

RANCHO CANYON NEWS

AN EDITION OF THE
REGISTER

FRIDAY, FEB. 28, 2014

OCREGISTER.COM/RSM and OCREGISTER.COM/CANYONS

Fashion show to put rare disease in spotlight

BY SARAH DE CRESCENZO
STAFF WRITER

LAKE FOREST • Today is World Rare Disease Day, but if the Spooner family had its way, that day wouldn't exist.

The annual event, which started in 2008, is intended to draw attention – and research funds – to thousands of rare diseases.

Cristy and Rick Spooner's oldest and youngest daughters, Cali, 15, and Ryann, 4, have a rare genetic disease, diagnosed in 2013. After years of confusion over the cause of the girls' challenges, a test called exome sequencing determined they had inherited mutations on a gene known as NUBPL, which prevent their bodies from producing the energy their cells need to function properly.

With the disease identified, UC Irvine geneticist



COURTESY OF THE SPOONER FAMILY

Parents Rick and Cristy Spooner sit with their daughters, from left, Cali, 15, Ryann, 4, and Raelyn, 8.

and pediatrician Virginia Kimonis has proposed further research, with the ultimate goal of finding a cure. To fund the project would cost \$250,000, she said.

When she told the Spooners the price tag, "we knew we had to do some fundraising," Cristy Spooner said.

Inspired by a girl with ce-

rebral palsy who founded a pageant for girls with disabilities, the Spooners invited local children with special needs to be in a fashion show, set for Sunday at Peppino's Italian Family Restaurant in Foothill Ranch.

The event is free; any donations will go to the Spooner Fund, set up

How to help

To donate to the Spooner Fund, visit ua-web.uadv.uci.edu/eGiving.

through UCI to fund the research proposal.

"The Spooners are doing the right thing," Kimonis said. "I cannot raise money from the (National Institutes of Health) for one family, even a dozen families. You have to start somewhere, and I'm hoping that the preliminary data will lead to funding down the road."

For more information, visit [facebook.com/spoonerfund](https://www.facebook.com/spoonerfund) and spoonerbrain.blogspot.com.

CONTACT THE WRITER:

949-454-7307 or sdecrescenzo@ocregister.com