Diagnosis of Rare Diseases

Remains Difficult

TRENDING NOW: New research highlights the need for improved physician education and referral programs.


Patti Engel

Patients living with rare diseases visit an average of 7.3 physicians before receiving an accurate diagnosis, according to a recent survey of patients, family members, physicians, and allied healthcare professionals (HCPs). The survey results, published in The Journal of Rare Disorders (JRD), also reveal a significant level of physician interest in helping patients with rare diseases.

The survey, conducted on behalf of Global Genes, reflects a growing appreciation among primary care physicians (PCPs) and specialists of the societal impact of rare diseases. Key findings include:

- Most physicians (60% of PCPs, 80% of specialists) say they welcome the challenge that rare diseases bring and want to be part of finding a diagnosis.
- 40% of PCPs and 24% of specialists indicate that they lack sufficient time to do a workup for a rare disease even when they suspect the patient may have one.
- For patients, the mean length of time from symptom onset to accurate diagnosis is 4.8 years (range: 0-20 years).
- 44% of patients agree with the statement: "Because of a slow diagnosis, treatment was delayed and the impact on my condition has been negative."

"It's encouraging to see the growing investment in rare disease research across industry, academia, and government," says Patti Engel, the lead author of the report and CEO of Engage Health. "The survey results show that innovative therapies are only part of the solution. There is an urgent need to bridge the knowledge gap by educating and connecting patients, families, physicians, and specialists."

For more information, visit engagehealth.com.

Breakthrough Designation Has POTENTIAL TO SHORTEN DEVELOPMENT TIME


Christopher-Paul Milne

The recently launched Breakthrough Therapy Designation (BTD) program in the United States, aimed at expediting development and review of drugs intended to treat a serious condition, has the potential to shorten development time considerably, according to the Tufts Center for the Study of Drug Development, which recently completed an assessment of the BTD program.

While the FDA approved only 30% of the first 113 BTD requests (with 60% denied/withdrawn and 10% pending) in the year and a half following the program's 2012 launch, Christopher-Paul Milne, director of research at Tufts CSDD, says early intensive guidance from senior FDA managers, a key element of the program, suggests that BTD-designated development programs will likely learn, sooner rather than later, whether BTD products will be successful.

"A key success factor for the program will be whether it serves the goal of helping drug sponsors and the FDA work together to cut development time, while encouraging the utilization of new development tools and methodologies, such as targeted diagnostics and adaptive clinical trial designs," Mr. Milne says.

The FDA already has tools to speed drug development — priority review, fast track, and accelerated approval — but these programs are limited in their ability to address scientific, regulatory, and economic factors that are dramatically shifting the R&D landscape, he says.

The assessment found that:

- Central nervous systems drugs (those for neurodegenerative and psychiatric disorders) and diagnostics currently comprise a small share of FDA approvals with special program designation (3% and 5%, respectively), but are likely to benefit from receiving the breakthrough designation.
- Although orphan designation technically is not an FDA expedited program for serious conditions, it often applies to the same investigational compounds as BTD.

For more information, visit csdd.tufts.edu.

Cross-Functional Communication OF REGULATORY STRATEGY BOOSTS IMPACT

Problems with understanding regulatory processes and requirements account for 38% of reported internal communication challenges, finds new Cutting Edge Information research.

The study discovered that regulatory affairs teams may encounter several challenges when interacting with interdisciplinary teams. Among surveyed companies, 48% of the reported difficulties revolve around misunderstanding regulatory requirements and strategy.

"One possible reason for these challenges is that some internal teams may not be aware of how frequently the regulatory environment changes," says Jacob Presson, research analyst at Cutting Edge Information. "In other cases, a team may think an agency will approve a drug simply because of its clinical benefits, and it may underplay agencies' priorities around cost-effectiveness and trial design."

About 11% of communication challenges revolve around issues in understanding regulatory strategy, including submission processes. Some functions may not realize the potential impact of regulatory strategy on product development, market authorization and post-approval marketing.

Regulatory affairs teams can correct these misunderstandings through discussions about regulatory guidances, processes, and strategy. Regulatory groups offer clarity on current agency guidelines and highlight how they will impact the product's development. In these discussions, regulatory affairs underlines agency perspectives and priorities to explain why certain guidelines exist.

For more information, visit cuttingedgeinfo.com. PV

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Growth Expected in
eClinical Solutions Market

The global eClinical solutions market has been categorized into two major segments, namely, products and professional services. The products market is further classified into clinical data management systems (CDMS)/electronic data capture (EDC), clinical trial management systems (CTMS), electronic clinical outcome assessment (eCOA), randomization and trial supply management (RTSM), safety solutions, and others. The other segments comprise core lab integration systems, regulatory information management services, and coding systems.

The CDMS/EDC solutions segment accounted for the largest share of the global market, by product at an estimated $899.1 million in 2013; while the eCOA solutions market is expected to grow at the highest CAGR of 21.2% from 2013 to 2018, according to MarketsandMarkets. The professional services market includes implementation services, training, support services, and consulting.

The global eClinical solutions market is estimated to grow at a CAGR of 13.5% from 2013 to 2018.

For more information, visit marketsandmarkets.com.

THERAPEUTIC TRAX...

AUTOIMMUNE

While the systemic lupus erythematosus (SLE) treatment market is predominantly limited to Benlysta and off-label Rituxan, new emerging entrants, which appear to offer significant therapeutic benefits, will cause dramatic changes to the future SLE biological treatment market landscape. There are extensive research efforts into innovative biologic programs in the SLE pipeline, which target a wide range of cytokines modulators, as well as cellular targets of the immune system. In fact, cytokine modulators now constitute the largest proportion (33%) of all active pipeline programs for SLE.

Source: GBI Research, Frontier Pharma: Systemic Lupus Erythematosus - Identifying and Commercializing First-in-Class Innovation

For more information, visit gbiresearch.com.

CANCER

The market value for monoclonal antibodies (mAbs) in breast cancer treatment will experience a massive increase from $4.8 billion in 2012 to $10.9 billion in 2019, demonstrating a CAGR of 12.2%. This double-digit increase will be driven mainly by the uptake of two recent approvals, Perjeta and Kadcyla, along with the rapid year-on-year growth in specialty pharmaceuticals in the U.S., which boasts the largest regional market for breast cancer treatment. Roche is currently the dominant player within this market, with a franchise of drugs (including the blockbuster Herceptin) that target the HER-2.

Source: GBI Research, Monoclonal Antibodies Market in Breast Cancer to 2019 - Strong Uptake of Novel HER-2 Targeted Therapies to Drive Robust Growth

For more information, visit gbiresearch.com.

There are currently three monoclonal Antibody (mAb) treatments approved for metastatic colorectal cancer with the purpose of extending the patient's lifespan; Avastin, Erbitux, and Vectibix. The current market leader is Avastin, which generated revenue of $6.2 billion in 2012 across a number of oncology indications. While compelling late-stage candidates are absent from the current pipeline, the anticipated rise in total prevalence of colorectal cancer in the top eight markets may inspire further investment. The global market has the potential to grow to a value of $5.5 billion by 2019.

Source: GBI Research, Monoclonal Antibodies Market in Colorectal Cancer to 2019 - Favorable Pricing Policy in the US and Rising Prevalence in Europe and Japan Ensures Market Growth

For more information, visit gbiresearch.com.

The bladder cancer therapeutics market in the six major countries — the U.S., France, Germany, Italy, Spain, and UK — is forecast to climb from $239.3 million in 2012 to $297.5 million by 2017, at a CAGR of 4.5%. The U.S. market will continue to generate the majority of sales, with its revenue increasing from $139.4 million in 2012 to $181.1 million by 2017, at a CAGR of 5.4%. This is due to the presence of higher-priced drugs in the U.S. market, as opposed to other countries.

Source: GlobalData, OpportunityAnalyzer: Bladder Cancer – Opportunity Analysis and Forecasts to 2017

For more information, visit globaldata.com.

CNS

Due to upcoming patent expirations for four high-profile drugs, the global Parkinson's disease (PD) market is expected to decline from $3.4 billion in 2012 to $2.9 billion by 2019, at a CAGR of 2.3%. PD drugs, such as Azilect (rasagiline mesylate), Stalevo (levodopa, carbidopa, entacapone) and Comtan (entacapone), will lose their patents by the end of the forecast period. Generic alternatives for these treatments have already been approved, which will result in further market competition. Additionally, the loss of patent for Mirapex (pramipexole dihydrochloride) and Exelon (rivastigmine tartrate) in 2010 and 2012 will continue to halt future PD market growth.

Source: GBI Research, Parkinson's Disease Therapeutics Market to 2019 - Pipeline Shows Shift towards Long Term Disease Management

For more information, visit gbiresearch.com.

The global schizophrenia market value is expected to undergo a modest increase over the coming decade, climbing from $6.3 billion in 2012 to $7.9 billion by 2022, at a CAGR of 2.4%. The U.S. dominated the schizophrenia space in 2012 with sales of $5.2 billion and a global share of 83%. It will maintain its market-leading position by 2022 with sales of $6.1 billion, representing a CAGR of 1.6% during the forecast period.

Source: GlobalData, PharmaPoint: Schizophrenia – Global Drug Forecast and Market Analysis to 2022

For more information, visit globaldata.com.

DIABETES

While the global type 2 diabetes market is crowded with inexpensive generics and marked by a pipeline filled with me-too drugs, this market is projected to undergo substantial growth between 2012 and 2022, more than doubling over this period. The main driver of this enormous expansion will be the dramatic increase in disease prevalence, which is attributable to increased life expectancy and an increasingly sedentary and stressful lifestyle. The second largest driver will be the physicians' efforts to delay disease progression and reduce the costly burden of diabetic complications through the use of combination therapies and novel branded drugs.

Source: GlobalData, PharmaPoint: Type 2 Diabetes - Global Drug Forecast and Market Analysis to 2022 – Event-Driven Update

For more information, visit globaldata.com.

GASTROINTESTINAL

The global ulcerative colitis (UC) market value will increase steadily over the coming years, growing from almost $4.2 billion in 2012 to about $6.6 billion by 2022, at a CAGR of 4.7%. The company's latest report states that out of the 10 major countries (the US, Canada, France, Germany, Italy, Spain, the UK, Japan, China and India), the US will continue to hold the largest UC market share by 2020, with 52%. France and Germany will follow with shares of 11% and 8%, respectively.

Source: GlobalData, PharmaPoint: Ulcerative Colitis - Global Drug Forecast and Market Analysis to 2022

For more information, visit globaldata.com.

INFECTIONS

Due to a large untreated patient pool and the recent approval of new treatments, the hepatitis C market will grow from $5.7 billion in 2012 to $18.6 billion by 2019, at an impressive CAGR of 18.5%. The hepatitis C market will receive this major boost with the 2013 approval of Sovaldi, which has the potential to transform the treatment algorithm for hepatitis C.

Source: GBI Research, Hepatitis C Therapeutics in Major Developed Markets to 2019 - Outstanding Recent Approvals and Late-Stage Pipeline to Transform Clinical and Commercial Landscape

For more information, visit gbiresearch.com.

The global hepatitis B virus (HBV) vaccine market value will experience modest growth over the coming years, climbing from about $959.9 million in 2012 to $1.19 billion by 2022, at a CAGR of 2.2%. The majority of HBV vaccine sales will take place in the US, which will boast a higher CAGR of 3.4% and expand its market share from 55% to 61.7% during the forecast period. The US will be followed by Canada and the five European countries, with expected CAGRs of 1% and 0.5%, respectively.

Source: GlobalData, PharmaPoint: Prophylactic Hepatitis B Virus Vaccines - Global Drug Forecast and Market Analysis to 2022

For more information, visit globaldata.com.

KIDNEY DISEASE

After an initial decline driven by generic and biosimilar competition in the G7 markets and changes in reimbursement practices in the United States, the total chronic kidney disease (CKD) market will return to growth, achieving total sales of $11.7 billion in 2022 compared with $11 billion in 2012. This market forecast includes erythropoietin-stimulating agents (ESAs), phosphate binders, calcium mimetics, active vitamin D analogues, antihypertensive agents, IV irons and emerging CKD therapies for the CKD non-dialysis and dialysis patient populations.

Source: Decision Resources Group

For more information, visit decisionresourcesgroup.com.

RESPIRATORY

The value of the cystic fibrosis (CF) market is expected to increase across the leading eight developed nations, from $695.6 million in 2012 to almost $4.5 billion in 2019. This equates to a CAGR of 30.4%. Novel treatments with disease-modifying mechanisms of action are the primary factor driving the growth of the value of the market. CFTR modulators are the first type of CF therapy to treat the root cause of the disease rather than the symptoms, and as a result, they offer patients improved relief and quality of life. Kalydeco (ivacaftor), was the first CFTR modulator to reach the market.

Source: GBI Research, Cystic Fibrosis Therapeutics In Major Developed Markets To 2019 - CFTR Modulators Initiate Drive Towards Personalized Treatment And Market Growth

For more information, visit gbiresearch.com.