

CARES Foundation looks to serve those with rare disease



(Photo: ~Courtesy of CARES Foundation)

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While the CARES Foundation is completely unknown to many people, there is a small but significant group of the population that heavily relies on the organization.

That group is those affected by CAH — Congenital Adrenal Hyperplasia — a family of rare genetic diseases that can be life-threatening to male and female children and will affect them for the rest of their lives.

The term CAH most often refers to 21-hydroxylase deficiency (21-OHD), the most common form of the disease, which is characterized by an absence of the important 21-hydroxylase enzyme in the body. Without this enzyme, the body's adrenal glands are unable to produce cortisol, an essential hormone responsible for blood sugar levels, and physical and emotional stress.

"The emotional toll and the feeling of helplessness that this mom had really resonated with me," Dina Matos, executive director of CARES, said of an exchange with a parent whose child was diagnosed with the disease. "And I realized at that point what an amazing organization this is, and the amount of support we are able to provide these patients."

As the leading organization in the CAH community, CARES works to improve the lives of those with the disease by guiding parents to valuable information and doctors, while also leading advocacy efforts to fight misconceptions against the disease.

The most notorious of those misconceptions has to do with another effect of CAH on the body: CAH also manifests itself with elevated levels of testosterone during gestation, which can result in female infants being born with atypical genitalia.

These physical effects of the disease have caused many people to place affected women alongside the transgender and intersex communities in public discourse, despite the fact that women with CAH are internally female and can reproduce effectively.

According to Dr. Karen Lin Su, who serves as the director of CARES, most women affected by CAH still choose to identify as women throughout their lives.

"It is not correct," Su said of the links between CAH and the transgender community. "I mean, again, there's a very, very small percentage who probably are transgender, but it's not a large number."

In addition to battling the unfounded links between CAH and gender identity, CARES also works to take the stigma away from surgery performed on CAH-affected children, which has drawn unsavory comparisons to genital mutilation.

CARES works to guide parents toward specialized doctors and surgeons, and provide them with a platform for making decisions on their child's health.

"CARES does not take a position on surgery, for or against," Matos said. "Our job is really to help people make informed decisions, and there's a whole section on our website about that. But most importantly, our goal is to make sure that these parents make a decision by consulting with highly specialized urologists, surgeons who have performed surgeries."



Dina Matos said that her organization focuses on providing parents with the best information and resources possible, rather than pushing them toward a specific decision with regards to surgery. (Photo: ~Courtesy of CARES Foundation)

Matos and Su repeatedly emphasized that CAH surgery, if performed, is not merely a cosmetic decision, and said that the recommended time frame for surgery is at the age of 6 to 9 months.

"If they choose to wait — you know when a child is verbal — then they start asking questions, other people start asking questions," Matos said about the choice to perform surgery on a child. "And that does shape how a child's personality forms. And it will probably shape the way they interact with other children, with adults."

CARES, whose programs benefit 3,600 families in the United States and abroad, was originally an advocate for newborn screening of CAH, which has become increasingly common for the disease.

According to Su, the procedure is especially important for boys, who don't show the physical signs of the disease that girls do (increased testosterone levels don't have a noticeable effect on their bodies) but are no less vulnerable to its potentially fatal effects.

However, while CARES has made significant strides in raising awareness of CAH, there is still much work to be done to normalize the disease.

According to Matos and Su, routine activities such as going to the bathroom, going on a field trip or even allowing one's child to be babysat become far trickier with society's lack of knowledge of the disease.

"School nurses, school administrators, even teachers and coaches, are trained on how to administer epinephrine in an epipen, but that's not done for CAH patients," Matos said. "So we advocate for those patients, because they shouldn't be treated any differently than another patient."

While CARES doesn't expect to develop a widespread understanding of the disease overnight, they'll continue to educate parents of children affected with the disease while serving as a de facto CAH community for affected families.

"A doctor, a pediatrician, a psychologist, another parent, another child as they get older that they can talk to," Matos said. "It's important that they have that type of support."

To learn more about the CARES foundation and CAH, visit <https://www.caresfoundation.org/>.