Teamwork and genetics uncover extremely rare diseases and patients who share them

In February 2013, Stanford geneticist Gregory Enns called the Wilsey family with news: He was “99 percent sure” that doctors had found the cause of their 3-year-old daughter’s terrible congenital disease.

“That was a huge relief,” said Kristen Wilsey, whose daughter, Grace, is now 4. “Other people may not understand how it can be such a relief to be told that your daughter has a very rare genetic disease for which there is no cure or treatment. But when you don’t know what you’re fighting, you have no direction. It meant we could start moving forward.”

Grace is a patient in a research paper published last month about the newly discovered disease, called “NGLY1 deficiency” for now; the gene mutation that all the patients share is in the NGLY1 gene. Eight patients have been confirmed with the disorder so far, but doctors believe they’ve found at least another half dozen who haven’t yet been formally diagnosed.

Read the full, original story: Families, doctors team up to solve mystery of missing enzyme