INVITAE

Support H.R. 5989 *Precision Medicine Answers for Kids Today* Act to enable faster, more accurate diagnosis of pediatric diseases

This bill removes barriers and enables physicians to select the genetic test that is best for patients and their families.

Genetic testing transforms the way children are diagnosed

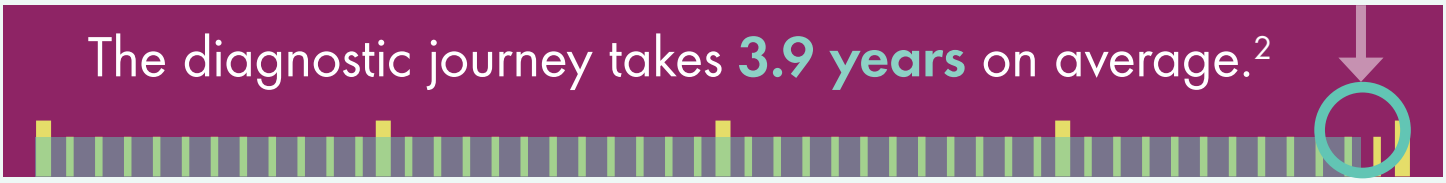
Early genetic testing for children with rare diseases can lead to faster diagnosis, improved outcomes and lower financial burden.

Rare diseases are costly—and common

Rare diseases affect up to **30 million people** in the United States alone, half of them children.[1]

On average, patients with a rare disease undergo up to **14** **non-genetic diagnostic tests** and receive up to **5** **misdiagnoses** before they finally find answers.[2]

The diagnostic journey takes **3.9 years** on average.[2]



That's 46 months of doctor's visits, tests, and medical bills which is similar to the amount of time it took to diagnose patients with a rare disease 30 years ago.[3]

While searching for a diagnosis,

60% **of rare disease patients** incur an unexpected financial burden,

leaving **29%** **in debt.**[2]

Fast, accurate diagnosis through genetic testing

Genetic testing can dramatically shorten the search for answers for many patients, leading them more quickly to an accurate diagnosis.

Approximately 16% of pediatric patients admitted to the hospital have a genetic condition, meaning that something in their DNA is contributing to their illness.[4]

For example, when a child presents with seizures, genetic testing can help identify one of the more than 100 underlying genetic causes, shortening the time to diagnosis and helping doctors tailor care to a patient's specific needs.[5]

Participants in a sponsored genetic testing program for pediatric epilepsy received an accurate diagnosis 1–2 years sooner than historic averages.[6]

Most physicians agree that genetic testing shortens the diagnostic journey for rare diseases.[2]

In many cases, an accurate diagnosis enables doctors to quickly identify the treatment or therapy most likely to succeed, leading to improved outcomes by connecting patients to successful treatments earlier in disease progression. In a 2020 study, 81% of physicians reported **adjusting treatment within 3 months** of receiving a genetic test result. In the same study, after receiving a positive genetic result for epilepsy, half of physicians changed their patients' treatment plan.

Of patients who received different treatment and subsequent follow-up visit,

75% reported improved outcomes,

with **65%** reporting reduction or elimination of their seizures.[7]

A solution: High-quality, low cost testing

Genetic testing has historically been seen as too expensive (and reimbursement from insurance companies unreliable), causing mainstream healthcare providers to sometimes shy away from ordering it.

Yet the paradigm is changing. Technology has evolved such that it's now possible to offer genetic testing that is both accurate and affordable. One genetic information company, Invitae, is in-network for more than 300 million Americans and also offers a **$250 self-pay price** for diagnostic testing.

That type of affordable and accessible genetic testing can enable many children, their families, and their doctors to receive timely information about the child's rare disease and intervene early, when treatments can be most effective.

The cost of sequencing the human genome has fallen by a factor of 10,000 over a decade, and continues to fall today.[8] Advances in sequencing technology allow physicians to use the most appropriate diagnostic sequencing strategy for each patient depending on what is most cost-effective and clinically appropriate.

$250 self pay price

Patient spotlight: Naya Edouard

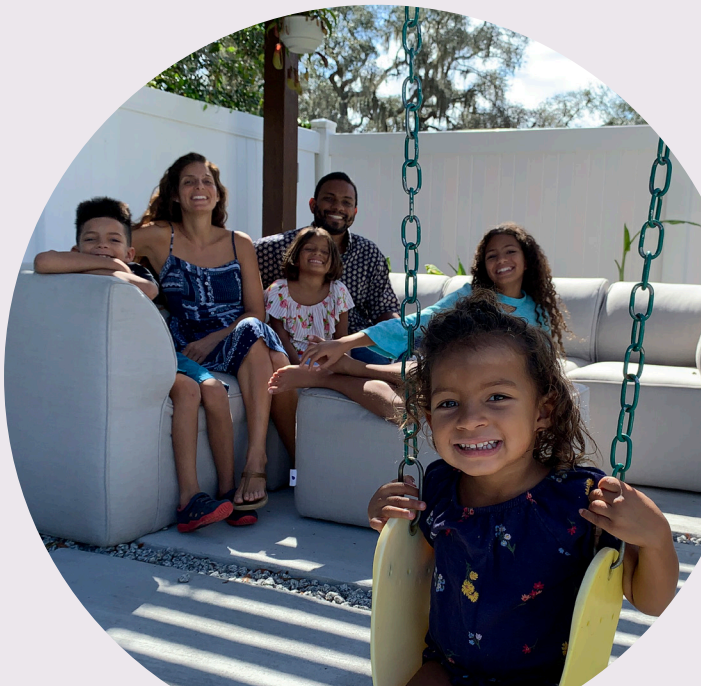
At 18 months old, when Naya Edouard had only recently started sitting on her own, hadn't started to talk, and occasionally experienced brief muscle jerks almost like a newborn's startle reflex, her medical team suggested genetic testing to figure out what was wrong.

"But we just couldn't get the genetic testing done," said Naya's mother, Alexis. The family was stuck in a catch-22: the hospital required an insurance pre-approval, something the insurance company didn't actually offer. Alexis spent months trying to bridge the gap, but neither institution would budge—it was against policy.

Finally, 10 months later, Naya was connected to a sponsored testing program that offers genetic testing for children who have experienced an unprovoked seizure. Just a few weeks later, Naya had her genetic results—and an important diagnosis: SynGAP1 disorder.

"Before that, every time we'd see another specialist, they'd tell us that Naya would eventually catch up," Alexis said. But the diagnosis showed that wouldn't be the case. "Now, instead of trying to figure out what we're doing wrong as parents, we can move forward."

Although there's no cure for Naya's rare neurodevelopmental disorder, a diagnosis can inform medication choices, and several therapies are under development that could be available to patients in a few years.



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