



**Greg Cooper is a faculty investigator at HudsonAlpha. (HudsonAlpha)**

Sequencing Exploratory Research (CSER) project, which is helping end a diagnostic odyssey for many families.

Some conditions, such as Down syndrome, have an obvious genetic cause. But there are other overlapping symptoms and disabilities that also have a genetic origin, and they are not as easy to pinpoint.

"With genetics, we're hoping to go after those underlying causes," Cooper said. "A major advancement has been our ability to go from looking at whole chromosomes to being able to look at every bit of those chromosomes. CSER looks at whole genome sequencing to look at every piece of a child's genetic code."

And while the diagnoses don't come with a cure-all for the children, they do offer direction on medication, therapies and support groups. Parents say that simply having an answer is invaluable.

David and Miranda Ainsworth of Florence spent eight years going to countless doctor appointments searching for a diagnosis for their daughter, Anna Brooke. In February, she was diagnosed through the CSER project with **Cornelia de Lange syndrome** (CdLS), a rare genetic developmental disorder.

"This has been such a long journey for us, and we were just thrilled to have an answer," Miranda Ainsworth said. "We can deal with whatever, but to get an answer that puts it all together, this is what we needed. We needed that for our family."

CSER is funded by a **National Institutes of Health** grant, and Phase 1 will end next year. But HudsonAlpha is applying for Phase 2 funding to continue the work. Most of the children involved are from North Alabama, but some hailed from other parts of the state and surrounding states. More than 100 children, and counting, have been diagnosed.

In a similar effort, the institute is partnering with **Alabama's Children's Rehabilitation Service** and recruiting families from South Alabama to participate.



**Anna Brooke Ainsworth of Florence was diagnosed with **Cornelia de Lange syndrome (CDL)**, a rare genetic developmental disorder, through a HudsonAlpha project. (HudsonAlpha)**