RARE DISEASES: A FOUNDATION FOR ALE AND CHILDREN WITH THE KIF1A GENE MUTATION

By *Barbara Bertocchi* (Giornale di Brescia – local newspaper)

Ale's father is founding an association in Italy to support Alessandro Andrea Lamari, a 6-year-old boy suffering from a neurodegenerative disease diagnosed in just 18 people across Italy and 550 worldwide.

Alessandro Andrea Lamari is just 6 years old.

In Ale’s eyes, one can see the vastness of the ocean, the empathy he shares with others, and the physical pain he endures multiple times a day. Ale - officially Alessandro Andrea Lamari - lives in Brescia with his mother Benedetta, father Ennio, and his older brother Davide. Ale is afflicted by a neurodegenerative disease so rare that only 18 people in Italy and 550 worldwide have been diagnosed with it. "It involves a mutation in the Kif1A gene," explains his mother, a professional gynecologist. "The effects can vary greatly depending on which part of the gene is mutated, ranging from leg problems to cognitive delays, spasticity, and epilepsy." Benedetta and Ennio first became suspicious when Ale was 13 months old: "He was too calm, wouldn’t try to stand or walk, behaved oddly with toys, and didn’t respond to certain stimuli."

The Diagnosis

The pediatrician tried to reassure them, but they sought advice from a child neuropsychiatry specialist. In September 2019, the initial diagnosis was autism. "Ale then began a behavioral-psychomotor therapy program," they continue. "During those months, we noted every small progress on the calendar: 'Today he took seven little steps.' Meanwhile, we managed to consult with Costanza Colombi, an expert on early autism diagnosis and author of reference books; due to COVID,

she assisted us remotely, emphasizing the importance of immediate intervention. Indeed, the progress was noticeable." Then, in February 2021, "the results of the genetic tests came in, and we finally learned the exact name of his condition."

Now Ale can walk short distances, does not speak, and makes vocal sounds, "but he makes himself understood with his eyes and hands," his parents emphasize. "One of our major concerns is the severe leg pain he experiences several times a day. The medication we give him provides some relief, but does not solve the problem." Research related to this disease is ongoing in the United States at Harvard University and Boston Children's Hospital: "They are testing a drug on a girl with promising results. However, her gene mutation differs from Ale's. He needs something else. We remain hopeful…"

The Mission

Last summer, the family traveled to New York to meet other children and adults with the same genetic disease as Alessandro: "It was a touching and emotionally enriching experience: for the first time, we realized we were not alone, and that mutations in the same Kif1A gene can have very different consequences: there are children who cannot walk, a young woman who graduated at college, and even a 60-year-old musician who only recently discovered his condition."

In the United States, father Ennio conceived the idea of founding a national association of little superheroes with Kif1A also in Italy: "beside the USA association, in Europe here is one in Spain and France, but not in Italy. Ours will be operational in a month: it will help raise funds to advance research and also spread information about the disease so that other families dealing with unnamed syndromes can identify with our issue. We are few, but together we can do a lot. For our children and for the children of tomorrow." Because the little and big superheroes who, like Ale, communicate with their eyes, need to be heard.