Preventing gatekeeping delays in the diagnosis of rare diseases

GPs acting as gatekeepers render a healthcare system easily accessible as well as affordable. However, gatekeeping can have an important drawback: it may hamper timely diagnosis and treatment of patients suffering from a rare disease (incidence <1:2000),[1] especially if patients present with common symptoms. This delays referral, possibly resulting in permanent organ damage, reduction in quality of life, and increased healthcare costs. In this article we reflect on both organisational and GP factors that contribute to the problem, and offer suggestions on organisational and educational approaches to tackling it.

> *"GPs acting as gatekeepers … may hamper timely diagnosis and treatment of patients suffering from a rare disease …* [unless they] *make use of their core quality: their "gut feeling" based on "pattern recognition".'*

USING AN ORGANISATIONAL PERSPECTIVE WHILE STUDYING THE PATIENT JOURNEY

Timely recognition is not only a problem for GPs: patients also travel between different healthcare professionals throughout the healthcare system. Ideally, these patient 'journeys' are organised as 'care chains' with the GP guiding and supporting the patient, but, in practice, this is often not the case. This causes problems that are also *organisational* in nature. Two related business disciplines that are rather unconventional in the (bio)medical field, operations management (OM), and supply chain management (SCM), could be used to reflect on the problem.[2,3] OM concentrates on the operational activities necessary for the creation of a single company's output (goods and/or services). SCM aims to improve the delivery of orders to customers that require the collaboration of different organisations. For patients travelling through various dissimilar parts of the healthcare system, the SCM approach can support our analyses.[4] An SCM approach would particularly focus on interorganisational requirements needed for a smooth patient journey through the system.

The many patient handovers during troubled patient journeys increase the possibility of errors because information is lost.[5] Specialisation is required for in-depth understanding of rare diseases, but it can cause fragmentation of care. Collaboration through integration, with internal processes becoming interlinked, spanning the traditional boundaries of the separate organisations, is likely to be helpful,[6] and may be achievable through enhancing information sharing, leading to a shared understanding among all concerned. Effective tools for information exchange and feedback are indispensable.

We investigated the problem in an exploratory case study. We focused on the specific flow from primary to secondary care because this is the first, crucial step in a patient's journey.

ILLUSTRATION OF THE PROBLEM IN AN EXPLORATORY CASE STUDY

We used the clinical pathway of immunology in the Jeroen Bosch Hospital (JBZ) as an example, which is specifically designed for recognition and treatment of patients with rare and difficult-to-recognise primary (intrinsic) immunodeficiencies (PIDs). Primary antibody deficiencies (PADs) are the most prevalent (but still rare) PIDs, and lead to a higher frequency of infections in the upper and lower airways, often accompanied by severe chronic fatigue. In themselves, these airway infections are common diseases; this renders the recognition of underlying PAD even more difficult, leading to significant delay in diagnosis and adequate treatment.[1]

We have explored this area through qualitative research involving semi-structured interviews with GPs and patients.

The five GPs thought that they should take a role in recognising potential immunodeficiency, because they have the best lifelong overview of the patient's medical history. They also felt that it was their task to differentiate more severe from milder diseases. They found it very difficult to recognise rare diseases, especially when patients presented with common symptoms. The GPs rated their own knowledge on immunodeficiency as generally insufficient, not knowing the warning signs, nor the potentially useful screening tests and their interpretation. They would refer children to a paediatrician, but were less sure about adults. The GPs showed interest in methods to enhance their role; they mentioned useful warning signs, help in interpretation of simple tests, and accessible consultation with experts. They did not know what happened to their patients once they had referred them.

The patients described many respiratory infections and antibiotics, repeatedly visiting the GP, ENT surgeon, and paediatrician. They felt they had to get angry before something happened, demanding referral, further investigations, or treatment with antibiotics. They experienced miscommunication between healthcare professionals, lack of information exchange, and too little attention to the whole picture.

Organisational reflection on the case study's results

Although they each have a very different perspective, both patients and healthcare professionals in fact described similar interorganisational problems. The most striking observation in the patients' interviews was the disturbed flow through the healthcare system, accompanied by a lack of coordination, collaboration, and adequate exchange of information during patient handovers between healthcare professionals in the different organisations, and lack of guidance and coordination from their GPs. The GPs described problems during patient handovers, and felt they did not get enough information from the hospital specialists to increase their knowledge for better recognition of potential underlying disease, or to know what to do with diagnosed patients.

Potential solutions

The GPs actually mentioned important warning signs during the interviews; simply offering more education about those core warning signs will not help. The problem they face is *how to recognise them in the backdrop of the general population*, where patients often suffer similar symptoms. We

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believe the best way to tackle this problem would be to educate GPs to make use of their core quality: their 'gut feeling' based on 'pattern recognition', the detection of a divergent pattern in a patient compared with the usual patterns seen in other patients.[7] To use this for the recognition of a rare disease, in-depth knowledge of the rare disease in question is not needed. However, the positive awareness that rare diseases do exist, and that they are indeed encountered in general practice, is crucial. This could be reached by *targeting postgraduate education to the phenomenon 'rare disease'*, and not to limit it to one or more specific (groups of) rare diseases. Being alerted to the phenomenon may make GPs less likely to overlook those signals, without alerting them unnecessarily to specific symptoms when the divergent pattern is not present. This is feasible, and an important message to convey.

In contrast to GPs' primarily holistic and person-focused care, medical specialists have a disease-focused perspective, in which they focus on the most likely diseases in their specific field that their referred patients may have.[8] In that way, the overall pattern may be lost in repeated referrals between different specialists in secondary or even tertiary care. Here is another important chance for GPs that can be targeted in postgraduate education: their person-focused, holistic nature of care enables them to oversee the patients' troublesome journeys through the healthcare system, being both the first as well as the continuing point of contact for the patient. They can use this for safety-netting, for themselves, and for instructions to their patients (*'come back to me for further investigations when this continues to occur'*).[9]

Unfortunately, even when alert to potential rare diseases, pattern recognition can be problematic for rare diseases presenting with common symptoms, as is the case in our rare disease PAD, which presents with common respiratory infections and, often, chronic fatigue. GPs see many patients who suffer from such problems, and, in most cases, no rare disease is present and referral is not necessary.

Innovative tools making use of modern data-driven methods linked to electronic patient files in GP practices (or hospitals or pharmacies) are most likely to get a desirable outcome for this problem. They are already in use, leading to electronic alerts; for example, with guideline adherence and drug interactions. However, these methods should be developed in the right context, adapted to the fact that these patients are hidden in the background of the general patient population. Warning signs purely developed on data from expert centres will not solve the problem. If this is neglected, an overwhelming number of false-positive warnings will ensue (the 'false-positive paradox'),[10] and GPs will soon stop making use of the system. It will be very interesting to further elaborate this approach in future research.

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