RPCPE
ISSN 2613-943X (print)
ISSN 2620-5572 (online)
Journal Homepage:
https://jurnal.ugm.ac.id/rpcpe
Review of Primary Care Practice and Education
(Kajian Praktik dan Pendidikan Layanan Primer)

Managing Rare Disease for Practicing Primary Care Physicians

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To cite this article:
Mahendra C. Managing rare disease for practicing primary care physicians. Rev Prim Care Prac and Educ. 2021; 4(2): 2-3.

A 5-year-old boy was brought to the primary care with primary complaints of fever and coughing for 5 days. The patient was given antipyretic and cough liquor by his mother to ease the symptoms but it did not give any improvement. The symptoms were sometimes accompanied by occasional diarrhoea. No other symptom was observed at that time. According to his mother, the patient has had similar symptoms a few times during the last year, about 3-4 times. The mother said that he seems to be relatively 'easy to get sick'. The symptoms arise and worsen if he is exposed to smoke and cold wind. Every time he gets sick, several drugs were prescribed by the doctor and he was treated as an outpatient. No history of allergy was known. The patient is the first child in his family. History of similar symptoms for the mother and father of the boy was denied.

INTRODUCTION

Primary care physicians (PCPs) are trained to provide care for a wide scope of medical conditions within their patient population. They are supposed to be the very first healthcare provider the patient should visit when having an illness. Accordingly, it is customary for PCPs to encounter many types of patient cases in their practice. In addition to common diseases, some patients may present with symptoms of a rare disease (RD).

Unfortunately, PCPs often lack knowledge to promptly detect and manage the patients with such disease. A research from Ireland reveals that only a third of PCPs were able to establish a diagnosis for RD and refer the patient to the appropriate specialist[1]. In Spain, more than half the PCPs were deemed fair or poor in knowledge of RD[2]. Similar result was also obtained from an expert survey in Belgium, stating that the basic RD knowledge of most PCPs in the country is low[3]. It is therefore justifiable for Evans to say that many patients with RD are un- or misdiagnosed[4] and thus, it may take years to finally get a correct diagnosis[3]. In Indonesia, there has been no published local- or nationwide research surveying the PCP's knowledge concerning presentations of RD.

DISCUSSION

Most RDs are seriously disabling and life-limiting or even life-threatening[4,5]. Moreover, the therapies for these RDs are few and costly, especially if treated at a late stage[5]. Consequently, patients with RD often face serious financial burdens, as well as physical and emotional burdens caused by the disease[3]. As such, early detection and treatment are important for these patients. Although the specialists are the ultimate healthcare providers to manage these patients, the role of PCPs is crucial as the 'first gate keeper' to suspect and refer them to higher care where needed. This is important to reduce the 'diagnostic odyssey' that many patients have to take in seeking help and eliminate unnecessary hospital visits, investigations, and inappropriate treatments[4].

As the name already implies, rare disease occurs rarely in daily practice encounters. It is likely that most PCPs will not see the RD cases often during their training years, resulting in inadequate experience to deal with rare cases. The predicament seems to be compounded by the fact that PCPs are sometimes lacking sufficient academic training by relevant tutors during the training years[2,3]. Most patients with RD are just referred to specialists directly by the PCPs, without trying to consult one to establish a diagnosis[1]. Furthermore, most PCPs are not even aware of web-based resources to look for RD information (e.g. findzebra.com, omim.org, orpha.net)[4] and improve their knowledge[1,2,3]. The lack of knowledge and skills to handle RD cases of the PCPs may leave the suffering patients unsatisfied with the PCPs performance during the visit and drive them to look on the Internet themselves to find more knowledge about their disease[2,6]; sometimes it even occurs that the patient knows more about their condition and thus educates the

PCP about the disease[2,7].

What can be done to improve PCPs' knowledge and management's performance in the office? The most important step is to start raising the awareness about existing RD resources as a means for PCPs to get access to knowledge about RD. The most concrete approach is through the continual medical education (CME) activity. The regulatory authorities may mandate the PCPs to undertake the compulsory CME credits about RD as an introduction to new developments. This may be in the form of journal reading, seminars, and taking online quizzes through RD websites. As a PCP, one should know the essential basics, especially the 'red flags' that warrant the case to be referred[3] such as the most conspicuous presentations pertaining to the disease, the progression nature of the given disease, and the potential dangers if ignored or left untreated.

Access to a shared electronic health record (EHR) could also improve communication and patient care[1], since this may allow coordination and exchange of information between the treating specialist and PCP. In that way, the PCP may get knowledge from the specialist's experience in managing the referred patient by learning from what was recorded in the EHR such as detailed physical examination results, laboratory parameters requested and checked, and drugs and/or interventions given. These will update the PCP's knowledge and may contribute toward better, and early detection of RD cases at the PCP office.

Another effort is to look at the course of the patient's disease during the practice. Unusual presentations or symptoms at first office visits should raise the awareness of the treating PCP of the possibility of a RD case. Combined with the worsening symptoms which are resistant to preliminary treatments or medications and/or unexplainable progression, these signs should alert the PCP to evaluate the patient thoroughly before referring to a higher care. The use of genomic testing seems to gain the spotlight in the diagnosis and management of RD, but this certainly needs the PCP's knowledge to translate and convey the findings to the patient[4]. Although advanced, the ability to order and interpret such testing for PCPs will be a great advantage for daily practice as part of the initial management of suspected patients before referring. This challenge could be solved by means of the aforementioned CME activity. The CME activity should therefore be structured well – from pathophysiology, through typical findings, to preliminary interventions - for the PCPs to study and understand about some 'common' RD that may be occasionally encountered in the practice.

The treating PCP as the 'family physician' needs to provide holistic care to the patient and reduce the burden of suffering that could result from the 'devastating' RD case. Although a PCP will eventually refer the patient to a specialist care, at least the patient should be well-informed about the disease first. The patient should be counseled to cope and be readied to deal with therapies he/she will have to undertake later[6]. This information seeking and sharing is what should be expected after the CME activity intervention given to the PCPs. In short, education is the primary and most important step for PCPs to start thinking about rare diseases that a patient could possibly present with in primary care.

References

1. Byrne N, Turner J, Marron R, Lambert DM, Murphy DN, O'Sullivan G, et al. The role of primary care in management of rare diseases in Ireland. Irish J Med Sci. 2020;189:771-6.
2. Ramalle-Gómara E, Domínguez-Garrido E, Gómez-Eguílaz M, Marzo-Sola ME, Ramón-Trapero JL, Gil-de-Gómez J. Education and information needs for physicians about rare diseases in Spain. Orphanet J Rare Dis. 2020;15(1):1-7.
3. Vandeborne L, van Overbeeke E, Dooms M, Beleyr BD, Huys I. Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in Belgium. Orphanet J Rare Dis. 2019;14(1): 1-11.
4. Evans WRH, Rafi I. Rare diseases in general practice: recognizing the zebras among the horses. Br J Gen Pract. 2016;66(652):550-1.
5. Jo A, Larson S, Carek P, Peabody MR, Peterson LE, Mainous AG. Prevalence and practice for rare diseases in primary care: a national cross-sectional study in the USA. BMJ Open. 2019;9(4):e027248.
6. Litzkendorf S, Babac A, Rosenfeldt D, Schauer F, Hartz T, Luhrs V, et al. Informational needs of people with rare disease–what information do patients and their relatives require?. J Rare Dis Diagn Ther. 2016;2(2):40.
7. von der Lippe C, Diesen PS, Feragen KB. Living with a rare disorder: a systematic review of the qualitative literature. Mol Genet Genomic Med. 2017;5(6):758-73.