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The Breakfast Club "Bottom Line"

Summary of Outputs from the 10th Blue Matter Breakfast Club™ Session

Tuesday, 26 October 2021

Patient Identification in Rare Diseases

A Live Consulting Session

What is the Breakfast Club?

Organized by Blue Matter, the Breakfast Club is a group of biopharmaceutical industry leaders that regularly meets by invitation to address and discuss important issues related to the industry. Breakfast Club members come from a wide range of companies, and many have a focus on rare diseases.

On 26 October 2021, Breakfast Club members participated in a "live consulting session", facilitated by Blue Matter, that addressed a key topic: **Patient Identification in Rare Diseases.** During two hours of breakouts and discussions, members pooled their knowledge to examine this topic and answer difficult questions rare disease companies often face.

Initially, participants examined the drivers and barriers to patient identification. Next, they discussed potential strategies and tactics to overcome barriers.

In this paper, we summarize the group's insights.

The Patient Identification Challenge for Biopharma Companies:

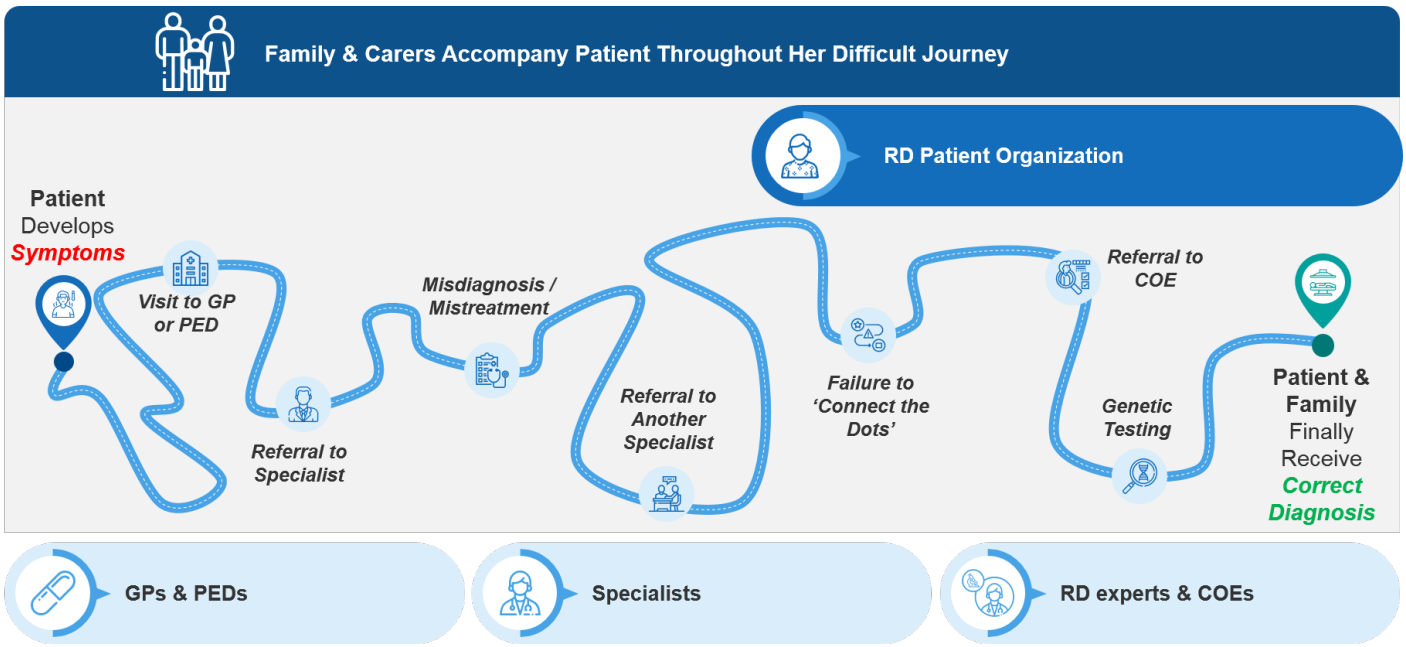
It can be quite challenging for biopharma companies to identify rare disease (RD) patients. This is because there are very few patients for any given RD, there are even fewer disease experts, and disease awareness is typically very low. As a result, biopharma companies often find it difficult to effectively size their markets, develop commercial strategies, and maximize the commercial value of RD therapies.

Patient Identification Drivers, Barriers, and Potential Solutions

When assessing the drivers and barriers for RD patient identification, it's helpful to consider them in two different contexts:

1. **The patient journey to diagnosis and treatment**: Patients often must travel a "long and winding road" to get a correct diagnosis and initiate treatment. Often, it can take years from the development of initial symptoms until the correct diagnosis is eventually established. The drivers and barriers to patient finding can manifest differently at different points along that journey.

Figure 1: The Long and Winding Road to Diagnosis



2. The key players who inhabit the RD ecosystem: These stakeholders experience or affect the drivers and barriers in different ways. The key players are described in Table 1.

Table 1: Key Players Surrounding the Patient in the RD Ecosystem

Parents, Family, and other Caregivers	Those who accompany the patient throughout his or her life and difficult journey from the first symptoms until the final diagnosis is established (and beyond)
General Practitioners and Pediatricians	First-line medical professionals who typically are the first to see a patient when initial symptoms develop
Specialists	Those who come in to evaluate and attempt to diagnose and treat specific symptoms
Patients and Patient Advocacy Groups (PAGs)	Other patients in a similar situation or affected by the same rare disease and/or patient organizations who typically come in later in the journey when a certain RD is suspected
Rare Disease Experts and Centers of Excellence (COEs)	Those who will—in most cases—finally establish the correct diagnosis and play a central role in the treatment decision(s) and in providing long-term care

With those two contexts in mind, the group:

1. Identified key drivers and barriers to patient identification across various stages of the patient journey
2. Provided some details / examples related to those drivers and barriers
3. Outlined some potential strategies and tactics that RD companies can use to overcome barriers

The tables below summarize the group's insights relevant to different stakeholders across key stages of the patient journey.

General Drivers and Barriers Across the Patient Journey (for Patients and Caregivers)

Drivers and Barriers	Detail / Examples	Strategies and Tactics
High unmet need coupled with high patient and caregiver burden	• Typically, RDs are associated with disproportionately high unmet need and burden for the patient • There are few treatment options for RDs, so new treatment options provide hope • The time to diagnosis is unusually long, and it may take several years until a clear diagnosis is obtained	• Provide better support for patients and caregivers, e.g., with targeted services • Drive disease awareness and broadly support patient organizations • Make more information accessible online; leverage social media
Disease rarity	• Difficult to find disease knowledge and expertise • Ultra-rare diseases are even more challenging • Patients have low share of voice	• Augment social media campaigns • Support patient umbrella groups (see below) • Educate patients on their (legal) rights • Make more information accessible online
Social media & digital education	• Can drive awareness • Can help patients get the right help	• Launch digital awareness campaigns • Leverage social media
Disease information in digital channels	• Caregivers, family, and friends can help to research symptoms to suspect diagnosis and self-refer to experts	• Leverage Google, social media, digital medical education • Create the "Rare Disease Internet" (one place where all ecosystem players can find information)

Drivers and Barriers Early in the Patient Journey (related to GPs and Pediatricians)

Drivers and Barriers	Detail / Examples	Strategies and Tactics
Very low RD awareness	• GPs and even specialists often never see a particular RD in their practice, so it can be a very low priority; they may not suspect the RD nor recognize symptoms • Pediatricians may sometimes attribute symptoms to developmental problems • Awareness is even lower in emerging markets, compounding the challenge there • Educating GPs and pediatricians is challenging, as they are difficult to reach and have lower interest in education for very rare diseases that they might never see; their focus is on more common diseases	• Raise disease awareness (GPs, patients and caregivers, specialists; need to tailor communication in language, content, etc. to the respective audiences) • Sequence approach to educate COEs, KOLs and HCPs; also need to remain open and adapt • Media campaigns, publications, medical education, social media, etc. • Targeted awareness campaigns: Identify key target groups, e.g., certain specialties that have previously referred patients • Motivate quicker patient referral to specialists & COEs

Drivers and Barriers in the Middle of the Patient Journey (related to Specialists)

Drivers and Barriers	Detail / Examples	Strategies and Tactics
Focus on symptom treatment	• Some specialists focus on treating symptoms; don't have time, interest, or expertise to look for the underlying cause of the disease • Treatment of symptoms can be a focus because there is no awareness of potential treatment	• Motivate / incentivize doctors to refer to experts • Targeted awareness campaign, e.g., for novel treatment options • Establish partnerships with COEs to educate specialists
Specialists unwilling to refer patients	• Want to keep patient and treat the condition themselves • Don't know to where patients should be referred • Due to time constraints and lack of knowledge, not motivated to diagnose	• Establish relationships with COEs, referral networks; educate, incentivize • Encourage earlier patient referral to specialist and/or to RD COEs • Map referral networks, targeted education, establish relationships, etc. • Increase awareness of RD COEs
Unclear diagnostic criteria	• Some RDs are ill-defined and lack clear diagnostic criteria • Particular RD maybe part of larger family lacking clear criteria for this sub-type • Natural history unknown	• Establish clear diagnostic criteria • Develop diagnostic tool, e.g., to provide to experts, patient organizations, etc. • Set up patient registries (1st step identify, then track)
Diagnosis difficult due to heterogenous symptoms / slow disease progression	• Hard to identify RD • Symptoms overlap with other more common diseases • Differential diagnosis very hard	• Get access to claims data, hospital patient data • Analyze patient data for clusters of symptoms to identify RD patients (possible in certain geographies; possible via a consulting project or 3rd party partners)
Diagnostic tests / genetic testing	• No funding for genetic testing in some places • Genetic testing cannot be done in certain markets and samples need to be sent to other countries • Genetic tests may carry stigma in some countries	• Drive earlier and more widespread genetic testing • Establish / coordinate with national testing programs • Drive biomarker tests • Develop RD test battery / synergize with other RDs • Newborn screening, RD genomic testing • Establish patient self-referral diagnostic service • Work with other companies / partners to develop multi-disease diagnostic tests • Work with geneticists • Empower patients and family members to get a referral to a geneticist

Drivers and Barriers Later in the Patient Journey (related to Other RD Patients and PAGs)

Drivers and Barriers	Detail / Examples	Strategies and Tactics
Other patients with same rare disease	• Patient to patient engagement is a huge opportunity as they share the same reality and experience	• Patient ambassadors can help other patients to suspect a diagnosis and convince them to get tested • Drive social media networks
Patient organizations strengthen share of voice for RD patients	• PAG partnerships are critical as they will drive awareness and help diagnose other patients • Social media can be effective to reach other patients • There are compliance / regulatory hurdles for companies to engage with patients & PAGs directly	• Help to organize patients themselves • Drive, support, and empower PAGs; co-create solutions and build win/win partnerships • Provide support to PAGs (e.g., tools / technical, legal, medical / scientific) • Company takes lead in linking national groups & organizing EU umbrella organizations; link / network existing PAGs in big countries with smaller ones • Provide financial support e.g., for travel, RWE studies, technical; e.g. through donations in a compliant way

Drivers and Barriers Later in the Patient Journey (related to RD Experts and COEs)

Drivers and Barriers	Detail / Examples	Strategies and Tactics
Isolated and geographically scattered expertise; very few COEs (if any)	• Because of disease rarity, there are also very few experts and COEs • Satellite centers are geographically dispersed • Big centers have more capability (opportunity)	• Link dispersed expertise by establishing RD / COE networks • Help establish / support COEs • Drive multidisciplinary approach to increase chance of connecting the right dots • Telemedicine, rare patient boards • COEs differ by market; pharma should support building higher-level COEs
Multidisciplinary RD expertise and COEs	• Referral and educational networks (e.g. COEs with specialists)	• Critical Success Factors: Create the ecosystem that can enable people to "sit around the same table" to provide best patient care

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New technologies, such as AI & deep learning	• Huge opportunity but has its challenges, e.g.: ◦ Getting the system set up ◦ Lack of coding (e.g., co-morbidities, biomarker data isn't recorded) in database	• Leverage technology to drive patient identification • Train 'Dr. Google' (e.g., to recognize clusters of symptoms typical for a particular RD) • Leverage AI & machine learning (examples: IBM Watson; collaboration between Microsoft & EURORDIS) • Analyze eHealth records with algorithm
New treatment and early access programs (EAP)	• Initially, limited awareness of new treatment • Difficult to find right channel	• Explore / establish patient finding strategy early (e.g. in clinical program / EAP) that can be adapted for commercial stage • News / media campaign
Pedigree analysis / founder effects	• Identify founder effects in hereditary diseases	• Literature search, pedigree analysis, look for affected families, analyze pockets of immigration (note the Alexion example with hypophosphatasia: mining of social media to identify those with teeth falling out, etc.)
Development / clinical studies	• Opportunity to generate interest & awareness	• Establish early patient finding strategies

Parting Thoughts

Before designing a patient-finding strategy for a particular RD, a company should map and deeply understand the patient journey from symptom development to diagnosis. This will help the company to more effectively

- Identify key drivers and barriers
- Determine their quantitative impact
- Identify key leverage points

The deeper understanding that this process offers will enable the company to prioritize leverage points, identify the highest-impact activities, and develop the most effective patient-finding strategy.

In any case, a patient-finding strategy must be realistic, scalable and also sustainable. It's also important to remember that plans should be country or region-specific, as some approaches will not work in all regions or countries.

A key to success is flexibility. While planning is important, companies must be adaptable and willing change course where necessary.

New Ideas. Better Results.

Blue Matter is a consulting firm serving the life sciences industry. We strive to bring a new approach to consulting with original ideas that deliver a meaningful impact.

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