Join Antonio: an injection of energy and hope in the face of rare disease

LOCATION: San Fernando (Cádiz)
DURATION: 2'09"

SUMMARY: Antonio, a three year old boy from Seville, is one of 8 people in the world to have TK2-related DNA Depletion Syndrome, Myopathic Form, classified as a rare disease due to its scarce prevalence. 6 months ago, he was in a critical situation, however his parents researched and found an experimental treatment in the USA. They have covered the cost until now. Antonio's improvement is an invitation for hope.

VTR

Today, Antonio plays and smiles like any other three year old child. However, he is one of the 4 people in Spain, and 8 in the world, who live with TK2-related mitochondrial depletion syndrome, Myopathic form.

ANTONIO BLANCO
Father

"The mitochondria are where energy is produced in the organism, and he has a defective gene which means that this energy is not produced."

ELENA ÁLVAREZ
Mother

"The diagnosis, which we received 9 months to a year ago, was to assume and wait that the end was near."

But this is not a sad story, nor one of resignation. Antonio and Elena did not accept this ending; they investigated for themselves, and in the USA they found an experimental treatment which has been approved by the health authorities. This little boy now has the energy he once lacked, and his extreme thinness has disappeared. You can't see it, but he has the face of a happy child.

ANTONIO BLANCO
Father

"What it does is bypass, essentially, the genetic defect that he has. We externally administer the enzyme that he lacks."

LY HAFNER
Association of Mitochondrial Disease Patients

"These children need rehabilitation from the day they are born, they need treatment, and no one else spends money on it, the family has to pay for it."

This is the collective demand of people with rare diseases. They ask for research and for the public health system to pay for the treatments. Antonio and Elena have spent 40,000 euros on medicine in the last 6 months. The Andalusian Administration has promised to reimburse them.

ELENA ÁLVAREZ
Mother

"From today, there is a massive glimmer of hope."

ANTONIO BLANCO
Father

"And no sadness, the will to fight, live, and continue looking for solutions to the problems."

The World Health Organisation has identified 7,000 rare diseases, the least common throughout the population. In Spain, there are 3 million people with one of these serious illnesses. 3 million stories. Antonio's one, however, is not sad, but a story of hope.

For more information or support please call +34 662 369 820 or email info@andalusianstories.com