and now is trying to army crawl.

- Frances Pimentel, Mom to Violet, Diagnosed at 9 months | Folsom, CA



If Luke had been diagnosed at birth we would have been given a two-year head start in giving him all the necessary tools to thrive. These include the ketogenic diet and additional supplements that are vital for Luke to grow, achieve developmental milestones, and have enough energy to live life. Due to this lack of knowledge, Luke spent 10 weeks in the pediatric intensive care unit at Kaiser Oakland. He was initially admitted for an RSV infection, but was ultimately diagnosed with PDCD while inpatient. He was extremely weak and the outlook was grim. If Luke had the proper nutrition from the start, he would have been in a much better position to fight the RSV infection and achieve milestones. Knowledge is power and without the knowledge of our son's condition he has been at a serious disadvantage for the past two years.

- Sarah Matthews, Mom to Luke, Diagnosed at 2 years | Vacaville, CA

Signed by:



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cc:
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