The medical odyssey of an undiagnosed child

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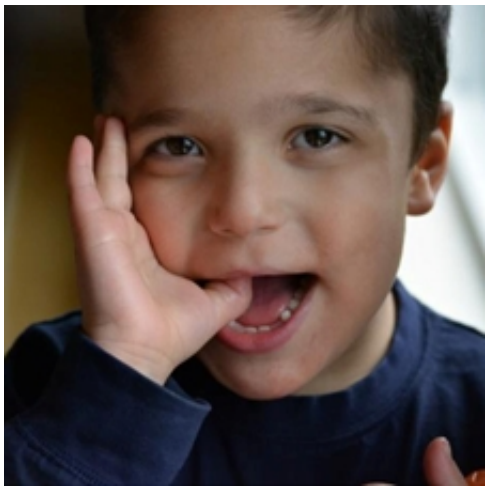

Children born with ADNP-related autism syndrome suffer from a heart-breaking combination of ambiguous developmental problems injurious to both their physical health and cognitive functioning. For parents, the mystery surrounding their infants' suffering can be even more agonizing than the syndrome itself, which has no known cure.

Recent research from Tel Aviv University and the University of Antwerp is easing some of that agony. Activity-dependent neuroprotective protein (ADNP), discovered by TAU's Prof. Illana Gozes 15 years ago, has now been shown by Prof. Frank Kooy at the University of Antwerp to be mutated in autism, causing developmental and cognitive delays, speech and feeding difficulties, heart abnormalities, and a laundry list of other debilitating symptoms.

"Nothing is as frightening as having a son with an unknown medical/genetic condition," writes Sandra Bedrosian Sermone, the mother of Tony, a child with the syndrome, in a recent *Journal of Molecular Neuroscience* article. "What does this mean? Will he be normal? Can they fix this? Will he die? Is his twin brother OK? Was this because of me? It was agonizing waiting to get the results of that first genetic test. But the call came, and it would be the first of many in this journey of an undiagnosed child."

A 15-year-old breakthrough

Prof. Gozes, the incumbent of the Lily and Avraham Gildor Chair for the Investigation of Growth Factors and former director of the Adams Super Center for Brain Studies at the Sackler Faculty of Medicine and a member of TAU's Sagol School of Neuroscience, first discovered ADNP as well its crucial role in brain formation, learning, and memory. For 15 years, she has been leading the crusade to understand ADNP and to develop a drug reversing the effects of ADNP deficiencies, which have been linked to schizophrenia, Alzheimer's disease, and dementia, among other neurological and psychiatric diseases.

Tony was diagnosed at the age of five with a new mutation of ADNP previously seen in only 10 other children around the world. The family's "aha" moment, as Sermone calls it, occurred after complete exome sequencing conducted at Duke University revealed the specific genetic mutation.

While the diagnosis was a blow to Sermone and her family, knowledge is power. Through Sermone's Facebook page, more and more people have gained awareness of the syndrome and its probable cause. "Our journeys have all been different in ways, but identical in others," writes Sermone. "[My son] has earned the name 'Superman,' because he is the strongest, toughest, sweetest little man on the planet, and he is helping other children every day by being a part of this new genetic testing and syndrome research with the team and doctors who discovered it."

Creating public awareness

According to Prof. Gozes, who is intimately familiar with Tony's case and that of other ADNP children, greater public awareness of the syndrome will more likely lead to the development of more appropriate care in the future.

"I hope it will become a routine screening in the future for undiagnosed cases that come to the clinic," said Prof. Gozes. "Teaming up with Prof. Kooy's Cognitive Genetics group in Antwerp and other leading researchers around the world will promote our understanding. Early diagnosis is key so these children can obtain the right training and the right help, and so the parents don't feel so alone in the world. The more we understand about the mutation, the better we can understand why on earth the gene is susceptible to mutations."

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