



# CRA Insights: Life Sciences

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## How collaborations with patients in rare diseases can support increased access to medicines

Patient advocacy groups (PAGs) are playing an ever-widening role across disease areas, from supporting individual patients to driving shifts in therapy development, market access and policy. Nowhere is this potentially more significant than in rare diseases where the awareness and understanding of patients' conditions and needs are lower, regardless of treatment availability.

### The rare disease contradiction

#### Rare diseases



Small patient numbers, paucity of data and the high cost of therapies are all prevalent in rare diseases and areas where PAGs can make a difference. PAGs can have an important role in improving the understanding of patient needs across all stakeholder groups. For example, PAG involvement in patient recruitment for clinical or natural history studies has helped to enable rare disease drug development.

As industry evolves to become more patient-centric, effective collaboration with PAGs may provide a competitive advantage. This is particularly relevant in the rare disease space where patients play an even more important role in influencing treatment choices and access to therapies.

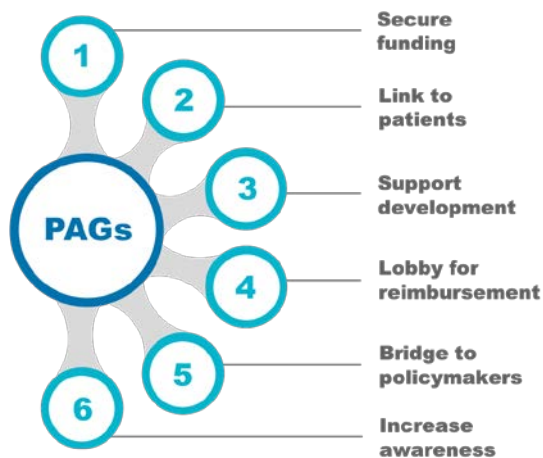
In this paper, we explore recommendations for more impactful collaborations with PAGs in the rare disease space. We look at where PAGs have been effective in influencing decisions and processes, and the impact of pharma/PAG collaboration in rare diseases.

## How PAGs have impacted rare disease decisions and processes

When successful, collaboration between PAGs and industry in rare diseases can be mutually advantageous. PTC Therapeutics leveraged such a relationship with the Parent Project Muscular Dystrophy (PPMD) PAG. This relationship served as a bridge to patients, policy makers, researchers, physicians and caregivers.<sup>1</sup> The partnership was viewed as beneficial on both sides. Early work with PPMD to identify lead compounds enabled PTC Therapeutics to secure state funding for early-stage research and gain a more in-depth understanding of Duchenne muscular dystrophy. For PPMD, the collaboration allowed them to provide valuable information on R&D progress to their patient members as well as to influence development decisions by PTC Therapeutics to align with their members' needs.

PAGs can also support the drug approval process in rare diseases by compensating for the dearth of available data and challenges in finding patients. For example the development of Kalydeco, approved by the US Food & Drug Administration (FDA) in 2012, represents a successful partnership between a PAG, the Cystic Fibrosis (CF) Foundation, and Vertex Pharmaceuticals.<sup>2</sup> Kalydeco was indicated for approximately 4% of CF patients with a specific mutation and the CF foundation recognized early on, given the rarity of the indicated population, the importance of establishing a patient registry and promoting patient participation in the search for a treatment. The partnership with Vertex was so successful that it was even mentioned by the FDA at time of approval.

### How PAGs have impacted rare disease decisions and processes

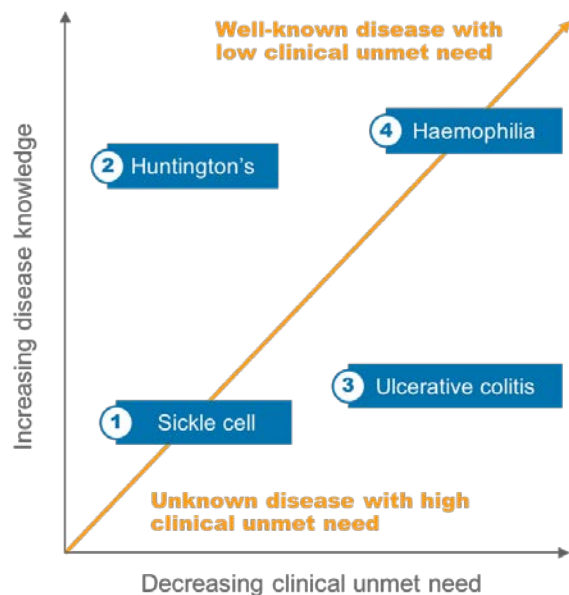


PAGs can also have an important impact on reimbursement decisions in rare diseases. In Belgium, patient and PAG letters have been included in reimbursement dossier submissions.<sup>3</sup> One letter highlighted the user-friendliness of Tobii's Podhaler to suppress chronic lung infection in cystic fibrosis patients and contributed to its acceptance. Another letter, documenting experiences with Exjade relative to other treatments for chronic iron overload, helped secure reimbursement. These examples show how the input from PAGs can increase payer awareness of the patient experience and help support the clinical and practical value of novel treatments.

In developing countries, such as Latin America, PAGs have had a considerable impact on the policy landscape by being strong voices for the rights of rare disease patients.<sup>4</sup> The lack of national policy and dedicated funding for rare diseases in developing countries can mean that patients struggle to gain access to treatments. Efforts by organisations such as the Geiser Foundation in Argentina and the Colombian Federation for Rare Diseases (FECOER) in Colombia have contributed to the implementation of national plans for rare diseases with goals to improve research, care and support. With government-backed priorities for rare disease, there is improved access to treatment.

## Collaboration with PAGs adds value by bringing more stakeholders to the table

**Figure 1: Spectrum of rare diseases according to disease awareness and level of clinical unmet need**



Source: CRA analysis of various sources

PAGs can add value and may play different roles depending upon disease awareness and the efficacy of existing treatments.

Figure 1 highlights these two dimensions of rare disease (awareness and unmet need) and suggests some examples in each quadrant.

1. Sickle cell is still relatively unknown and is characterised by high clinical unmet need. In this immature disease area, collaboration opportunities could be many and varied and include raising awareness of unmet need, enhancing physician education, promoting research or supporting development efforts. Bolstering the evidence base and providing patient perspectives on disease burden could also be areas of added value. However, PAGs in disease areas like Sickle cell, are likely to be fragmented and underdeveloped; therefore industry could help PAGs by providing financial and strategic support.
2. Huntington's disease is relatively well-known but with a high clinical unmet need due to a lack of effective treatment. PAG/industry collaborations focused on engaging physicians and research institutes could help support research and development and drive the discovery of effective treatments.
3. Public awareness of ulcerative colitis is considered low, but patients with the disease are currently well cared for with anti-TNF treatment. Therefore, PAG/industry collaborations should focus on building awareness and education across all policy influencing stakeholders (political, institutional and the media).
4. Haemophilia is one of the best known and most effectively treated rare diseases, with a range of treatment options. Whilst PAGs are more established and have a history of integration and collaboration in this area, they continue to play a critical role. PAGs work to maintain treatment availability and funding by engaging with payers and regulators via registries and real world evidence (RWE), raising standards of care where relevant, e.g., by supporting medical training and treatment centres, and building wider networks and using the media to communicate the continued needs and importance of the disease.

For each of the four disease examples, PAGs gravitate to identifiable roles based around the specific needs of their patient community, closely correlated with the nature of the disease, the availability of treatment and specific local factors. PAGs operating in disease areas with high unmet need or low awareness, such as Huntington's disease or ulcerative colitis, are often focused on what is lacking to drive progress to increase awareness of the disease and/or to reduce unmet clinical need.

Because collaboration with a PAG needs to be based around the role or mission of each individual PAG, the focus of activity will necessarily be determined by where the disease sits on the spectrum in Figure 1. Part of this activity will involve consideration of how PAGs interact with wider stakeholders and therefore how collaboration with industry can drive wider engagement among interested groups. In addition, an assessment of the resources and capabilities of a PAG will help identify areas where pharma support or partnership can make a material difference to the PAG's ability to deliver on its goals. This could be as simple as providing training or technical assistance on becoming more established and how best to support R&D, which small PAGs find difficult to afford or access.

Whilst we have anecdotal evidence of successful PAG collaboration, there are considerably more opportunities for PAG/industry collaboration in the rare disease space.

### How to build a successful Industry-PAG partnership



Collaboration needs to start with both parties understanding the benefits of working together and a willingness to compromise to ensure mutual gains. Industry can drive this, but needs to better understand the needs of PAGs to foster complementary long-term partnerships.

To do this, rethinking the role of the patient as a partner, and not only a customer, may require dedicated teams on both sides to handle this special relationship. Whilst this requires an investment in people, such teams are critical to ensure alignment of

goals, incentives and communication. To a certain extent, the skills required for these teams may also depend on how developed PAGs are, and the status of the disease, in terms of awareness and level of clinical unmet need.

### Begin now

Starting early is a wise strategy. Engagement of PAGs at pre-clinical stages to help define disease characteristics and crystallise patient burden with natural history studies can help plan more impactful clinical programmes. PAG involvement in clinical trials or even RWE provides a clear win-win. Whilst assistance with recruitment could speed up industry's development process, PAGs can provide their patient members with information on the latest advances in clinical research and help shape clinical development to better answer patients' needs.

To ensure mutual benefit, any PAG/industry collaboration requires commitment and open communication from both parties. Objectives, roles and responsibilities, timelines and expected outcomes all need to be clearly laid out at the outset of any partnership. Lack of familiarity in working together will also need to be counteracted through candid, honest and regular dialogue and a commitment by all to succeed.

Opportunities for PAG/industry collaborations are many and varied in the rare disease space. Although the path to effective collaboration has its difficulties, companies that harness PAG potential will reap the benefits, namely more patients and “customers” with increased access to treatment.

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