

# Childhood Genetic Disorders



Two out of every 100 children are born with a developmental, physical, or intellectual disability, many of which have a genetic origin. Oftentimes, physicians cannot make a diagnosis or identify the cause of a child's condition, leaving parents with painful questions and uncertainty about their child's future. Through genomic sequencing and analysis, HudsonAlpha is able to give the benefits of its ground-breaking research to families who desperately need answers.



**Developmental disabilities and delays are a group of conditions due to an impairment in physical, learning, language, or behavior areas. Often, these conditions begin before birth and may impact day-to-day functioning, and usually last throughout a person's lifetime.**

## Model for discovery

HudsonAlpha researchers have created a model for using the information in human genomes to identify the genetic causes of developmental delay and unexplained neurological conditions in children. Our goal is to make discoveries that allow physicians to provide a diagnosis to patients and their families.

These children have often lived for years with an undiagnosed or misdiagnosed condition, and those who love them have watched them endure tests that provided no answers. To help end that diagnostic odyssey, HudsonAlpha geneticists and bioinformatics experts sequence and analyze the entire genomes of children and their parents – groups known as “trios”. HudsonAlpha genetic counselors then explain findings from the analysis and what those findings mean to patients and their families.

The team of scientists and physicians associated with the research project have delivered answers to hundreds of children and their families, providing life-changing hope and access to treatments and resources that are best for the child's specific condition.

## Newborns in the NICU

In an effort to help more patients and families earlier in the diagnostic journey, HudsonAlpha researchers are sequencing neonatal intensive care unit (NICU) babies across the south through a program called *SouthSeq*. The project provides genome sequencing and analysis with the hope to provide patients with earlier diagnosis and a pathway for better care and treatments.

Genomic information can reveal the underpinnings of diseases to give diagnostic information that other medical technologies may miss. HudsonAlpha utilizes the best DNA sequencing technology to produce and analyze genomic data, along with a team of highly-experienced clinical analysts to draw meaningful conclusions that impact patient care.



**1 in 6** children between the ages of 3 and 17 have one or more developmental disabilities.



**40%** of children with special needs do not have an exact diagnosis.



**\$268B in 2015** the cost of caring for Americans with autism. This could rise to \$461B by 2025.



**4 States** - HudsonAlpha's *SouthSeq* project is working with medical facilities in Alabama, Mississippi, Louisiana and Kentucky.



**More than 1,100 patients** sequenced, along with more than 1,500 family members.



**30%** of families enrolled in HudsonAlpha's projects have received a diagnosis, ending their search for answers.



Research leading to the discovery of **more than a dozen** previously unknown genetic conditions, knowledge of which allows labs all across the world to improve their testing results.