

## **FAQs on The Ending the Diagnostic Odyssey Act (S. 3116/H.R. 4144)**

### **What would the bill do?**

- The bill would allow states to conduct a three-year pilot program to increase the Federal Medical Assistance Percentage rate (FMAP) to provide Whole Genome Sequencing (WGS) services for children on Medicaid with a disease suspected to have a genetic cause, and it includes strict reporting requirements.

### **Who supports the bill?**

- The bill has been endorsed by 100+ rare disease groups and children's hospitals, the Genetic Alliance, Biocom, and the Personalized Medicine Coalition.

### **How many rare genetic diseases are there?**

- There are more than 7000 genetic conditions we know of today.<sup>i</sup>

### **How many families are affected by these genetic diseases? How many children?**

- 350 million people worldwide, 6-10 million in the U.S, and roughly half of these are children.<sup>ii</sup> The individual genetic diseases themselves are "rare," but the collective population is quite large.
- For comparison, worldwide, roughly 37 million people are affected by HIV<sup>iii</sup>, and 219 million by Malaria.<sup>iv</sup>

### **How long does the average family struggle through a battery of other tests before they receive a diagnosis?**

- The average Diagnostic Odyssey takes 5-7 years.<sup>v</sup>

### **What is Whole Genome Sequencing? How can Whole Genome Sequencing (WGS) help these families?**

- WGS is a comprehensive method for analyzing the genome. WGS provides a high-resolution view of the genome and delivers large volumes of data in a short amount of time.<sup>vi</sup>
- WGS looks across the genome and can be used to detect variants. By looking at the whole genome, we can better understand the genetic origins of disease, and often, which treatments are most likely to be effective.

### **How does Whole Genome Sequencing compare to other tests?**

- There are single gene tests, and panel tests which test for approximately 50-150 genes, and finally, Whole Exome Sequencing (WES), the second most comprehensive test, which tests only 2% of the entire genome.
- WGS is the most robust genetic test available, and the only test that can detect nearly all types of genetic variants.<sup>vii</sup> The likelihood of diagnosis with WGS (55-70%) is roughly twice that of WES (24-33%), and roughly 3x greater than panels.<sup>viii</sup> It also produces results faster than WES when accounting for preparation and data interpretation; this time-savings is closing what little gap remains in terms of cost-savings between WGS and WES.<sup>ix</sup>

### **Who will be doing the testing?**

- Wherever the clinician usually orders tests – including medical centers, private labs, state labs. This will be decided by the clinician and his/her employer.

### **What is the cost of such testing?**

- CMS estimates this to be about \$5,000 per newborn. This includes the preparation, testing, and back-end analysis.

### **Will this be used in the prenatal period?**

- No. This is only for newborns where the clinician suspects a genetic disorder.

### **What does a timely, accurate diagnosis mean for families struggling with a genetic disease?**

- Once a diagnosis is in hand, physicians will often make a change in treatment or management<sup>x</sup>, and as more cases are discovered, the urgency and demand for new treatments and clinical trials grows. In terms of cost, an earlier diagnosis can mean hundreds of thousands of dollars in savings.<sup>xi</sup>
  - In a recent study, Rady Children's Institute for Genomic Medicine showed that by sequencing a group of 42 infants, the recovered savings from sequencing was far greater than the cost of sequencing for the entire group. Sequencing diagnosed 14 more patients than standard care would have (33%), and changed the medical care from what the standard of care had recommended for 11 patients (26%) – resulting in savings across the cohort of approximately \$100,000 to \$1.1 million.<sup>xii</sup>
  - A separate study estimated that the average cost of the tests alone prior to receiving a Next Generation Sequencing-based diagnosis was \$19,100, and that total average cost of care was \$305,428.<sup>xiii</sup>
- In addition, there are psychological benefits that come from the peace of mind of simply knowing what is affecting your child, and finding other families dealing with the same struggle.<sup>xiv</sup>

## How many people have access to WGS today?

- Today, only six states (KY, MN, NJ, ND, OH & IL) explicitly provide for clinical WGS for Medicaid patients. Combined with commercial coverage from two insurers, this means only 14 million total people are eligible for the most comprehensive diagnostic, and even then, only in very limited circumstances.<sup>xv</sup>
- Compare this to WES which Medicaid covers in 20 states. Combined with greater coverage from commercial payers, 152 million people already have access to this less comprehensive test.<sup>xvi</sup>
- A clear signal and incentive from the federal government would drive clinical adoption in and out of the Medicaid setting at the beginning of a child's struggle, providing a quicker path to diagnosis and treatment.

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## Citations

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- <sup>xv</sup> Policy Reporter. [Policyreporter.com](http://Policyreporter.com)
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