Genetic tests help in diagnosing babies with unknown ailments

Within hours of entering the world, little Sebastiana Manuel's entire body froze in a rigid spasm. Her neck twisted, her face turned blue, and one side of her body stiffened as if someone were yanking her violently.

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Dr. Stephen Kingsmore launched a genomics institute at [Rady Children's Hospital] designed to help babies like her. From any infant younger than 4 months who has a sickness that can't be explained, Kingsmore's team takes a vial of blood to run a genetic test. Within days, they sequence the entire genome of the baby.

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Sebastiana's DNA told him that an antiseizure drug different from those that doctors normally use–one that is rarely used in infants–would be more effective at treating her seizures. […]Once he made the switch, she became more alert, responding to her parents and eating as any healthy infant would. And her seizures stopped.

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If Kingsmore gets his way, mapping the DNA of these babies will one day be as standard as ordering a blood test. These infants often carry the answer to their own mystery illnesses in their very DNA; it's just a matter of recognizing and reading the genetic clues.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Genetic Testing Is Providing New Hope for Babies Born with Mysterious Ailments