Opportunity
While some 7,000 rare diseases have been identified that are estimated to affect 30 million people in the US and 350 million people globally, with most being children, effective therapies are still not available to more than 95% of the patients suffering from these diseases. Most major pharmaceutical companies are now working to develop rare disease therapies to improve lives and penetrate the $86B orphan drug market. Also, to take advantage of the tax credits, grants, fee waivers, market exclusivity, and accelerated regulatory approval process, made available through the US Orphan Drug Act, to promote investment. Since the passage of the Act in 1983, 2,755 agents have received an orphan drug designation in the US, with 424 orphan drugs now approved.

Problem
While tremendous investments have been made to develop new therapy, little has been done to accelerate patient testing upon launch of a new therapy. Testing is critical to achieve sustainable profitability and ensure patients are treated early, before damage occurs, so they have every prospect of leading a normal life. Surveys indicate it takes on average, 7.6 years in the US and 5.6 years in the UK for a patient with a rare disease to receive a proper diagnosis. A journey that, on average, requires a patient visit eight physicians; four primary care, four specialists and receive two to three misdiagnosis before confirming the cause of their condition. It has been projected that for the top 350 rare diseases, approximately 27% of patients will not reach their first birthday.

An orphan drug is defined as a drug that has been developed to treat a rare disease. The US FDA defines a rare disease as a disease with an incidence of less than 1:5,000 of the general population. While rare diseases have traditionally received no attention from pharmaceutical companies as the small target audiences do not justify the huge investment needed for drug development, the US was the first to propose an Orphan Drug Act (ODA) in 1983 to encourage the development and availability of orphan drugs. Some of the key factors involved in the growth of the orphan drug market include exclusivity options for multiple orphan indications, off-label usage, expansion to non-orphan indications, and freedom from generic competition. The growth is due in part to grants available through the US FDA for clinical research and clinical studies as well as orphan drug acts being established in all major countries. Total global revenue for orphan drugs was $86.1 billion 2012 and is expected to grow at a CAGR of 5.4% to reach $112.2 billion by 2017.
The conventional expectation is for a patient to rely on a physician for an accurate diagnosis of a common disease and an effective treatment. Often neither of these presumptions applies to a patient with a rare disease. Given the small universe of patients, a typical physician may never encounter a rare disease patient over the course of his or her entire career. Physicians also may not have the expertise to recognize or diagnose a rare disease. As such, pharmaceutical companies must play a key role in enabling physicians to recognize and properly diagnose such disorders.

**Need for Diagnostic Tools and Testing Infrastructure**

The challenge of finding patients post-approval is critical for the commercial success of any rare disease therapy but in order to do so, a sustainable diagnostic system must be set up so potential patients have a reasonable expectation of being seen and diagnosed by a disease expert so that informed decisions about treatment can be made.

While the Genzyme patient-centric business model established that investment in rare disease therapies can be a viable business model for pharmaceutical companies, Genzyme was forced to fund researchers to develop the diagnostic tests, validate these tests for clinical use and then implement the process to manufacture the required reagents for testing to go live. Additionally, Genzyme had to lead the efforts to source and register all of the components required for patient sample collection and then set up the laboratory testing infrastructure required to perform testing in each target region. Luckily this investment paid off with Genzyme now generating over $18 in annual revenue treating over 5,000 patients in 90 countries. However, not all companies have the competency and/or appetite to spend the tens of millions of dollars or the many years required to create this infrastructure or deal with the regulatory issues involved in offering diagnostic testing to different regions across the globe. Ultimately, Genzyme recognized that in order to maximize shareholder value, these resources were better spent elsewhere. Genzyme has since sold off its pharmaceutical intermediates, diagnostic products, and laboratory business to focus on its core competency of drug development.

**PerkinElmer’s Solution**

The PerkinElmer global diagnostics division is dedicated to commercializing complete solutions for prenatal, newborn and adult testing. With resources and global reach to provide testing to over 40 million newborns and adults each year, PerkinElmer leads the industry in CE-approved and FDA-cleared dried blood spot sample collection devices, test kits and CLIA-certified testing services. Pioneers in the development and commercialization of mass spectrometry, immunoassay, enzymatic and molecular assays for newborn screening, PerkinElmer can provide all the products and services required to identify patients with specific rare diseases.

These services* include:

- Optimizing and validating the required diagnostic screening tests according to CLIA requirements
- Manufacturing and distributing the reagents and quality control materials required for screening and diagnostic testing
- Contract manufacturing of sample collection kits to include all the components required, with instructions in local language, for physicians to obtain patient samples for testing
- Performing diagnostic screening on a fee per test basis according to CLIA or other country specific guidelines
- Providing an electronic report and genetic counselling to physicians to interpret test results and define next steps for patient follow-up

Contracting PerkinElmer to perform these services can save a pharmaceutical company millions of dollars and years of time when trying to establish the mechanism to identify patients that would benefit from a new therapy once launched.

Figure 1. Summary of Services
Taking advantage of PerkinElmer’s global network of certified laboratories also provides the testing and counselling required to allow physicians to find patients with a specific rare diseases faster so these patients can ultimately lead better lives. The application of these products and services is summarized in Figure 1.

*Please note that the Products/Services mentioned above may not be available in all countries.

**Summary**

While the targeting of rare diseases offers substantial opportunity for pharmaceutical companies, given the small universe of patients, pharmaceutical companies must play a key role in educating and enabling physicians to recognize and properly diagnose such diseases. PerkinElmer can save a pharmaceutical company millions of dollars and years of work by creating the diagnostic tests, diagnostic reagents, patient sample collection kits and testing services required for use by physician’s globally to identify patients as early as possible in the course of their disease. Patients that are critical to generating the revenue and market acceptance required to ensure commercial success of any new rare disease therapy.

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