

THE DIAGNOSTIC ODYSSEY

What Genetics Providers Need to Know



THE DIAGNOSTIC ODYSSEY

- Approximately **15 MILLION** children in the United States have an undiagnosed condition, with the vast majority having a genetic basis.
- The average time from onset of symptoms to an accurate diagnosis is almost **5 years**.
- Parents are learning how to care for a child with health issues while facing **uncertain prognosis**.
- Approximately **40% of parents** of undiagnosed children experience high rates of anxiety and depression.

BARRIERS TO GETTING DIAGNOSIS



WORKFORCE SHORTAGE

Only **1 in 100,000** Americans has access to a clinical genetic counselor.

Genetic tests are **ordered by Pediatricians** who also explain testing results.

50% of pediatricians report not feeling competent to provide care related to genetics and genomics.

Pediatricians report **difficulties in developing relationships** with genetic specialists to fill in the gaps in their own genetic literacy.



RACIAL BIAS

Use of race to diagnose genetic conditions has contributed to the **underdiagnosis of cystic fibrosis in African Americans** because cystic fibrosis is considered a “white” disease.

Due to historical events, **minority families are not as trusting about genetic testing** compared to white families.

Mistrust within the healthcare system among minorities is compounded by **underrepresentation of diverse healthcare providers** in the workforce.

The “historical legacy of mistreatment” by medical professionals continues to **manifest as mistrust** between African Americans and their doctors.



INSURANCE PROBLEMS

Insurance does not always cover needed tests, which places the **financial burden on families**.

BY THE NUMBERS:

Individuals with a Genetic or Inherited Condition

- 14%** live in households where the primary language is not English
- >50%** live in households where the income is <199% of the Federal Poverty Level
- 26%** live in households where an adult has some college or technical school
- 20%** of adults have a high school diploma or GED

More data can be found at www.childhealthdata.org

IMPACT OF THE DIAGNOSTIC ODYSSEY



Psychological, emotional, and financial stress affecting access to services



Distressing and traumatic tests for extended periods of time



Delayed care



HELPING FAMILIES NAVIGATE OBTAINING A DIAGNOSIS

- VALIDATE THEIR CONCERNS
- PROVIDE A PROGNOSIS
- REFER THEM TO PEER SUPPORT OR ADVOCACY GROUPS (www.familyvoices.org, www.p2pusa.org)
- BE PREPARED TO PROVIDE INFORMATION TO COMMON PATIENT QUESTIONS SUCH AS:

- What are common diagnoses that might explain my child’s symptoms and responses?
- How well does my child fit those profiles?
- Are there other specialists with whom we should consult?
- Should we consider a rare disease?
- Should we get a genetic test?
- Where can we access more information about conditions that might explain these symptoms?

FOR MORE INFORMATION, VISIT WWW.EXPECTINGHEALTH.ORG/PROGRAMS/FAMILYCENTER

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National Genetics Education and Family Support Center

