
Consideration of a Legislative Framework to Support the Diagnostic Odyssey Commonly Encountered in the Instance of Rare Disease

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The diagnostic odyssey refers to the struggle to achieve a diagnosis for a medical condition in the face of significant implications if a diagnosis is not made. It is a common experience for people living with a rare disease. Western Australia has led the way in Australia in being the first State to establish a rare disease policy framework and an Undiagnosed Diseases Program (UDP). The UDP includes an expert panel made up of various specialists brought together with the aim of arriving at a diagnosis through collaboration. This article looks at the possibility of enhancing initiatives such as the UDP through a legislative framework. Relieving the medical, financial and emotional implications of the diagnostic odyssey is particularly important when one considers that taken together, rare diseases affect millions of people globally.

Keywords: *rare disease; diagnostic odyssey; diagnostic panel; Undiagnosed Disease Program; medical panels*

“I don’t know”, said the doctor. Therein ensued a glaring duel between patient and doctor as the eons ticked by. In an effort to break the awkward silence, the doctor added, “Trust me, this is good news. You don’t ever want to be one of my patients.” Agreed. But now what?

These few words seek to capture the frustration arising from the “diagnostic odyssey” – the struggle to achieve a diagnosis for a medical condition in the face of significant implications if a diagnosis is not made. At its most basic, a diagnosis determines the route to take when navigating the labyrinthine corridors of a hospital (Cardiology? Endocrinology? Haematology?). At its most consequential, lack of a diagnosis has an impact upon a patient not only physically and mentally, but also socially and financially.¹ The problem is compounded when a person, relieved by the identification of one diagnosis, learns that the diagnosis is wrong. The consequences are all the more devastating if the time delay occasioned by the wrong diagnosis, or wrong treatment in the face of the wrong diagnosis, means irreversible harm to health.²

The diagnostic odyssey is often considered in the context of rare disease, and while not all rare diseases will be difficult to diagnose, an early and accurate diagnosis has been acknowledged as a particular challenge in the rare disease context.³ In terms of quantifying the diagnostic odyssey, an European study found that a quarter of patients did not obtain a diagnosis for between 5 and 30 years, and 40% of

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¹ BM Kuehn, “US Launches Undiagnosed Disease Program” (2008) 300(1) *Journal of American Medicine* 27.

² National Academies of Sciences, Engineering and Medicine, *Improving Diagnosis in Health Care* (The National Academies Press, 2015).

³ Department of Health, Commonwealth of Australia, *Draft National Strategic Action Plan for Rare Disease* (July 2019) 6; see also *Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Disease: Europe’s Challenges*, Brussels, 11.11.2008 COM(2008) 679 Final at para 2.



patients experienced a wrong diagnosis. An Australian study found that a diagnosis was not reached for about a third of participants following more than five years on the diagnostic odyssey, and that more than half had experienced an incorrect diagnosis.⁴

In the light of this, the following will briefly describe rare disease, some barriers to diagnosis peculiar to the rare disease context and possible avenues of redress for those struggling through the diagnostic odyssey. Against this background, the Undiagnosed Diseases Program (UDP) established in Western Australia will be explored and high-level consideration given to whether such a program could be enhanced by being underpinned by a legislative framework.

WHAT IS RARE DISEASE?

A common approach to defining rare disease is to do so by way of how often a disease occurs within a given population.⁵ For example, in the European Union, rare diseases are defined to include diseases affecting up to 1 in 2,000 people.⁶ On the other hand, in the United States, the *Rare Diseases Act 2002* (US) defines rare disease as a condition affecting fewer than 200,000 people.⁷ In Australia, the Draft National Strategic Action Plan released for further consultation in July 2019 adopts the European Union definition of rare disease.⁸ Notwithstanding these different definitions, the paradox is that while a given disease may be “rare” in terms of its occurrence, rare diseases as a group are cumulatively common, with approximately 5,000 to 8,000 different rare diseases⁹ affecting around 6–10% of the population. This translates to an estimated 63,000 children in Western Australia alone, and many more on a worldwide scale.¹⁰

Rare diseases are typically complex (involving various systems of the body), chronic and frequently require multidisciplinary care. They are often life-threatening, genetic and incurable. Rare diseases can be painful, disabling and lead to early death.¹¹ About 70% of rare diseases start exclusively in childhood making the particular circumstances of the paediatric patient an added dimension to the treatment and care of patients with rare disease.¹²

BARRIERS TO DIAGNOSIS

These characteristics of rare disease compound the urgency to establish streamlined and efficient methods to arrive at an early and accurate diagnosis. Diagnosis is a complex process which generally involves the medical practitioner taking a history of symptoms, undertaking a physical examination, and then ordering any appropriate medical tests as guided by a working diagnosis. The medical practitioner will then review these test results when they become available so as to arrive at a final diagnosis.¹³ This process is best illustrated as follows:¹⁴

⁴ C Molster et al, “Survey of Healthcare Experiences of Australia Adults Living with Rare Diseases” (2016) 11 *Orphanet Journal of Rare Diseases* 30; G Baynam, S Broley and J Goldblatt, “Initiating an Undiagnosed Diseases Program in the Western Australian Public Health System” (2017) 12 *Orphanet Journal of Rare Diseases* 83, 2.

⁵ Department of Health, Government of Western Australia, *WA Rare Diseases Strategic Framework 2015–2018* (Office of Population Health Genomics, Public Health Division, 2015) 3.

⁶ *Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Disease: Europe’s Challenges*, n 3, para 1.

⁷ *Rare Diseases Act 2002* (US) s 2(a)(1); see also National Center for Advancing and Translational Sciences, <rarediseases.info.nih.gov>.

⁸ Department of Health, Commonwealth of Australia, n 3, 6, making reference to the Ministers, Department of Health, “Australia’s first National Rare Diseases Framework” (Media Release, 16 November 2018) <www.health.gov.au/ministers/the-hon-greg-hunt-mp/media/australias-first-national-rare-diseases-framework>.

⁹ Baynam, Broley and Goldblatt, n 4, 2.

¹⁰ Department of Health, Government of Western Australia, n 5, 3.

¹¹ Department of Health, Commonwealth of Australia, n 3, 6.

¹² S Nguengang Wakap et al, “Estimating Cumulative Point Prevalence of Rare Disease: Analysis of the Orphanet Database” (2020) 28 *European Journal of Human Genetics* 165–173, 168.

¹³ CMPA Good Practice Guide, *The Diagnostic Process* <https://www.cmpa-acpm.ca/serve/docs/ela/goodpracticesguide/pages/manage_risk/The_diagnostic_process/steps_to_diagnosis-e.html>.

¹⁴ National Academies of Sciences, Engineering and Medicine, n 2.

While not all rare disease patients travel the diagnostic odyssey, for those whose condition escapes diagnosis, it means navigating this diagram over and over again with different medical practitioners, often having to “start at the beginning” of the information-gathering process each time. This is at best a tiresome and uncomfortable experience, although the repetition of a traumatic situation by way of talking has been recognised in itself as leading to further trauma.¹⁵

So why is the diagnostic odyssey such a feature of the experience of dealing with rare disease? In this context, four barriers to diagnosis can be identified:

- (1) the nature of the patient/doctor relationship;
- (2) the quantity of information now available;
- (3) increased specialisation; and finally
- (4) the quantity of resources available to the medical system as well as to the patient.

Medical practitioners spend years training in their fields, and as with other professions, a certain language develops in the efficient and accurate communication of information. However, while a competent doctor may take time to explain the medical situation in detail, the lay patient has no means to discern either the suitability nor the evidence behind a particular course of action. This inherent inequality in the patient/doctor relationship means that a patient, presented with the wrong diagnosis or treatment, will not have the information base to either argue the position or seek another opinion. Even if a second opinion is sought, this may not happen until sometime later which in the instance of childhood rare disease, may have irreversible developmental consequences. Put another way, the rare disease patient, unless equipped with the same level of medical skill, cannot evaluate the diagnosis (or reasons for the lack thereof) on an equal footing.¹⁶

Another barrier to diagnosis arises from the explosion in medical information now available, although paradoxically, in the context of rare disease, lack of information is an equally recognised barrier. Masys noted in 2002 in relation to the volume of data in the United States National Library of Medicine’s Medline data, that “a conscientious practitioner who reads two articles each evening will, at the end of a year, be approximately 550 years behind in keeping up with the literature”.¹⁷ The situation, nearly two decades later has compounded. On the other hand, in the context of genetic-based rare disease, while knowledge and high quality of data are recognised as important for accurate diagnosis, “the rarity also represents a barrier to diagnosis and discovery as there are limited data due to the relatively small number of affected individuals”.¹⁸

The nature of information available, taken together with imperatives to meet professional indemnity obligations which are in turn tied to registration of medical professionals¹⁹ have resulted in medical professionals practising in increasingly specialised fields. In securing professional indemnity insurance in order to ensure continued registration, a medical practitioner must detail practice areas, types of patients and other facts that may increase the level of risk in their practice.²⁰ Clause 2.2 of *Good Medical Practice: A Code of Conduct for Doctors in Australia*²¹ provides that not only is maintaining a high level of medical competence critical to good patient care, but a doctor must also recognise and work within the limits of their competence and scope of practice. As at June 2018, the list of medical

¹⁵ B Van der Kolk, *The Body Keeps the Score: Mind, Brain and Body in the Transformation of Trauma* (Penguin Random House, 2015) especially Ch 14.

¹⁶ AVS Kanwar and MM Rahim, “Social Responsibilities of the Global Pharmaceutical Companies: Towards an Ethical Health Care Paradigm” (2019) 26 JLM 75, 754.

¹⁷ Daniel R Masys, “Effects of Current and Future Information Technologies on the Health Care Workforce” (2002) 21(5) *Health Affairs* 33, 35.

¹⁸ MG Hansson et al, “The Risk of Re-identification versus the Need to Identify Individuals in Rare Disease Research” (2016) 24 *European Journal of Human Genetics* 1553, 1554.

¹⁹ AHPRA Fact Sheet: Professional Indemnity Insurance Arrangements 6 January 2016; Changes to the PII arrangements registration standard July 2019.

²⁰ Changes to the PII arrangements registration standard, n 19.

²¹ Medical Board of Australia (March 2014).

specialties recognised by the Medical Board of Australia to support a specialist registration provides for 23 specialties with a subset of 64 “Fields of Specialty Practice”.²² Given these obligations, it is suggested that while the focus narrows to meet professional indemnity insurance obligations, rare disease which often requires consideration of multiple aspects of a patient’s condition results in patients often being referred to multiple specialists resulting in fragmented care which itself may be a barrier to diagnosis.²³

A final barrier arises from the extensive resources required to fund the search for a diagnosis.²⁴ To the extent that a patient must personally fund these expenses, then it stands to reason that access to medical specialists in the quest for a diagnosis will be limited by the resources available to that patient. Indeed, a recent survey considering key issues of rare disease reported that almost half of respondents had out-of-pocket costs of over \$10,000 per year related to their disease.²⁵ As a corollary, medical systems will be limited by the funds available to see patients and then provide advanced diagnostic techniques, medical skills and information that could lead to a diagnosis.²⁶ This issue is compounded by what has been described as a “macroeconomic allocation dilemma” which requires this allocation of resources to be made in circumstances where more common diseases, which affect a greater number of people, could be treated using relatively less resources.²⁷

AVENUES OF REDRESS

So, what avenues of redress are available to a patient frustrated by the diagnostic odyssey but also perhaps, at resources end? In answering this question, pages could be spent analysing a “right to a diagnosis” as an aspect of a “right to health”.²⁸ However, in Australia – where the Medicare system guarantees Australians access to public hospitals and subsidised health care²⁹ – such an argument is not entirely apposite because such analysis misunderstands the plight of those travelling the diagnostic odyssey. This is because the problem is not necessarily with access to a health system, but rather, access to a health system *that can provide an answer*. As acknowledged by the *Report of the United Nations High Commission for Human Rights*:³⁰

Even when access to some form of health care exists, coverage usually mirrors health issues experienced by the general population, with little or no attention paid to the specific needs and rights of persons living with rare diseases. ... [The] paucity of medical and scientific knowledge about rare diseases drives this marginalization, with the result that many people remain undiagnosed and therapies are difficult to develop.

Further to this acknowledgment is the notion that there comes a time for the undiagnosed patient where continuing with the available standard diagnostic approaches of attending one more medical practitioner who requests one more blood test, one more magnetic resonance imaging or one more computed tomography scan becomes just too much. Where the standard model of diagnosis is inherently unsuitable to the particular circumstances of the undiagnosed patient and unacceptably compounds the stress of the diagnostic odyssey.

With this in mind, the analysis is perhaps better framed by considering what actions a person navigating the diagnostic odyssey can take in a situation where the health system’s standard approach is not leading

²² Medical Board of Australia, *List of Specialties, Fields of Specialty Practice and Related Specialist Title* (1 June 2018).

²³ WA Gahl and CJ Tiff, *The NIH Undiagnosed Diseases Program – Lessons Learned* (2011) 305(18) JAMA 1905.

²⁴ Department of Health, Commonwealth of Australia, n 3, 30.

²⁵ The McKell Institute, *Disability and Rare Disease: Towards Person Centred Care for Australians with Rare Disease* (October 2019) 37.

²⁶ Department of Health, Commonwealth of Australia, n 3, 14.

²⁷ Ministry of Health and Family Welfare, Government of India, *National Policy for Treatment of Rare Diseases* (22 March 2017) 4, 15 <mohfw.gov.in/sites/default/files/Rare%20Diseases%20Policy%20FINAL.pdf>.

²⁸ *Universal Declaration of Human Rights* (UDHR) 1948 (resolution 217A) (adopted 10 December 1948) Art 25.

²⁹ Australian Government, *Medicare* <health.gov.au/health-topics/medicare>.

³⁰ *United Nations Economic and Social Council*, 2019 sess, Agenda Item 19(f) (15 May 2019) par 29.

to answers. At this point, the relevance of any rights in negligence will have little practical value. An action in negligence requires a plaintiff to establish “damage” – a difficult requirement when one is not yet diagnosed, still living in hope and the implications of a lack of a diagnosis perhaps not yet fully known. These arguments may arise once the diagnostic odyssey has run its course, but that is likely to be too late for practical and healthful benefit for the undiagnosed because the focus of negligence is compensatory – the damage is done. Instead, the following considers what a patient, frustrated by the absence of a diagnosis, can do “along the way” to advance a diagnosis.

One possibility is to consider rights such as those under s 25 of the *Health and Disability Services (Complaints) Act 1995 (WA) (HDSC Act)* which allow a person to make a complaint alleging that “a provider has acted *unreasonably*” (emphasis added) either in providing (or not providing) a health service for the user, or “in the manner of” providing a health service.³¹

The *HDSC Act* provides a definition of “health service” sufficiently broad to cover someone who is yet to be diagnosed, as the definition of “health service” means any service provided “by way of ... diagnosis or treatment of physical or mental disorder or suspected disorder” and “by way of ... health care”.³² However, the difficulty with founding such a complaint is immediately apparent: a complaint will only be established where the provider has acted “unreasonably” – where a diagnosis is not known, and still being sought, can the provider be said to be acting “unreasonably” by requiring one more diagnostic test or making a referral to another specialty? To this end, s 4(2) provides that the question of whether there has been “unreasonable conduct” is to be determined by the guiding principles set out in s 4(1), which includes in s 4(2)(b) whether the provider has complied with any “professional standards commonly accepted by members of the provider’s profession”. As a general proposition, it would be difficult to argue that there has been “unreasonable” behaviour where the standard model of diagnosis has been followed. Instead, the complainant would have to argue that they have reached the stage in the diagnostic odyssey where following the standard model does not promote “respect for the ... dignity of persons receiving health care” as required by the guiding principle set out in s 4(1)(b). While picking this point in time to lodge a complaint may be problematic (particularly in the light of the Director’s obligation under s 26 to reject a complaint that is “without substance” or which “does not warrant any further action”) the *Health and Disability Services Complaints Office 2018–2019 Annual Report* provides that since 2016, there has been a 72% increase in complaints related to “diagnosis”. Other aspects of treatment which have also been the subject of complaint include “co-ordination of treatment”, “experimental treatment”, “no/inappropriate referral” and “wrong/inappropriate treatment”.³³

When the Director determines that a complaint is acceptable to be dealt with under the *HDSC Act* (this may require consultation with the Australian Health Practitioner Regulation Agency depending on the nature of the complaint³⁴), the powers available to deal with the complaint are set out in the *HDSC Act*. The Director may attempt to settle the complaint by negotiation (a paper-based approach where the Director facilitates the exchange of information), refer the complaint for conciliation (a meeting in person to encourage a settlement) or investigate the complaint.³⁵ If the Director, following an investigation, decides that there has been “unreasonable conduct”, the Director must give written notice to the provider specifying “any action that the Director considers ought to be taken to remedy the matter”.³⁶ A provider then has 45 days to report the remedy undertaken to the Director or else risk a penalty of \$2,500.

³¹ *Health and Disability Services (Complaints) Act 1995 (WA)* s 25(1)(a),(b) and (c).

³² *Health and Disability Services (Complaints) Act 1995 (WA)* s 3(1).

³³ Health and Disability Services Complaints Office (WA), *2018–2019 Annual Report* (10 September 2019) 118.

³⁴ *Health and Disability Services (Complaints) Act 1995 (WA)* ss 31, 32A; see also Health and Disability Services Complaints Office (WA), n 33, 26.

³⁵ *Health and Disability Services (Complaints) Act 1995 (WA)* s 34; Health and Disability Services Complaints Office (WA), n 33, 18.

³⁶ *Health and Disability Services (Complaints) Act 1995 (WA)* s 50(2).

The emotional aspects of the diagnostic odyssey include the stress of “not knowing” which adds to a sense of isolation and even exclusion.³⁷ While the Director’s involvement under the *HDSC Act* could not guarantee a diagnosis, the Director’s involvement could potentially mitigate these emotional aspects of the diagnostic odyssey. Some of the reported redress outcomes achieved using the powers under the *HDSC Act* include the service provider giving an explanation, or even an apology, to the individual making the complaint or a particular service being obtained for an individual.³⁸

Another avenue is to consider whether the diagnostic odyssey is tantamount to indirect discrimination. Generally speaking, indirect discrimination describes the situation where a practice, policy or rule that applies to everybody in the same way actually has a more detrimental or disadvantageous effect on some people than on others.³⁹ In the instance of the diagnostic odyssey, the argument would be that continually applying the standard model of diagnosis has a less favourable impact on those navigating the diagnostic odyssey (especially where the standard model has been applied numerous times without result.) Dawson and Toohey JJ in *Waters v Public Transport Corp*⁴⁰ described indirect discrimination in terms of where “the treatment is on its face neutral but the impact of the treatment on one person when compared with another is less favourable”. In *Griggs v Duke Power Co*⁴¹ it was said that indirect discrimination makes it unlawful to engage in “practices that are fair in form, but discriminatory in operation”.

Notwithstanding these general descriptions of indirect discrimination, whether a complaint on the basis of indirect discrimination will be able to be argued successfully depends upon being able to establish each of the particular elements of the anti-discrimination legislation applying in that jurisdiction.⁴² This is where the difficulty of analysing the diagnostic odyssey as an aspect of “indirect discrimination” comes to the fore.

In Western Australia, s 66A of the *Equal Opportunity Act 1984 (WA) (EO Act)* deals with discrimination on the ground of “impairment”. “Impairment” is defined under s 4(1) of the *EO Act* to mean a “condition” involving “any defect or disturbance in the normal structure or functioning of a person’s” brain or body, and includes an impairment “which is imputed to the person”. While the inclusion of an impairment “imputed” to a person expands the ordinary meaning of this term, there remains a question as to whether a person who does not yet have a diagnosis has a “condition” so as to fall within the definition of “impairment”. Unless an undiagnosed state of itself constitutes a “condition” within the meaning of “impairment” then a complaint for indirect discrimination on the grounds of impairment may not be available to a person dealing with the diagnostic odyssey.⁴³ The term “condition” is not defined in the *EO Act*, but its dictionary meaning according to the *Macquarie Dictionary* includes “state of health”. On the other hand, the *Shorter Oxford Dictionary* defines “condition” to include “a state resulting from a physical or mental illness; sickness or malady”. In *Edoo v Minister for Health*,⁴⁴ the State Administrative Tribunal stated, relying on the decision in *Re Prezzi and Discrimination Commissioner*,⁴⁵ that “the word ‘condition’ appears to denote an underlying cause and not merely symptoms”.⁴⁶ Applying this reasoning, it could be argued that a cluster of symptoms, not yet given a formal diagnosis, is not sufficient to fall within the meaning of impairment under the *EO Act*.

³⁷ Swan UK et al, *International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients* (October 2016) 3.

³⁸ Health and Disability Services Complaints Office (WA), n 33, 27.

³⁹ *Indirect Discrimination* <citizensadvice.org.uk>.

⁴⁰ *Waters v Public Transport Corp* (1991) 173 CLR 349, 392.

⁴¹ *Griggs v Duke Power Co*, 401 US 424, 431 (1971).

⁴² *IW v The City of Perth* (1997) 191 CLR 1.

⁴³ *Bolton v Centrecare Inc* [2011] WASAT 198, [116].

⁴⁴ *Edoo v Minister for Health* (2010) 72 SR (WA) 16; [2010] WASAT 74.

⁴⁵ *Re Prezzi and Discrimination Commissioner* (1996) 39 ALD 729, 742.

⁴⁶ *Edoo v Minister for Health* (2010) 72 SR (WA) 16, [71]; [2010] WASAT 74.

Even assuming impairment extends to someone yet to be diagnosed, a complainant would have to establish each of the criteria in s 66A(3) in order to argue successfully that indirect discrimination has occurred.⁴⁷ Section 66A(3) provides that indirect discrimination can occur on the ground of impairment:

- if the discriminator requires the aggrieved person to comply with a requirement or condition –
- (a) with which a substantially higher proportion of persons who do not have the same impairment as the aggrieved person comply or are able to comply; and
 - (b) which is not reasonable having regard to the circumstances of the case; and
 - (c) with which the aggrieved person does not or is not able to comply.

In the context of s 66A(3)(b), a person navigating the diagnostic odyssey would have to establish that being sent for one more test, or being forced to source one more medical practitioner “is not reasonable” and that the provision of the health service therefore has an indirectly discriminatory effect. Sackville J stated in *Commonwealth Bank of Australia v Human Rights & Equal Opportunity Commission*,⁴⁸ that the “fact that a distinction has a ‘logical and understandable basis’ will not always be sufficient to ensure that a condition or requirement is objectively reasonable”. However, at what point of the diagnostic odyssey could an aggrieved person establish that the health service has an indirectly discriminatory effect on the basis that while at the beginning of the diagnostic odyssey, the standard model of diagnosis had a “logical and understandable basis” after so many times through that model, the effect on a person of seeking a diagnosis is not reasonable? It has been acknowledged that discriminatory conduct to found a complaint is not easily established “when one descends from generalised expectations or sentiments to the technical language in which the anti-discrimination legislation is expressed”.⁴⁹ In the context of the diagnostic odyssey where a “condition” is yet to be determined and the standard model of diagnosis has an inherently reasonable, medically accepted basis, the legislative elements to establish indirect discrimination may be insurmountable.

THE PROCESS HAS STARTED: THE WA RARE DISEASES STRATEGIC FRAMEWORK 2015–2018

Against these hurdles, Western Australia has led the charge in addressing the particularities of the diagnostic odyssey, in the context of rare disease, by being the first Australian State to establish a rare disease action plan.⁵⁰ The *WA Rare Diseases Strategic Framework 2015–2018* sets out a “framework for the coordination of WA Health initiatives for rare diseases”.⁵¹ Objective 7 provides for building on existing WA Health Services for the screening and diagnosis of rare disease. Objective 8 then encourages the use of evidence-based, best practice guidelines to deliver health care for rare disease. Each objective is grounded in an acknowledgment that “[an] accurate and early diagnosis provides those living with a rare disease and their families and carers an understanding of future healthcare requirements”.⁵² In response to these objectives, Initiative 2 of Objective 7 proposes to:

Scope the need for and feasibility of mechanisms (eg a [Rare Disease] clinic) to facilitate referral pathways from primary/community care for diagnosis of [Rare Disease].

Similarly, Initiative 2 of Objective 8 proposes to:

Scope the need for a WA model of care for [Rare Disease].

In advancing this framework, the UDP was established in March 2016 and “incorporates a team of doctors from a broad range of specialties working together in partnership with researchers”.⁵³ The UDP

⁴⁷ *Edoo v Minister for Health* (2010) 72 SR (WA) 16, [61]; [2010] WASAT 74 and more recently, *D’Alto v Curtin University* [2019] WASAT 61, [21].

⁴⁸ *Commonwealth Bank of Australia v Human Rights & Equal Opportunity Commission* (1997) 80 FCR 78.

⁴⁹ *IW v The City of Perth* (1997) 191 CLR 1, 52 (Kirby J).

⁵⁰ Molster et al, n 4; Department of Health, Commonwealth of Australia, n 3, 7.

⁵¹ Department of Health, Government of Western Australia, n 5, 1

⁵² Department of Health, Government of Western Australia, n 5, 16.

⁵³ Project, <projectyudp.com>.

seeks to address a particular aspect of the diagnostic odyssey that arises from the way the medical system works: that while a patient is referred to each specialist individually, the likelihood of a diagnosis may be improved by the collaboration of these experts.

The UDP is made up of the Program Director, Program Co-ordinator, Genetic Counsellor and Expert Panel.⁵⁴ The Expert Panel is made up of specialists from a deliberately diverse range of medical specialties at differing career stages. Members meet each month to discuss possible diagnoses and further assessment for referred patients following a review of the patient history. Patients come from within the Western Australian (WA) public health system, and at this stage, are limited to paediatric and transition age patients (defined as 16–25 years of age). Patient self-referral is not accepted. Of note, it has been observed that the open collaboration of clinicians, coming from specialties as diverse as genetics, cardiology and metabolic medicine, allows for consideration of “hunches or intuitions, which can spark solutions”.⁵⁵

The earliest example of outcomes “sparked” by the collaboration of experts came with “Lily”, the first patient seen within the UDP. Lily’s condition presented at birth, and affected her heart, bones, hair, liver and growth. By seven years of age she had had more than 50 inpatient hospital admissions, 13 general anaesthetics for invasive diagnostic procedures, and more than 200 hospital outpatient admissions. She had an incredibly extensive list of medical investigations and many individual doctors (involving more than 10 medical specialties) had tried to achieve a diagnosis. Part of Lily’s experience with the UDP included a meeting with the Expert Panel (covering eight medical specialties). This Expert Panel identified a presumptive diagnosis (trichohepatoenteric syndrome) in 20 minutes, and a definitive diagnosis was achieved in 48 hours. Within one week, Lily and her family were connected to 37 other families living with this condition around the world.⁵⁶

STRENGTHENING THE WA APPROACH

The processes of the UDP lead to questions as to whether they could be enhanced by a legislative framework. This is in no way to downplay the medical realities of rare disease, the diagnostic odyssey and consequent health implications, or to suggest that legislation could be drafted to easily deal with these matters. Moreover, this is not to undermine the advances already made by the UDP or its method of operations. However, a framework which brings together experts in their fields for the purposes of arriving at a diagnosis could be established under legislation in a way that promotes access by patients themselves, the identification of appropriate expertise, consultation and knowledge building.

Legislation could set out criteria which provide equity and clarity for those who wish to apply for the assistance of the diagnostic panel. In particular, rather than relying upon a medical practitioner’s referral as is currently the situation with the UDP (and which is also currently limited in any event to patients within the public health system), legislation could provide for patients to refer themselves. A medical practitioner’s referral could be specified as a relevant consideration in determining whether access is to be granted to the panel, but not a mandatory requirement. This would allow a patient some control to escalate their plight with the aim of bringing their condition to the attention of suitable experts rather than having to rely upon the co-operation of their medical team.

The appointment of appropriate expertise, including a pool of suitable practitioners, and access to medical resources could also be set out in legislation. Furthermore, legislation could also ensure that compensation for consultation between medical practitioners is addressed to the extent that it is not covered by the Medicare system. In this way, the “core power” of the UDP could be enhanced as it would allow such a panel to focus “assessments and expertise in one place at one time and centred on one

⁵⁴ Baynam, Broley and Goldblatt, n 4, 4.

⁵⁵ G Hay and F Klonek, *Solving the Unsolvable: How De-centralized Knowledge Sharing Helps Diagnose Children with Rare Diseases* <sketchingworkdesign.com>.

⁵⁶ Baynam, Broley and Goldblatt, n 4, 2.

patient at a time, [thereby harnessing] the richness of face-to-face discussion and real-time and in-person clinical assessments”.⁵⁷

This openness and the opportunity for consultation in arriving at a diagnosis is another objective that could be enshrined as a characteristic of the legislative framework. So far, the discussion has focused on consultation between medical practitioners, but in order to address the inherent inequality between patient and doctor, the right to consultation could also be as between patient and panel, with perhaps even a legislative right to access assistance by way of, for example, counselling to provide support and explanation. As the UDP currently operates, patients accepted into the UDP speak first with the Program Co-ordinator (a genetic counsellor) to learn more about the program and after going through the medical assessment, speak with the Program Director to discuss the outcomes.⁵⁸

Finally, legislation would be able to set out a requirement that outcomes be recorded, and be able to be accessed, so that information can be applied to future similar situations thereby allowing a body of knowledge to be developed. This output will need to be considered carefully in the light of privacy and ethical obligations, as well as concerns such as fear of discrimination and reduced access to opportunities.⁵⁹ Moreover, in the context of rare disease, the rarity of each disease may allow for either “identity disclosure” or “attribute disclosure” thereby resulting in breaches of privacy even when it has been de-identified by removing details such as names, addresses and date of birth.⁶⁰ Another possibility would be for outcomes from the panel to be encompassed in specific health system coding for undiagnosed rare diseases. This would provide the means to flag and monitor patients systematically, as well as evaluate the system-wide effect of the diagnostic odyssey and interventions to reduce that odyssey. Indeed, the collection and effective use of rare disease data is recognised as critical to the successful implementation of strategies to deal with rare disease.⁶¹

Knowledge could also be built for the direct benefit of the patient once the panel process is concluded, if the patient is given a document akin to “reasons for decision” which summarises and explains the approaches, tests and matters in a medically acceptable format. Indeed, with so many test results gathered along the diagnostic odyssey,⁶² patients often lack the knowledge and skills to order these in an acceptable format, or analyse which aspects of the patient’s condition have been considered (and discounted). This possible output from the panel would be helpful even if a diagnosis is not made as the patient could take this away to introduce their condition to the next specialist (assuming the patient still has the stamina to go on) thereby avoiding the trauma of having to repeat the experience. Such “reasons for decision” are usually required to be produced by administrative law review panels established under legislation. In this context, providing reasons for decisions has been described as recognising “the dignity of members of society and treats those subject to decisions with respect”.⁶³ Given the trauma of having to repeatedly explain the situation “from the beginning” when navigating the diagnostic odyssey, a more humane reason to provide “reasons for decision” is unlikely to be found.

THIS IS NOT NOVEL

Applying a system set up by legislation to lead to, or support, a medical diagnosis is not novel. In this regard, two examples come to mind: medical panels in Victoria and the *Mental Health Act 2014* (WA) (*MH Act*). Each will be considered in turn.

⁵⁷ Baynam, Broley and Goldblatt, n 4, 2.

⁵⁸ Baynam, Broley and Goldblatt, n 4, 5.

⁵⁹ McCormack et al, “‘You Should at least Ask’. The Expectations, Hopes and Fears of Rare Disease Patients on Large-scale Data and Biomaterial Sharing for Genomics Research” (2016) 24 *European Journal of Human Genetics* 1403, 1406.

⁶⁰ Centre for Epidemiology and Evidence, *HealthStats NSW: Privacy Issues and the Reporting of Small Numbers* (NSW Ministry of Health, 2015) paras 4.1, 4.2.

⁶¹ Department of Health, Commonwealth of Australia, n 3, 21, 44.

⁶² D Taruscio, “Undiagnosed Diseases Network International (UDNI): White Paper for Global Actions to Meet Patient Needs” (2015) 116 *Molecular Genetics and Metabolism* 223, 223.

⁶³ R Douglas et al, *Douglas and Jones Administrative Law* (The Federation Press, Sydney, 2018) 47.

Medical panels in Victoria are established under workers compensation legislation and the *Wrongs Act 1958* (Vic). The mission of the Office of the Convenor of Medical Panels is to “support the fair and timely resolution of disputes, through the provision of final and binding opinions and reasons in relation to medical questions referred”.⁶⁴ Medical Panels are made up of specialist and general medical practitioners⁶⁵ and, while there is no limit to the number of practitioners who may be assigned to a Medical Panel, the nature of the expertise assigned will depend on the particular circumstances of the referral.⁶⁶ Medical questions able to be referred to Medical Panels include determining aspects of a WorkCover-related injury or medical condition, or the extent of impairment.⁶⁷ The Medical Panel provides an opinion which is legally conclusive and binding on any court, body or person. As such, opinions regarding these matters affect rights to compensation, medical services and other entitlements. In reaching a determination, a Medical Panel uses its own knowledge and expertise⁶⁸ and may undertake its own medical examination of the claimant.⁶⁹

Examinations are also authorised under the *MH Act* and a diagnosis made or confirmed in accordance with the procedures under the *MH Act* could lead to an inpatient treatment order which is one of the most prominent examples of the intersection between legislation and the diagnosis of a medical condition. Inpatient treatment orders authorise a person’s detention at a hospital with a view to the person being able to be treated without informed consent for the mental health condition.⁷⁰ Under s 26(1), a medical practitioner may refer a person for an examination by a psychiatrist if the practitioner “reasonably suspects” that the person requires such an order because, among other things, the person “has a mental illness for which the person is in need of treatment”.⁷¹ The *MH Act* nevertheless recognises the implications of this course of action so that s 386 requires that the Mental Health Tribunal review the order “as soon as practicable after an involuntary treatment order is made”. Unlike Medical Panels in Victoria, the Mental Health Tribunal is made up a medical professional (a psychiatrist) as well as a lawyer and a community member.⁷²

Each of these panels provides an example of the intersection of a medical diagnosis and legislation which has potentially serious consequences for the individual concerned such as affecting rights to compensation, or in the instance of the *MH Act*, personal liberty and consent to treatment. The diagnostic panel described for the purpose of supporting the diagnostic odyssey does not have a specific objective of establishing rights and obligations. However, a diagnosis of itself may have the consequential benefits of providing the basis for the patient to access medical services, obtain welfare assistance or even claim employment entitlements that would otherwise require a diagnosis in order to be available.

LEGISLATION IS ALREADY CREEPING IN

Addressing the diagnosis of rare disease through legislative intervention has already commenced in some countries. The following examples involve legislation which only authorises or requires the bringing together of experts for the purposes of diagnosing rare disease. However, the rules and obligations setting out how these experts are to work together is contained outside the terms of the legislation.

The Office of Rare Disease was established at the National Institutes of Health (NIH) in Washington DC pursuant to the *Rare Diseases Act of 2002* (US). The United States Act aims to “increase the national investment in the development of diagnostics and treatments for patients with rare diseases

⁶⁴ Medical Panels Victoria, *The Office of the Convenor of Medical Panels End of Year Results 2015/2016* (September 2016) 3.

⁶⁵ Medical Panels <medicalpanels.vic.gov.au>.

⁶⁶ C Newlands, “Judicial Review of Medical Panels Decisions” (2017) 24 JLM 538, 539, 540.

⁶⁷ Medical Panels Victoria, n 64, 3.

⁶⁸ Newlands, n 66, 539.

⁶⁹ Convenor of Medical Panels, Convenor’s Direction, *Medical Panel Examinations*.

⁷⁰ *Mental Health Act 2014* (WA) s 22.

⁷¹ *Mental Health Act 2014* (WA) ss 25(1)(a), 26.

⁷² *Mental Health Act 2014* (WA) s 383.

and disorders”.⁷³ In 2008 the Office of Rare Disease established a UDP⁷⁴ where accepted patients are evaluated at the NIH following their medical records being reviewed by up to five medical professionals representing 25 different specialties.⁷⁵ Of note, one of the broader benefits of this program has been the discovery of “insights into human physiology, biochemistry and genetics” which in turn provide valuable information for more common diseases.⁷⁶

Rare disease, including references to diagnosis, has also been dealt with by legislation in Taiwan and the Philippines. In Taiwan, the *Rare Disease and Orphan Drug Act 2015* (Taiwan) includes the early diagnosis of rare disease as one of its objectives,⁷⁷ and ensures that funding is available for diagnosis, treatment and drug costs for rare diseases that are not covered by the national insurance scheme.⁷⁸ Similarly, the *Rare Diseases Act of the Philippines 2015* seeks to “improve the access of patients diagnosed to have a rare disease or patients highly suspected of having a rare disease to comprehensive medical care”⁷⁹ Under this Act, the Department of Health is charged with the responsibility of being the lead agency for implementing the Act, including being required to “organize a pool of medical specialists who will be responsible in the diagnosis and management of persons afflicted with rare disease and their families”.⁸⁰ This follows the requirement under s 7 for the Department, with assistance from NIH (USA) to “develop a system to train a sufficient number of medical specialists to diagnose and manage persons with rare disease”.

In Australia the *Draft National Strategic Action Plan for Rare Diseases* does not advocate a legislative regime for Australia but it does seek that “the Australian Government to establish a dedicated Rare Disease Unit within a central agency”.⁸¹ This call is made in recognition that rare disease policy is complex and that a national approach is required to ensure that the necessary health infrastructure exists to support the early diagnosis of rare disease.⁸²

CONCLUSION

Rare disease is “rare and everywhere”⁸³ and its correlation with the diagnostic odyssey significant. Given the medical, emotional and financial implications of the diagnostic odyssey there must be room to consider a system of diagnosis informed by characteristics of existing legislation based on medical-type panels. This panel could further enhance the existing frameworks of the UDP, as well as mandate two particular outcomes for patients dealing with the diagnostic odyssey: a mode of empowering patients to bring together experts, rather than leaving patients merely to hope that their doctors will confer, as well as providing patients with a document akin to “reasons for decision” which could summarise and explain the approaches, tests and matters considered to date in a way that sets out the information in a conventional medical format. A diagnostic panel will not guarantee a diagnosis, and indeed, a diagnosis may not be able to be treated – but for the person concerned, a name and direction provided by a diagnosis can provide life-altering benefits by directing a patient to comfort and support, and a sense of belonging.⁸⁴

⁷³ *Rare Diseases Act of 2002* (US) s 2(9)(b).

⁷⁴ Kuehn, n 1.

⁷⁵ Gahl and Tiftt, n 23, 1904.

⁷⁶ WA Gahl, “The Battlefield of Rare Diseases: Where Uncommon Insights Are Common” (2012) 4 *Sci Transl Med* 154ed7.

⁷⁷ *Rare Disease and Orphan Drug Act 2015* (Taiwan) Art 1.

⁷⁸ *Rare Disease and Orphan Drug Act 2015* (Taiwan) Art 33; see also Orphanews Europe, *How Specific Legislation for Rare Diseases and Orphan Drugs Is Changing Life for Some Patients in Taiwan* <<https://www.orpha.net/actor/EuropaNews/2009/doc/Taiwan.pdf>>; RD-ACTION, *Overview Report on the State of the Art of Rare Disease Activities in Europe 2018* <<http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf>>.

⁷⁹ *Rare Diseases Act of the Philippines 2015* (Philippines) s 3(a).

⁸⁰ *Rare Diseases Act of the Philippines 2015* (Philippines) s 17(e).

⁸¹ Department of Health, Commonwealth of Australia, n 3, 18.

⁸² Department of Health, Commonwealth of Australia, n 3, 18.

⁸³ Project, n 53.

⁸⁴ Kuehn, n 1.