

Supporting patients with a rare disease

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Do I write of the emotional impact I experienced when I learned of my 28-year-old son's rare disease late in 2013 and the sense of devastation, guilt, loss and anger I felt when he was diagnosed with primary sclerosing cholangitis (PSC)? Do I take a more pragmatic, academic perspective? In truth, both came to the fore after his diagnosis. While searching for evidence-based information on PSC, I spent as much time in tears, confronting the reality of his diagnosis. There is no cure for most rare diseases,¹ including PSC; as a mother, this was simply heartbreaking to read.

It is difficult to source Australian data on the incidence and prevalence of PSC, or indeed most of the rare diseases. Current Australian data on the prevalence of PSC are based on research adapted from a 2001 UK study. These data were extrapolated to the 2012 Australian population, providing an estimate of 872 people with PSC that year.² The only research on PSC I could locate in the academic and grey literature was from the US, UK and European countries.

Epidemiological data are unavailable for the majority of rare diseases worldwide due to the absence of a universally recognised coding system to establish adequate databases.¹ With the advent of the International Classification of Diseases 11th Revision (ICD-11), it is anticipated that there will be a mechanism to more accurately gather data on the incidence and

prevalence of rare diseases.³ With many thousands of rare diseases, and low incidence and prevalence in each, there is a concomitant lack of researchers working in the field for each specific disease. Difficulties then arise when seeking research collaborations aimed at increasing knowledge and developing treatment options.⁴

Feeling we were the only people in Australia dealing with PSC, I searched for a support group, to find none existed. In a recent study, two-thirds of patients with rare diseases were not given details of relevant patient support groups.⁵ It is not hard to see why. With low numbers and diverse geographical spaces where those with a specific rare disease live, the potential for face-to-face support groups is severely hindered. The use of online support groups (OSGs) then becomes a reality as the only available option. A growing body of research on the use and efficacy of OSGs, including a limited amount of research considering rare diseases, is demonstrating overall positive findings.⁵⁻⁷

I am a member of a number of closed Facebook support group pages for PSC and my lived experiences concur with that of the published literature. Information and emotional sharing have been found to be the most frequently sought and offered in OSGs, and are among the strongest features of the OSGs in which I am involved, followed by networking, esteem and, finally, tangible support.⁶⁻⁸ Interestingly, advances in

public knowledge and awareness of rare diseases, as well as research funding and legislative changes to support orphan drug development, have derived largely from the strong advocacy of patient support groups.^{1,9}

Many patients (and caregivers) do not receive information about their disease when diagnosed and are left to find their own reliable and accurate information,⁵ an often difficult task. The usual patient–doctor relationship may become challenged when a rare disease is involved. Given the high number of rare diseases, combined with limited knowledge and treatment options, the general practitioner (GP) is no longer necessarily the expert in this medical encounter, as rare diseases are also uncommon for the GP. Thus, the more 'traditional' roles of GP and patient may be disrupted, and the patient/caregiver becomes the expert regarding the disease.¹⁰ This disparity could be mitigated by a generic general practice approach to rare diseases as suggested by Knight and Senior.¹¹

A multidisciplinary team is usually required in the management of rare diseases, given the impact on multiple body systems, yet, coordination and communication between health professionals may be arbitrary, with the patient/caregiver being the one to facilitate information-sharing and care. We have taken the lead in our situation, providing an up-to-date, tabled chronology of our son's complex medical history to

each health professional. This details who he has seen and why, and all results and reports for each investigation. This adds to the development and construct of 'expert' patient.¹⁰

GPs can actively facilitate their patient's developing expertise. Advising of trustworthy websites, such as Rare Voices Australia, can empower patients to learn more about their disease, enhancing the clinical partnership (Table 1).

Given the beneficial findings of OSGs, together with the lack of face-to-face support groups, it would appear reasonable to suggest Facebook and other social networking websites to newly diagnosed patients. Not all will be interested in support networks, but it is important that they are informed and provided with options for those who do seek additional knowledge and support.

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Competing interests: None.

Provenance and peer review: Commissioned, externally peer reviewed.

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Table 1. Useful websites

Rare Voices Australia	www.rarevoices.org.au
GaRDN (Genetic and Rare Disease Network)	www.geneticandrarediseasenetworg.org.au
Association of Genetic Support of Australasia	www.agsa-geneticsupport.org.au
National Organisation for Rare Diseases	www.rarediseases.org
Rare Disease Dot Org	http://raredisease.org/index.html
Office of Rare Diseases	http://rarediseases.info.nih.gov
Genetic Alliance	www.geneticalliance.org
EURODIS	www.eurordis.org
National Human Genome Institute	www.genome.gov/27531963
Orphanet	www.orpha.net/consor/cgi-bin/index.php

Many of these websites provide links to more comprehensive information and disease-specific sites as well as online support groups.

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