Figuring out Anna Brooke with new genetic testing in Alabama

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"We just knew in our hearts there was something else going on," Miranda Ainsworth of Florence said Wednesday.

Ainsworth was talking about 9-year-old daughter Anna Brooke and the developmental delays she'd spotted as early as 5 months old.

A long series of tests – even a full genetic screening – led to a series of diagnoses "thrown at us," she said, including microencephaly, cerebral palsy, even gastro-intestinal problems. None was right; all left Ainsworth determined to keep searching.

Ainsworth and her husband, David, came to Huntsville Wednesday to celebrate the end of their search with researchers at the HudsonAlpha Institute for Biotechnology. The institute was marking the 100th child diagnosed in a partnership with the University of Alabama in Birmingham, and one of those 100 children was Anna Brooke.

'A great new idea'

HudsonAlpha started the screenings by approaching UAB neurology professor Dr. Martina Bebin. Bebin has more than 1,700 children as patients at clinics in Alabama, and HudsonAlpha wanted to team with her in new research.

"They had a great idea," Bebin said Wednesday. "This technology has advanced so quickly over the last two years."

The National Institutes for Health said "yes" to funding, and free whole genome screenings began using new technology. They continue into next year, and parents can still get in on the program. **They can contact HudsonAlpha** and ask about the CSER program (Clinical Sequencing Exploratory Research project) or reach out to Dr. Bebin through UAB, Children's Hospital of Alabama, or North Alabama Children's Specialists in Huntsville. (Watch a video about the project below.)

New screening "opens the door to diagnosing earlier and really being on the prevention side of some of these diseases patients face," Bebin said. "Preventive medicine, or preventive health maintenance, as I like to look at it, is such a better position to be than on the reactive side."

Dr. Greg Cooper of HudsonAlpha leads the team that does the genome sequencing and analysis using new rapid scanners. "What we often find," he said, "is ... a cause rooted in their DNA that contributes to the challenges they may be dealing with."

The final diagnosis

For Anna Brooke Ainsworth, the final diagnosis was Cornelia de Lange Syndrome, a very rare genetic disorder present from birth but not always found then. There will be challenges as she grows up, Miranda Ainsworth said, including the discussion to come about the 50 percent chance Anna Brooke could pass the syndrome to her own children.

"We can deal with whatever (comes)," Ainsworth said. And she had advice for other Alabama mothers wondering about their children.

"I would say, 'Do not stop,'" she said. "We have that mother's intuition. We know when something else is really going on.... We have had so many things thrown at us, but we needed something to pull it together. And that's what genetic testing has done for us."