

# Parents Sponsor Fundraiser To Fight Rare Disease

By Judy O'Gorman Alvarez

HALEY AND AMBER Le Vine of Avon suffer from a rare, debilitating disease. But there are no colorful wristbands, walkathons or celebrity appeals to raise money for research to find a cure for SSADH, the rare metabolic neurotransmitter disease these sisters suffer from.

Succinic Semialdehyde Dehydrogenase Deficiency (SSADH) has serious effects on memory and speech and is so rare that the sisters, now 20 and 22 years old, were the fifth and sixth people to be diagnosed in the U.S. with the disease.

As a result, Jess and Karen Le Vine have taken on the role of advocates for their daughters and others families whose lives are rocked by SSADH.

For the first time, the Le Vines, including two older daughters, Jacqui and Allison, are hosting a Wave of Hope Holiday Benefit for Amber and Haley on Thursday, December 29, from 7 to 11 p.m., at the Headliner, in Neptune. The evening will feature entertainment by the Pat Roddy Band and Paul Cilinski, discounted beverages and light fare (provided by McLoones and The Atlantic Club). Tickets are \$25.

Amber was seven when she and younger sister Haley were accurately diagnosed. Before that, the family had spent seven years and visited five major hospitals until specialists at Johns Hopkins Hospital in Baltimore identified their condition in 1996. Since then, some 350 patients worldwide have been diagnosed with SSADH.

The Le Vines have joined forces with other SSADH families – in Virginia and Michigan and elsewhere – raising awareness and holding fundraisers, but the results are painfully slow. "Because it is such a small cohort, it has been us against the world," says Le Vine.

SSADH families fall under the umbrella of the PND (Pediatric Neurotransmitter Disease) Association, which has been instrumental in assisting families with conferences, grants and other efforts to combat the disease, but the Le Vines believe a separate organization would help promote awareness of SSADH. As a result, Le Vine is launching a website, [www.center-forssadh.org](http://www.center-forssadh.org), and is reaching out to local and national media, trying to draw attention to SSADH and unite families affected by it. "We want to get the word out in a public sense," he says. "It's not in the public eye like autism or MS," he says. Patients are too few and far between to generate much interest.

Right now a drug, SGS 742, may hold the answer and provide some relief. Thanks to the National Institute of Health and a grant obtained through the PND Association, a clinical trial is progress and the Le Vine sisters await the chance to participate.

Many patients afflicted with diseases and conditions other than SSADH suffer similar symptoms such as gross motor problems, memory loss, and seizures. A newly discovered drug could help them as well.

After false starts, Le Vine says pharmaceutical companies have "played football with the rights to produce the drug." He has grown frustrated and hopes to bring his daughters' plight and others suffering from SSADH to the forefront and perhaps nudge pharmaceutical companies to put more interest in researching and developing medicines.

The Le Vine sisters, who have lived with SSADH for 15 years, are of special interest to families with recently diagnosed young children who look to the girls to see how their own children may fare in later years.

Amber and Haley are bright, vibrant young women who enjoy playing music and bike riding but struggle with speech, gross motor skills and serious memory problems.

Haley attends the Hawkswood School for students with special needs, where Amber graduated. Amber now takes classes at Brookdale Community College, where her father is a history professor. She also works bussing tables at a restaurant. "In our case, memory and speech are the hardest parts," says Le Vine. "It's bizarre. Amber may not remember what I told her yesterday but she can recall something from three years ago. The tragedy [with the disease] is you're trapped in a weird sense of time and not remembering."

With the website and center, the Le Vines hope to create awareness, generate an interest for donations and urge drug companies to push for research and answers. And perhaps spark creative minds, inside and out of the scientific community, to take up the cause. "Someone may say, 'I want to beat that,'" says Le Vine. "You just never know."

For more information on the benefit visit [www.waveofhope-ssadh.org](http://www.waveofhope-ssadh.org).