Part I of IV: Precision Medicine and Pharmacogenomics
New frontiers in genetic testing for employee benefits providers
February 2018
This paper is the first in a four-part series examining the role of genetic testing in employee benefits. The goal of this white paper series is to help employee benefits professionals understand this complex and evolving topic and to provide a framework for determining whether these new capabilities in medicine should be considered in crafting an employee benefits strategy. Genetic testing and precision medicine is a rapidly emerging and exciting area that will likely challenge the employee benefits industry for decades, so this white paper series will serve as a starting point to shedding light on the trend.
What is precision medicine?

Precision medicine, sometimes referred to as personalized medicine, is defined by the National Institutes of Health as “an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.” This approach allows physicians and researchers to predict more accurately which treatment and prevention strategies for a particular disease will work in which groups of people. It is in contrast to a one-size-fits-all approach, in which disease treatment and prevention strategies are developed for the average person, with less consideration for the differences between individuals. Because it is a tailored approach to medical care, it is a paradigm shift from the way medicine has been practiced and delivered for the last century.

HIERARCHY OF GENETIC TESTING

1. Genetic testing
2. Precision medicine
3. Pharmacogenomics
4. Cancer genomics

The emerging diagnostic capabilities of precision medicine and pharmacogenomics are just starting to gain the attention of benefits professionals. The near future will undoubtedly hold greater promise and significant challenges for how employers might leverage these new capabilities.
Precision medicine can do several things:

- Reveal genetic factors that increase disease risk.
- Inform optimal medication selection and dosage for improved outcomes and fewer side effects.
- Identify risk of inherited diseases.
- Offer insight into rare diseases.
- Individualize cancer care based on a tumor’s specific genetic fingerprint.

Pharmacogenomics is a subset of precision medicine and further defined as "the study of how genes affect a person’s response to drugs. This relatively new field combines pharmacology (the science of drugs) and genomics (the study of genes and their functions) to develop effective, safe medications and doses tailored to a person’s genetic makeup." Part II of this white paper series will explore pharmacogenomics in more depth.

### Precision medicine capabilities

Precision medicine has opened the doors to new potential in health management and care, so it is important for employee benefits administrators to understand the diagnostic capabilities. There are a number of advantages to having genetic testing done:

- More targeted prevention, early detection and treatment of disease.
- Encouraging healthier behavior in the covered population by having them take a more active role in their own healthcare.
- Extended quantity and quality of life.
- Avoidance of drug side effects and more effective therapies (pharmacogenomics).
- Discontinuation of useless drug therapies and toxic chemotherapies (pharmacogenomics).
Advances in precision medicine will help lead “doctors to use patients’ genetic and other molecular information as part of routine medical care.” Today, many healthcare providers are only just beginning to incorporate these capabilities in their clinical practices. Improved ability to predict which treatments will work best for specific patients will radically change prescribing patterns, as has already been seen in the field of oncology. Part III of this white paper series will take a closer look at how cancer genomics is revolutionizing cancer diagnosis and treatment. Practitioners in psychiatry and cardiology are also poised to begin incorporating precision medicine as their professional organizations begin to tackle the new capabilities offered by the field.

Practical applications for employers today

Precision medicine, genetic testing and pharmacogenomics are receiving increased attention from employers interested in enhancing their employee benefits. A major factor driving this interest is the promise of improved employee health resulting in less disability, better morale, less absenteeism, better productivity and increased company loyalty because the employee believes the company provides a valuable benefit the employee would not receive elsewhere.

There are a growing number of third-party vendors introducing precision-medicine capabilities as a voluntary benefit for employees or as a fully paid benefit for employees and their families. Growing consumer concern around heart disease, diabetes and Alzheimer’s disease are fueling this interest.

As noted in a BenefitsPRO article, one provider of these services “commissioned Survey Sampling International to query 536 U.S. consumers between the ages of 26 and 64 with employer-sponsored health insurance. The survey found a majority (65 percent) of respondents would be interested if their employer offered easy and affordable access to genetic testing for health purposes — provided the results were private and only shared between the employee and their doctor.

“Moreover, many people are willing to help pay for such testing if it were a workplace benefit.”
The results of this survey demonstrate relatively pervasive consumer awareness of an emerging medical practice, and those numbers are likely to increase. In a recent article in MedCity News, author Gavin Teo discusses what consumer genomics trends to watch in 2018, and he notes the reason direct-to-consumer marketing may play a larger role in growing the demand for precision medicine.

“[I]n 2017, the FDA issued a landmark decision to reverse its 2013 ban on 23andMe marketing their disease risk screen directly to consumers, leading to a more progressive regulatory environment in the United States. As a result, new distribution models are emerging. In addition to direct-to-consumer marketing and clinically prescribed tests, insurance carriers are now bundling tests with new policies, and employers are beginning to offer genomics tests as a risk screening tool and a covered employee benefit.”

As direct-to-consumer genetic testing companies, such as Ancestry.com and 23andMe, raise consumer awareness of precision medicine, employers are also starting to see an emergence of precision medicine companies offering new solutions for including genetic testing in employee benefits plans.

Currently, there are two ways precision medicine diagnostics are being offered through benefits plans:

1. Generalized testing (employer-sponsored, voluntary or shared cost) offered to everyone: This testing provides information to the tested individual on risks and propensities for medical conditions and responsiveness to certain classes of drugs where genetic phenotype can be a determinant of efficacy. Different providers of these services offer different combinations of these capabilities.

2. Targeted testing (usually employer-paid) offered to identified individuals: In this case, diagnostics through medical and pharmacy claims or administered questionnaires identify those individuals who will more likely benefit from a specific type of genetic testing. Those individuals are contacted and offered testing.
A framework for looking at precision medicine for employer benefits

When considering precision medicine or pharmacogenomics as part of an employee benefits plan, there are some basic questions employers should ask:

- What does the new capability offer?
- What is the upside of offering the testing?
- What are some of the potential downsides to offering the testing?
- Do the benefits outweigh the costs?
- Does the medical carrier pay for the testing? If no, why not?
- What are the ethical and legal ramifications of including genomic testing in the benefits plan?

In answer to the final question posed, Part IV of this white paper series takes a closer look at current policy addressing genomic information privacy as it relates to employers, but companies should always seek professional legal advice when making this type of change to their benefits plans.

For all testing it is important to consider:

- What is being tested?
- How is testing administered?
- How is confidentiality maintained?
- Will the employer see the results?
- What laboratories are being used, how are they credentialled, and how are quality controls conducted?
- How are results being delivered back to the recipient?
- Are genetic counseling resources made available? Are they mandatory?
- Are treating physicians included in the interpretation and next steps based on the results? Is treating physician involvement mandatory?
- What are the costs of testing?
- Are recipients able to maintain a connection with the service, and will they receive updates as new diagnostics become available?

If a company makes the decision to include genetic testing in an employee benefits plan, there are some best practices that should be followed.

- For testing that identifies inherited disease, provide mandatory telephonic counseling with a genetic counselor to all recipients screened. This counseling should be delivered by credentialled personnel who have been trained and equipped with tools to assist the recipient with accurately interpreting results.
- Include the recipient’s treating provider in a review of the test results, as well as any recommendations for changes in treatment.
- Consider the availability of longitudinal follow-up to update the employee’s report based on new diagnostic capabilities in the future. In theory, once the genome is sequenced, it may be run against newer algorithms not available for the initial report.
Challenges for precision medicine in employee benefits

As promising as these new technologies are, concerns around genetic testing for the employee benefits decision-maker may be considerable, including:

- Recipients having difficulty interpreting the significance of the results.
- Creating a false sense of security with false negative results.
- Creating a false sense of anxiety with false positive results.
- Recipients resigned to views that could be detrimental to their health.
  - “I am predisposed to a condition and there is nothing I can do to prevent it.”
  - “I am predisposed to a condition, so what’s the use changing my behaviors if I’m going to get the condition anyway?”
  - “I tested negative for the condition, so I don’t need to follow any preventive measures. I’m safe.”
- Patients making medical decisions without consulting their treating physician.

Employee benefits professionals should practice vigilance and seek guidance when considering what and how precision medicine diagnostics are introduced into their employee benefits plans. As best practices emerge, some of these areas will become clearer and more commonplace. Inevitably, new diagnostic technologies will continue to challenge what, when and how diagnostic testing is administered and results are delivered.
Where we will be tomorrow?

The promise of precision medicine is considerable. Better understanding of the underlying mechanisms by which various diseases occur can transform the way many diseases are diagnosed and treated. Improved approaches to preventing, diagnosing and treating a wide range of diseases has the potential to increase efficacy and ultimately lower healthcare costs.9

Technologic advancement is driving an accelerated growth in the genomics space. As noted in a recent medical delivery industry article, “Lab costs have continued to decline, consumer awareness is higher than ever, regulation has become more favorable in the U.S. and remains permissive in other parts of the world, and new models of payment and distribution have emerged.”10

As technology and delivery of these new diagnostic capabilities improve, employers will need to continue to monitor what’s available and what it means for their employee benefits offerings.
