



InformedDNA®

Genetics, Decoded.

Bringing the Power of Genetics to a Rare Disease Gene Therapy Program: A Case Study

Background

With the arrival of gene-based therapies, biopharmaceutical companies face significant challenges in incorporating genetics expertise into programs supporting clinical trials and emerging treatments. This is especially true for therapies that target rare diseases, where physician awareness is limited and patients are difficult to identify. By incorporating genetics expertise into clinical trial programs, sponsors can increase patient identification and recruitment efficiency, improve patient retention, and arm physicians with the genetics expertise they need to successfully participate as enrollment sites.

The Challenges

Our client, a gene therapy company, faced challenges in finding patients with the rare diseases targeted by their experimental therapy. They were further challenged to provide physicians the expertise and support needed to help their patients benefit from participation in a genetic testing screening program. Here are some reasons why:

- Pharmaceutical companies have difficulty recruiting genetics experts with specific rare disease expertise.
- Although physicians are eager to order genetic testing for their patients, many feel ill-equipped to interpret and communicate genetic test results.

- Appropriate rare disease clinical trial and therapy candidates are difficult to find, due to a lack of physician awareness and a typically low percentage of patients with the targeted diseases that have undergone genetic testing.

The Solutions

Genetics Program Development

- **Genetic testing program design:** We worked closely with the client's medical services team to develop and implement a genetic testing program. Targeted physicians were eager to test their patients and determine who might benefit from upcoming therapies. We developed a screening questionnaire for physicians to help determine eligibility for the client's genetic test, resulting in a very high sensitivity for the detection of patients with this rare disease.
- **Targeted physician education:** InformedDNA's rare disease experts played a key role in educating physicians on genetic program elements by participating on multiple advisory boards and presenting at specialty-specific scientific conferences.
- **Test results interpretation:** For physicians who chose to disclose genetic test results to their patients personally, InformedDNA made genetic specialists available to serve as advisors, available for one-on-one consults to discuss and review complex test reports and provide guidance to physicians in advance of the physician's appointment with their patient.

Patient Engagement

- **Genetic testing panel design:** Our deep experience working with patients and physicians who treat rare diseases enabled us to propose a custom genetic testing panel ideal for our client's patient community, to maximize the molecular diagnostic rate while minimizing costs.
- **Patient recruitment:** We developed a screening questionnaire for physicians to use in determining eligibility for the genetic test, resulting in a very high sensitivity for the detection of patients with this rare disease.
- **Genetic counseling:** We provided telephone-based, post-test genetic counseling to patients participating in the genetic testing program. This helped to eliminate geographic barriers in reaching patients, and reduce screening costs by performing genetic testing locally. Physicians could choose whether to refer patients to InformedDNA for genetic counseling or disclose results themselves (with expert guidance from InformedDNA genetics specialists).

The Results

The solutions we deployed added genetics-oriented elements to the program design that enabled more successful patient recruitment and retention. Over a period of 18 months, more than 1,000 patients have received post-test genetic counseling and test interpretation through this program from InformedDNA's genetic counselors.



By partnering with InformedDNA to provide post-test genetic counseling, the client was also able to collect data related to patient motivation and outcomes that would otherwise have been unavailable to them. For example, in this program, over half of the patients stated that they chose to undergo genetic testing to determine possible eligibility for clinical trial participation or therapy.

The client reported nearly universal praise for the program from both patients and referring providers.

"Genetic counseling was a wonderful addition to the program. I'm a smart guy and read the results, but it really didn't mean anything to me until she [the genetic counselor] broke it down and really explained it. I thought that was a great offering."

— Client's genetic testing patient



InformedDNA® is the authority on the appropriate use of genetic testing. Leveraging the expertise of the largest full-time staff of independent board-certified genetics specialists, InformedDNA helps to ensure health plans, hospitals, employers, community clinicians, and patients all have access to the highest quality genetic services. The company's key offerings include clinical genetic counseling, genetic testing utilization management, clinical trial support, and personalized hereditary risk assessment and prevention solutions.

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