

# Genetic Testing & Counseling for Rare Hereditary Diseases



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## **Genetic testing and counseling are useful tools for patients with rare diseases.**

Sophisticated genetic tests available in clinical settings now offer health care providers a closer look into patients' genes. The information there may allow them to diagnose a hereditary condition, assess a patient's risk for a particular disease or determine whether a certain drug might work.<sup>1</sup> Since the mapping of the human genome in 2003, more than 75,000 genetic tests have emerged, with about 10 new tests entering the market daily.<sup>2</sup>

Testing is typically complemented by guidance from a genetic counselor, who serves an internationally recognized role. These essential practitioners have traditional counseling skills as well as a specific education in assessing the nuances of genetics and genomic medicine.<sup>3</sup> Genetic counseling is an important tool in helping patients prepare for, interpret and make decisions based on testing results.

Genetic testing and counseling can help providers and patients make informed choices throughout a patient's course of treatment. Furthermore, they can accelerate the decision-making process, which is critical for patients with rare diseases that have poor life-expectancy rates.

Despite this valuable resource, accessing tests and the accompanying counseling services can be difficult, and the results may impact a patient's life and family in unexpected ways. How can the policies that shape genetic testing empower these patients?

## The Challenge of Diagnosing Rare Diseases

Diagnosing a rare disease is challenging, in part because so few general practitioners have direct experience with it.

Consider the example of hereditary transthyretin, or hATTR, amyloidosis. The disease affects approximately 50,000 people worldwide and can impact multiple parts of the body, including the nerves, heart and digestive system.<sup>4</sup> It is caused by build-up of an abnormal protein in the organs, but the symptoms can mimic other more common diseases or appear completely unrelated. Different variations of the disease have different causes and symptoms, often leading patients to see an array of providers. This makes it harder to identify the disease.<sup>5</sup>

Diagnosing a rare disease can take, on average, up to five years.<sup>6</sup> That can be a deadly delay for amyloidosis patients, whose life expectancy without treatment is as little as two years.<sup>7</sup>

Furthermore, there are likely fewer specialists for any disease that is especially rare. A patient may need to travel a great distance to see the right provider or wait months for an appointment.


This is why genetic testing is important. There has been a push in the genomic medicine community recently to encourage genetic testing as soon as possible. Results can save a patient precious time, and providers could potentially inform vulnerable family members sooner.

## Privacy Concerns with Genetic Testing

Not every patient, however, is comfortable with the idea of testing.

Genetic information is rightly perceived as among the most highly sensitive personal data. The possibility of consequences for employment, increased life insurance premiums or even social stigma weigh heavily on many patients' minds.

The Genetic Information and Nondiscrimination Act of 2008 protects the genetic privacy of the public. The law prohibits most employers or health insurers from requiring genetic information, and it forbids the discriminatory use of such information. But concerns remain about forms of insurance not covered by the law, such as life, long-term care and disability.<sup>8</sup> With these forms of insurance, information revealed by genetic testing could still seriously impact patients' eligibility, coverage or premiums.



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## Cost Barriers

Some patients must also weigh the potential benefits of testing against the financial costs.

Genetic testing is not always covered by insurance and can be expensive. Insurers may require prior authorization or impose other coverage barriers for necessary diagnostic tests, creating an increased burden on an already over-taxed health care system. Even worse, insurers and even Medicare may not be entirely up front on what they will cover and what they won't.

That may, in part, be because insurers face significant procedural barriers when it comes to genetic tests. The tests are billed using a standardized system of codes developed by the American Medical Association, and fewer than 200 codes exist for the tens of thousands of genetic tests.<sup>9</sup>

Patients have the option to pay out of pocket, and prices have declined in recent years to as low as a few hundred dollars. But that may still be cost prohibitive for some patients.

The many hurdles and delays in accessing testing can be difficult for patients and their families. Some pharmaceutical companies today partner with laboratories to offer free genetic testing for patients, which has greatly helped to ease the stress of the genetic testing process. In addition, some labs may offer discounts of their own on genetic testing.

## Easing Patient Hesitancy Through Genetic Counseling

Given the range of considerations patients must navigate, it may help to involve a genetic counselor even prior to testing. Genetic counselors are trained to present complex information about genetic risks, testing and diagnosis to patients. Together, the counselor and patient, in consultation with the specialty clinician, can explore privacy and cost concerns and find solutions to move forward.

Genetic counselors can also help patients cope with any preliminary anxiety, manage expectations throughout the testing process and course of treatment, and act as the point person to communicate and understand results. Educational tools such as papers, pamphlets and infographics can help patients understand their results and their options.

Once test results have been returned, a follow-up counseling session typically involves more than simply relaying the medical information. Genetic counseling aids patients in processing the emotions of receiving a genetic diagnosis, identifying support groups, offering educational resources and connecting newly diagnosed patients with fellow patients.



## Acting on Test Results

Combined with health history and details of a patient exam, diagnostic results can help the specialty clinician chart the path forward for an individual patient. Next steps, however, often present ethical and logistical questions that are difficult to answer.

When testing indicates an inherited mutation, clinicians must weigh their responsibility in getting the patient's relatives tested. Simply asking the patient to contact relatives may not be adequate. The patient may be distraught, reluctant or unsure how to communicate a positive test result to family members.

Should providers send free testing kits to relatives, even if those relatives are not their direct patients? This might be the most effective means, but it may not be practical or standard, and many providers worry about legal issues that may arise from testing patients who are not their own.

Some providers give their patients a family letter that can be customized, then mailed or copied and pasted into an electronic message. It explains the risk they face and provides instructions about how they can get tested.

Sometimes patients and providers face resistance from family members about testing.

With hATTR amyloidosis or Huntington's disease, another progressive and inherited condition, family members may avoid testing because they don't want to face the severity of the diagnosis. With Huntington's disease, family members may also opt not to test because treatment options are limited.

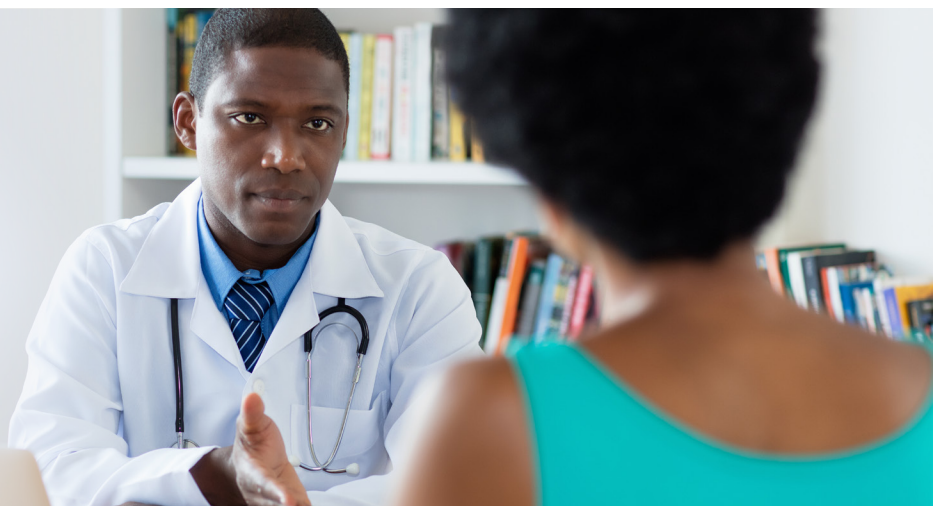
## The Value of Answers

Ethical and logistical challenges notwithstanding, test results can be empowering to patients and providers alike.

Even when testing reveals that a patient has a hereditary condition, that knowledge can end years of uncertainty. It can also allow the patient to begin treatment. Some rare diseases, including hATTR amyloidosis, have much better outlooks than they once did. After years with no treatment options, patients with amyloidosis now have three FDA-approved medications and several other new medications on the horizon. Meanwhile, researchers are investigating preventive treatment that could help patients with genetic conditions before they develop any signs or symptoms of disease.

Ultimately, knowledge is always beneficial if privacy can be reasonably assured.

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## Conclusion

Genetic testing and counseling empower patients to seek treatment and offer clinicians a roadmap to identify the best course of treatment for their patients.

While some patients are hesitant to seek out these services for privacy or cost concerns, it is essential to diagnose rare diseases like amyloidosis because outcomes can be improved with FDA-approved treatments.

Continued education and awareness, along with improved access, can help patients and health care providers make the most of genetic testing. Policymakers can and should address the loopholes that allow discrimination to persist in the life, long-term care, and disability insurance industries. They can also work to improve coverage for genetic testing and counseling, which can inform and empower people with rare diseases to seek treatment and maximize quality of life.

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